Supplementary Table 1: Gene-disease association reference list

Gene	Curated disease	Evidence for gene-disease association	Typical inheritance	Penetrance	Age of onset <18 Yrs	BabySeq Category	Meets NGSR citeria?	Reason for BabySeq category	Pediatric disease gene in Bell 2011	Key references used in curation (PubMed ID)	BabySeq IBA panel	Reviewed
AAAS	Achalasia-addisonianism-alacrimia syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12730363, 11159947	HYPOTO	Sept 2016 - Oct 2016
AARS	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22009580, 22206013	IEM, HYPOTO	Sept 2016 - Oct 2016
AARS2	Leukoencephalopathy, and ovarian failure in females	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24808023	HYPOTO	Sept 2016 - Oct 2016
ABAT	GABA-transaminase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20052547, 9746906,	SEIZ	Sept 2016 - Oct 2016
ABCA12	Ichthyosis, congenital, autosomal recessive	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19434086, 10712205	DERM	Sept 2016 - Oct 2016
ABCA3	Surfactant metabolism dysfunction, pulmonary, 3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16641205, 16728712	PULM	Sept 2016 - Oct 2016
ABCA4	Stargardt disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9054934, 10958763,	9973280, 14517951, 115279	Sept 2016 - Oct 2016
ABCB11	Cholestasis, progressive familial intrahepatic 2	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9806540, 15300568,	BIL	Sept 2016 - Oct 2016
ABCB4	Cholestasis, progressive familial intrahepatic 3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9419367, 17726488,	BIL	Sept 2016 - Oct 2016
ABCB7	Sideroblastic anaemia and ataxia	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10196363, 11050011	AN_TH, IEM, HYPOTO	Sept 2016 - Oct 2016
ABCC2	Dubin-Johnson syndrome	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		8235715, 9425227	BIL, SEIZ	Sept 2016 - Oct 2016
ABCC6	Pseudoxanthoma elasticum	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10835643 10811882	DERM	Sept 2016 - Oct 2016
ABCC8	Hyperinsulinemic hypoglycemia, familial	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14715863, 23275527	GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ABCC9	Atrial fibrillation, familial	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		17245405, 24439875	CM, COND, DERM	Sept 2016 - Oct 2016
ABCC9	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	No	B	Yes	Moderate evidence, Actionable in childhood		15034580, 24503780	CM, COND, DERM	Sept 2016 - Oct 2016
ABCC9	Hypertrichotic osteochondrodysplasia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22610116, 24352916	CM, COND, DERM	Sept 2016 - Oct 2016
ABCD1	Adrenoleukodystrophy	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7904210, 11992258 ,	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22922874, 23141461	AN_TH, SEIZ	Sept 2016 - Oct 2016
ABCG5	Sitosterolemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11378826, 11099417	BIL, AN_TH	Sept 2016 - Oct 2016
ACAD8	Isobutyryl-CoA dehydrogenase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17304052, 12359132	IEM	Sept 2016 - Oct 2016
ACAD9	ACAD9 deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21057504, 22499348	IEM, HYPOTO	Sept 2016 - Oct 2016
ACADL	Sudden infant death	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	24591516		Sept 2016 - Oct 2016
ACADM	Medium chain acyl CoA dehydrogenase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11349232, 22542437	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ACADS	Acyl-CoA dehydrogenase, short-chain, deficiency of	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	16926354, 9499414,	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency	Strong	AR	LOW (A)	Yes	C	No	Low penetrance	Y	20547083, 15615815	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ACADVL	VLCAD deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7479827, 10077518,	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ACAT1	Alpha-methylacetoacetic aciduria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17236799, 15877211	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
ACBD5	Thrombocytopaenia	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20626622	AN_TH	Sept 2016 - Oct 2016
ACE	Renal tubular dysgenesis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2359105, 22095942,	REN	Sept 2016 - Oct 2016
ACO2	Cerebellar-retinal degeneration, infantile	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		25351951, 22405087	IEM, HYPOTO	Sept 2016 - Oct 2016
ACOX1	Peroxisomal acyl-CoA oxidase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8040306, 18536048,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ACSF3	Combined malonic and methylmalonic aciduria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21841779, 21785126	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
ACTA1	Nemaline myopathy	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10508519, 12601110	HYPOTO	Sept 2016 - Oct 2016
ACTA1	Congenital myopathy with fiber type disproportion	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17387733, 15468086	HYPOTO	Sept 2016 - Oct 2016
ACTA2	Aortic aneurysm, familial thoracic	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		17994018, 19409525,	21248741, 21212136, 2575	Sept 2016 - Oct 2016
ACTB	Baraitser-Winter syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23756437, 23649928	HL	Sept 2016 - Oct 2016
ACTB	Neutrophil dysfunction and recurrent infection	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		10411937	HL	Sept 2016 - Oct 2016
ACTC1	Atrial septal defect	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17611253, 17947298	CHD, CM, COND	Sept 2016 - Oct 2016
ACTC1	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		9563954, 22464770,	CHD, CM, COND	Sept 2016 - Oct 2016
ACTC1	Cardiomyopathy, familial hypertrophic	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		17611253	CHD, CM, COND	Sept 2016 - Oct 2016
ACTC1	Left ventricular noncompaction	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17611253, 2520164	CHD, CM, COND	Sept 2016 - Oct 2016
ACTG1	Deafness, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		13680526, 19477959	HL	Sept 2016 - Oct 2016
ACTG1	Baraitser-Winter syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22366783	HL	Sept 2016 - Oct 2016
ACTG2	Megacystis-microcolon-intestinal hypoperistalsis syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24676022, 24337657	BOW	Sept 2016 - Oct 2016
ACTN1	Macrothrombocytopenia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23434115, 25949529	AN_TH	Sept 2016 - Oct 2016
ACTN2	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		14567970	CM	Sept 2016 - Oct 2016
ACTN2	Cardiomyopathy, familial hypertrophic	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		17097056, 20022194	CM	Sept 2016 - Oct 2016
ACTN4	Glomerulosclerosis, focal segmental, 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10700177, 12444222	REN	Sept 2016 - Oct 2016
ACVR1	Fibrodysplasia ossificans progressiva	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19085907, 18203193	CHD	Sept 2016 - Oct 2016
ACVR2B	Left-right axis malformation	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9916847, 21864452	CHD	Sept 2016 - Oct 2016
ACVRL1	Telangiectasia, hereditary hemorrhagic, type 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18312453, 17786384	PULM, DERM	Sept 2016 - Oct 2016
ADA	Severe combined immunodeficiency due to ADA deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9758612, 11807006,	3946419, 6863546	Sept 2016 - Oct 2016
ADAM17	Neonatal inflammatory skin and bowel disease	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22010916, 25804906	BOW	Sept 2016 - Oct 2016
ADAMTS13	Thrombotic thrombocytopenic purpura, familial	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14563640, 12576319	THROM, AN_TH, REN	Sept 2016 - Oct 2016
ADAMTS2	Ehlers-Danlos syndrome VIIc	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	10417273, 15373769,	23495203	Sept 2016 - Oct 2016
ADAMTSL2	Geleophysic dysplasia 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2090119, 18677313,	IEM, SK, DERM	Sept 2016 - Oct 2016
ADAR	Aicardi-Goutieres syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23001123, 25604658	THYR, AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ADAR	Dyschromatosis symmetrica hereditaria	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15955093, 24950769	THYR, AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ADK	Hypermethioninemia due to adenosine kinase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21963049, 2664297	BIL	Sept 2016 - Oct 2016
AGA	Aspartylglucosaminuria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11174635, 1904874,	IEM, SEIZ	Sept 2016 - Oct 2016
AGL	Glycogen storage disease IIIa	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11977176, 10982190	GLY, HYPOTO	Sept 2016 - Oct 2016
AGPS	Rhizomelic chondrodysplasia punctata, type 3	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	21990100, 24849933		Sept 2016 - Oct 2016
AGRN	Myasthenia, limb-girdle, familial	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21990100, 24951643	HYPOTO	Sept 2016 - Oct 2016
AGT	Renal tubular dysgenesis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	2359105, 22095942,	REN	Sept 2016 - Oct 2016
AGTR1	Renal tubular dysgenesis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	2359105, 22095942,	REN	Sept 2016 - Oct 2016
AGXT	Hyperoxaluria, primary, type 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1703535, 10453743,	THYR, REN	Sept 2016 - Oct 2016
AHI1	Joubert syndrome-3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16453322, 15322546	REN, SEIZ	Sept 2016 - Oct 2016
AHSP	Thalassaemia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20371604	AN_TH	Sept 2016 - Oct 2016
AIFM1	Cowchock syndrome	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23217327, 20362274	HL, IEM, HYPOTO	Sept 2016 - Oct 2016
AIP	Pituitary adenoma	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate evidence, Actionable in childhood		17244780, 23371967,	26186299, 16728643, 1789	Sept 2016 - Oct 2016
AIRE	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10677297, 9398839,	DERM	Sept 2016 - Oct 2016
AK1	Hemolytic anemia due to adenylate kinase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12649162, 10233365	AN_TH	Sept 2016 - Oct 2016
AKAP9	Long QT syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18093912, 23174487	COND	Sept 2016 - Oct 2016
AKR1D1	Bile acid synthesis defect, congenital, 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3198770, 8301429, 1	BIL	Sept 2016 - Oct 2016
AKT2	Severe insulin resistance and diabetes mellitus	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		15166380, 21979934	GLY, DERM	Sept 2016 - Oct 2016
AKT3	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22729224, 23745724	SEIZ, DERM	Sept 2016 - Oct 2016
ALAS2	Anemia, sideroblastic, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7592563, 24323989,	AN_TH, IEM, HYPOTO	Sept 2016 - Oct 2016
ALB	Analbuminemia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9266687, 20025859,	THYR	Sept 2016 - Oct 2016
ALDH18A1	Cutis laxa, autosomal recessive, type IIIA	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21739576, 22411858	DERM	Sept 2016 - Oct 2016
ALDH1A2	Tetralogy of Fallot	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19886994	CHD	Sept 2016 - Oct 2016
ALDH3A2	Sjogren-Larsson syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7485163, 2666627, 1	SEIZ, HYPOTO, DERM	Sept 2016 - Oct 2016
ALDH4A1	Hyperprolinemia, type II	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		9700195, 2624476	SEIZ	Sept 2016 - Oct 2016
ALDH5A1	Succinic semialdehyde dehydrogenase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14635103, 9266358,	SEIZ	Sept 2016 - Oct 2016
ALDOA	Aldolase A deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		14615364, 2825199,	HYPOTO	Sept 2016 - Oct 2016
ALDOB	Fructose intolerance	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8071980, 738900, 33	BIL	Sept 2016 - Oct 2016
ALG1	Congenital disorder of glycosylation, type Ik	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14973782, 20679665	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG11	Congenital disorder of glycosylation type 1P	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20080937, 22213132	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG12	Congenital disorder of glycosylation, type Ig	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15639192, 12093361	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG2	Congenital disorder of glycosylation, type Ii	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12684507	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG3	Congenital disorder of glycosylation, type Id	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10581255, 16053906	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG6	Congenital disorder of glycosylation, type Ic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10359825, 9710431,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG8	Congenital disorder of glycosylation, type Ih	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1523502, 19648040,	REN, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALG9	Congenital disorder of glycosylation, type Il	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15148656, 15945070	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ALMS1	Alstrom syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17594715, 11941370	HL, REN, SEIZ, DERM	Sept 2016 - Oct 2016
ALOX12B	Ichthyosis, congenital, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11773004, 11398099	DERM	Sept 2016 - Oct 2016
ALOXE3	Ichthyosis, congenital, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19434086, 10712205	DERM	Sept 2016 - Oct 2016
ALPL	Hypophosphatasia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	3174660, 8954059, 9	SK, SEIZ	Sept 2016 - Oct 2016
ALS2	Amyotrophic lateral sclerosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2328408, 12601111,	11586297, 17698795, 2001	Sept 2016 - Oct 2016
ALX4	Parietal foramina 2	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16319823, 11137991	SEIZ	Sept 2016 - Oct 2016
AMACR	Bile acid synthesis defect, congenital, 4	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	10594127, 12512044	BIL, IEM, HYPOTO	Sept 2016 - Oct 2016
AMACR	Alpha-methylacyl-CoA racemase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10655068, 21576695	BIL, IEM, HYPOTO	Sept 2016 - Oct 2016
AMELX	Amelogenesis imperfecta	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1483698, 7599636, 1	DERM	Sept 2016 - Oct 2016
AMN	Megaloblastic anemia-1, Norwegian type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12590260, 22929189	AN_TH	Sept 2016 - Oct 2016
AMPD1	Adenosine monophosphate deaminase deficiency	Moderate	AR	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	11331279, 1631143,	HYPOTO	Sept 2016 - Oct 2016
AMT	Hyperglycinaemia, non-ketotic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	6336599, 3297708, 8	SEIZ	Sept 2016 - Oct 2016
ANK1	Spherocytosis	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7883994, 2961992, 1	BIL	Sept 2016 - Oct 2016
ANK2	Long QT syndrome	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		16253912, 17242276	COND	Sept 2016 - Oct 2016
ANKH	Craniometaphyseal dysplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19449425, 20358596	HL, SK	Sept 2016 - Oct 2016
ANKRD1	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	No	B	Yes	Moderate evidence, Actionable in childhood		19525294, 19608030	CHD, CM	Sept 2016 - Oct 2016
ANKRD1	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19608031	CHD, CM	Sept 2016 - Oct 2016
ANKRD26	Thrombocytopenia 2	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21211618, 21467542	AN_TH	Sept 2016 - Oct 2016
ANO10	Spinocerebellar ataxia, autosomal recessive 10	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21092923, 25089919,	25182700	Sept 2016 - Oct 2016
ANO5	Muscular dystrophy, limb-girdle, type 2L	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20096397, 23606453	SK, HYPOTO	Sept 2016 - Oct 2016
ANO5	Gnathodiaphyseal dysplasia	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15124103, 23843187	SK, HYPOTO	Sept 2016 - Oct 2016
ANTXR2	Hyaline fibromatosis syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12214284, 14508707	IEM, DERM	Sept 2016 - Oct 2016
AP1S3	Pustular psoriasis	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24791904		Sept 2016 - Oct 2016

AP3B1	Hermansky-Pudlak syndrome 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23403622, 16551969	PULM, DERM	Sept 2016 - Oct 2016
AP4M1	Spastic paraplegia 50, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21937992, 24700674	HYPOTO	Sept 2016 - Oct 2016
APC	Adenomatous polyposis coli	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17938238, 1651174,	1316610, 1324223, 8395941,	Sept 2016 - Oct 2016
APC	Adenomatous polyposis coli, attenuated	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20105204, 8064829,	9585611, 20434453, 2107819	Sept 2016 - Oct 2016
APOB	Apolipoprotein B deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15805152, 8468533,	17570373, 17158591, 24842	Sept 2016 - Oct 2016
APOE	Sea-blue histiocyte disease	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		11095479, 16094309	AN_TH	Sept 2016 - Oct 2016
APP	Alzheimer disease 1, familial	Definitive	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		1671712, 2111584,	1415269, 10867787, 1044157	Sept 2016 - Oct 2016
APRT	Adenine phosphoribosyltransferase deficiency	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11532677, 20150536	REN	Sept 2016 - Oct 2016
APTX	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11176953, 11022012	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
AR	Androgen insensitivity	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12838569, 10995865,	8723113, 10690872, 11587	Sept 2016 - Oct 2016
AR	Spinal and bulbar muscular atrophy of Kennedy	Definitive	XLR	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		2062380, 25449081,	HYPOTO	Sept 2016 - Oct 2016
ARFGEF2	Periventricular heterotopia with microcephaly	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14647276, 19384555	SEIZ	Sept 2016 - Oct 2016
ARG1	Arginase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7649538, 6422160, 2	IEM, SEIZ	Sept 2016 - Oct 2016
ARHGAP31	Syndromic cutis aplasia & limb anomalies	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21565291	SK	Sept 2016 - Oct 2016
ARHGEF9	Hyperekplexia and epilepsy	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21633362, 17893116	SEIZ	Sept 2016 - Oct 2016
ARID1A	Coffin-Siris syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22426308, 23929686	SEIZ	Sept 2016 - Oct 2016
ARID1B	Coffin-Siris syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22426308, 23929686	SEIZ	Sept 2016 - Oct 2016
ARL13B	Joubert syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18674751, 24168557	REN, SEIZ	Sept 2016 - Oct 2016
ARMC4	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23849778, 24203976	PULM	Sept 2016 - Oct 2016
ARSA	Metachromatic leukodystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2574462, 2906225, 1	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1550123, 15324318,	IEM, SEIZ	Sept 2016 - Oct 2016
ARSE	Chondrodysplasia punctata, X-linked recessive	Strong	XLR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	12567415, 23470839	SK	Sept 2016 - Oct 2016
ARX	Lissencephaly, X-linked 2	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12379852, 18462864	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ASCL1	Congenital central hypoventilation	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		14532329, 18173746	PULM	Sept 2016 - Oct 2016
ASL	Argininosuccinic aciduria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12384776, 1594374,	IEM, SEIZ	Sept 2016 - Oct 2016
ASNS	Microcephaly, intellectual disability, cerebral atrophy & intractable seizures	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24139043, 25227173	SEIZ	Sept 2016 - Oct 2016
ASPA	Canavan disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8252036, 16437572,	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ASS1	Citrullinemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11941481, 2358466,	IEM, SEIZ	Sept 2016 - Oct 2016
ATIC	AICA-Ribosiduria	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	15114530		Sept 2016 - Oct 2016
ATM	Ataxia-telangiectasia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9915942, 12497634,	DERM	Sept 2016 - Oct 2016
ATN1	Dentatorubral-pallidoluysian atrophy 1	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		7824105, 8929958, 2	SEIZ	Sept 2016 - Oct 2016
ATP1A2	Hemiplegic migraine	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18513263, 18644608,	18056581, 24921013, 2083	Sept 2016 - Oct 2016
ATP1A3	Rapid-onset dystonia-parkinsonism	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		24523486, 17282997	SEIZ	Sept 2016 - Oct 2016
ATP2A1	Brody myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10914677, 8841193,	HYPOTO	Sept 2016 - Oct 2016
ATP6AP2	X-linked recessive intellectual deficit - epilepsy	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15746149, 1976568	SEIZ	Sept 2016 - Oct 2016
ATP6V0A2	Cutis laxa, autosomal recessive, type IIA	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22773132, 10406678	SK, SEIZ, HYPOTO, DERM	Sept 2016 - Oct 2016
ATP6V1B1	Renal tubular acidosis & hearing loss	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9916796, 12414817,	HL, REN	Sept 2016 - Oct 2016
ATP7A	Menkes syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17717039, 15981243	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ATP7A	Occipital horn syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8914740, 7887410, 1	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ATP7A	Spinal muscular atrophy, distal, X-linked 3	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19153371, 20170900	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ATP7B	Wilson disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8298639, 10441329,	BIL, IEM	Sept 2016 - Oct 2016
ATP8B1	Cholestasis, progressive familial intrahepatic 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15239083, 9500542,	BIL	Sept 2016 - Oct 2016
ATR	Seckel syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	8358044, 12640452,	SEIZ	Sept 2016 - Oct 2016
ATRX	Alpha-thalassemia/mental retardation syndrome	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9326931, 18409179,	AN_TH, SEIZ	Sept 2016 - Oct 2016
AUH	3-methylglutaconic aciduria, type I	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	112434311, 7130438	IEM, HYPOTO	Sept 2016 - Oct 2016
AVPR2	Diabetes insipidus, nephrogenic	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10820168, 8104196,	REN, SEIZ	Sept 2016 - Oct 2016
AXL	Hypogonadotropic hypogonadism	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24476074		Sept 2016 - Oct 2016
B3GALTL	Peters-Plus syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18798333, 16909395	THYR, HYPOTO	Sept 2016 - Oct 2016
B3GAT3	Multiple joint dislocations, short stature, craniofacial dysmorphism, and congenital heart defects	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21763480, 23664117	SK, HYPOTO	Sept 2016 - Oct 2016
B4GALT1	CDG syndrome type IId	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	11901181	SEIZ, HYPOTO	Sept 2016 - Oct 2016
B9D2	Meckel syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21763481	REN	Sept 2016 - Oct 2016
BAAT	Bile acid amidation defect	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12704386	BIL, AN_TH	Sept 2016 - Oct 2016
BAG3	Cardiomyopathy, dilated	Strong	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood		21898660, 21353195	CM, HYPOTO	Sept 2016 - Oct 2016
BAG3	Myopathy, myofibrillar	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19085932, 21361913	CM, HYPOTO	Sept 2016 - Oct 2016
BANF1	Progeroid syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21549337, 21932319	DERM	Sept 2016 - Oct 2016
BARD1	Tetralogy of Fallot	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22912587	CHD	Sept 2016 - Oct 2016
BBS1	Bardet-Biedl syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12118255, 12524598	REN	Sept 2016 - Oct 2016
BBS10	Bardet-Biedl syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16823392, 16582908	REN	Sept 2016 - Oct 2016
BBS12	Bardet-Biedl syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17160889, 20827784	REN	Sept 2016 - Oct 2016
BBS2	Bardet-Biedl syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11285252, 11567139	REN	Sept 2016 - Oct 2016
BBS4	Bardet-Biedl syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11381270, 12016587	REN	Sept 2016 - Oct 2016
BBS5	Bardet-Biedl syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15137946, 18203199	REN	Sept 2016 - Oct 2016
BBS7	Bardet-Biedl syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12567324, 9402160,	REN	Sept 2016 - Oct 2016
BBS9	Bardet-Biedl syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16380913, 20177705	REN	Sept 2016 - Oct 2016
BCKDHA	Maple syrup urine disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2703538, 9609836, 9	IEM, SEIZ	Sept 2016 - Oct 2016
BCKDHB	Maple syrup urine disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2022752, 14517957,	IEM, SEIZ	Sept 2016 - Oct 2016
BCL9	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
BCS1L	Complex 3 deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11528392, 12215968,	22277166, 17403714, 2047	Sept 2016 - Oct 2016
BDNF	Central hypoventilation syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		11840487	PULM	Sept 2016 - Oct 2016
BICD2	Congenital spinal muscular atrophy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24482476, 23664116	HYPOTO	Sept 2016 - Oct 2016
BIN1	Myopathy, centronuclear, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20927630, 21129173	HYPOTO	Sept 2016 - Oct 2016
BLM	Bloom syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7585968, 17407155,	DERM	Sept 2016 - Oct 2016
BLOC1S3	Hermansky-Pudlak syndrome 8	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16385460, 22709368	PULM, DERM	Sept 2016 - Oct 2016
BLOC1S6	Hermansky-Pudlak syndrome 9	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22461475, 21665000	PULM, DERM	Sept 2016 - Oct 2016
BMPR1A	Juvenile polyposis syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11536076, 11381269	CHD	Sept 2016 - Oct 2016
BMPR1A	Tetralogy of Fallot	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24127225	CHD	Sept 2016 - Oct 2016
BMPR2	Pulmonary hypertension, familial primary	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood	Y	10973254, 6703480,	CHD, PULM	Sept 2016 - Oct 2016
BNC2	Total anomalous pulmonary venous return	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21368915		Sept 2016 - Oct 2016
BPGM	Erythrocytosis due to bisphosphoglycerate mutase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		1421379, 25015942	AN_TH	Sept 2016 - Oct 2016
BRAF	Cardiofaciocutaneous syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16474404, 19206169	AN_TH, CHD, SEIZ, DERM	Sept 2016 - Oct 2016
BRAF	LEOPARD syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19206169	AN_TH, CHD, SEIZ, DERM	Sept 2016 - Oct 2016
BRCA1	Breast-ovarian cancer, familial, 1	Definitive	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		7894491, 7894492, 7	AN_TH	Sept 2016 - Oct 2016
BRCA2	Breast-ovarian cancer, familial, 2	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		8524414, 11257103,	AN_TH	Sept 2016 - Oct 2016
BRCA2	Fanconi anemia, complementation group D1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16115142, 16825431	AN_TH	Sept 2016 - Oct 2016
BSCL2	Silver spastic paraplegia syndrome	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		23553728, 15358725	HYPOTO	Sept 2016 - Oct 2016
BSCL2	Berardinelli-Seip lipodystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12362029, 11479539	HYPOTO	Sept 2016 - Oct 2016
BSND	Bartter syndrome with sensorineural deafness	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11687798, 16328537	HL, REN	Sept 2016 - Oct 2016
BTD	Biotinidase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7550325, 2502673, 1	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
BTK	Agammaglobulinemia, X-linked 1	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16159644, 9445504,	9486400, 15024743, 816205	Sept 2016 - Oct 2016
BVES	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		2340379	CHD	Sept 2016 - Oct 2016
C10ORF2	Spinocerebellar ataxia infantile-onset	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21681116, 8133312,	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
C15ORF41	Congenital dyserythropoietic anemia type I	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23716552	BIL, AN_TH	Sept 2016 - Oct 2016
C3	Haemolytic uraemic syndrome	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		20595690, 16621965	BIL, THROM, AN_TH, REN	Sept 2016 - Oct 2016
CA2	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9143915, 1301935, 1	REN, SK	Sept 2016 - Oct 2016
CACNA1A	Episodic ataxia, type 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19633872, 10371528	SEIZ	Sept 2016 - Oct 2016
CACNA1C	Brugada syndrome	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		17224476, 12676817	COND, GLY, SEIZ	Sept 2016 - Oct 2016
CACNA1D	Sinoatrial node dysfunction and deafness	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21131953	HL	Sept 2016 - Oct 2016
CACNA1F	Night blindness, congenital stationary (complete), 1A, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10900517, 15761389	SEIZ	Sept 2016 - Oct 2016
CACNA1S	Malignant hyperthermia	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9199552, 20861472,	25658027, 20431982, 19825	Sept 2016 - Oct 2016
CACNA2D1	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20817017, 25527503	COND	Sept 2016 - Oct 2016
CACNB2	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17224476, 12676817	COND	Sept 2016 - Oct 2016
CAPN3	Muscular dystrophy, limb-girdle, type 2A	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7720071, 7670461, 1	HYPOTO	Sept 2016 - Oct 2016
CARS2	Epileptic encephalopathy	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		25787132, 25361775	SEIZ	Sept 2016 - Oct 2016
CASK	Mental retardation and microcephaly with pontine and cerebellar hypoplasia	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19165920, 21735175	CHD, SEIZ	Sept 2016 - Oct 2016
CASP10	Autoimmune lymphoproliferative syndrome II	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10412980, 16446975	AN_TH	Sept 2016 - Oct 2016
CASQ2	Ventricular tachycardia, catecholaminergic polymorphic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21618644, 12386154	CM, COND	Sept 2016 - Oct 2016
CAV3	Cardiomyopathy, familial hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		14672715	CM, COND, HYPOTO	Sept 2016 - Oct 2016
CAV3	Long QT syndrome-9	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		17060380, 23631430	CM, COND, HYPOTO	Sept 2016 - Oct 2016
CAV3	Muscular dystrophy, limb-girdle, type IC,	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9537420, 9536092, 1	CM, COND, HYPOTO	Sept 2016 - Oct 2016
CAV3	Caveolinopathy	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15580566, 18930476	CM, COND, HYPOTO	Sept 2016 - Oct 2016
CAV3	Rippling muscle disease	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11431690, 12666119	CM, COND, HYPOTO	Sept 2016 - Oct 2016
CBL	Noonan syndrome-like disorder with or without juvenile meylomonocytic leukemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20619386, 2449367	CHD	Sept 2016 - Oct 2016
CBS	Homocystinuria, B6-responsive and nonresponsive types	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10364517, 8554066,	THROM, IEM	Sept 2016 - Oct 2016
CC2D2A	Joubert syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18950740, 22241855	BIL, REN, SK, SEIZ	Sept 2016 - Oct 2016
CCDC103	Primary ciliary dyskinesia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		2258122, 22581229,	PULM	Sept 2016 - Oct 2016
CCDC39	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21131972, 22693285	PULM	Sept 2016 - Oct 2016
CCDC40	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22693285, 23255504	PULM	Sept 2016 - Oct 2016
CCDC50	Hearing loss	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17503326, 24875298	HL	Sept 2016 - Oct 2016

CCDC78	Congenital myopathy with prominent internal nuclei and atypical cores	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22818856, 25635128	HYPOTO		Sept 2016 - Oct 2016
CCDC88C	Hydrocephalus	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23042809, 23042809	SEIZ		Sept 2016 - Oct 2016
CD2AP	Glomerulosclerosis, focal segmental, 3	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17713465, 18443213	REN		Sept 2016 - Oct 2016
CD36	Platelet glycoprotein IV deficiency	Strong	AR	LOW (A)	Yes	C	No	Low penetrance, non-disease trait, clinical relevance of CD36 deficiency is un		11352982, 10890433	AN_TH		Sept 2016 - Oct 2016
CD40LG	Immunodeficiency, X-linked, with hyper-IgM	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8097258, 8550833, 7	AN_TH		Sept 2016 - Oct 2016
CD46	Haemolytic uraemic syndrome	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		16621965, 17089378	BIL, THROM, AN_TH, REN		Sept 2016 - Oct 2016
CD96	C syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	3981579, 17847009	DERM		Sept 2016 - Oct 2016
CDAN1	Anemia, congenital dyserythropoietic, type I	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16141353, 16098079	BIL, AN_TH		Sept 2016 - Oct 2016
CDAN3	Congenital dyserythropoietic anemia type 3	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		7711721	AN_TH		Sept 2016 - Oct 2016
CDH1	Orofacial clefts	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23197654, 26123647			Sept 2016 - Oct 2016
CDH1	Gastric cancer	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		10477433, 20373070,	19269290, 16924464, 2370		Sept 2016 - Oct 2016
CDH23	Deafness, autosomal recessive	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1785063, 11090341,	17850630 12075507		Sept 2016 - Oct 2016
CDH23	Usher syndrome, type 1D	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21940737, 21569298	HL		Sept 2016 - Oct 2016
CDK5RAP2	Microcephaly 3, primary, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15793586, 17764569	SEIZ		Sept 2016 - Oct 2016
CDKL5	Epileptic encephalopathy, early infantile, 2	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22872100, 19793311	SEIZ		Sept 2016 - Oct 2016
CDKN1C	Beckwith-Wiedemann syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9341892, 20503313,	THYR, GLY, SK		Sept 2016 - Oct 2016
CDKN2A	Melanoma	Definitive	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood		9425228, 21801156,	9856841, 15761864, 798738		Sept 2016 - Oct 2016
CDON	Holoprosencephaly	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21802063, 26728615	SEIZ		Sept 2016 - Oct 2016
CDSN	Hypotrichosis	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12754508, 16307662	DERM		Sept 2016 - Oct 2016
CDT1	Meier-Gorlin syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21358632, 21358631	HYPOTO		Sept 2016 - Oct 2016
CEACAM16	Hearing loss, autosomal dominant	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21368133, 26648831	HL		Sept 2016 - Oct 2016
CENPJ	Primary microcephaly	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15793586, 16900296	SEIZ		Sept 2016 - Oct 2016
CEP152	Seckel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21131973, 26436113	SEIZ		Sept 2016 - Oct 2016
CEP290	Joubert syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17617513, 16682973	REN, SK, SEIZ		Sept 2016 - Oct 2016
CEP41	Joubert syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22246503	REN		Sept 2016 - Oct 2016
CFB	Haemolytic uraemic syndrome	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		19584399, 20513133	BIL, THROM, AN_TH, REN		Sept 2016 - Oct 2016
CFC1	Congenital heart defects	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18538293, 19853937	CHD		Sept 2016 - Oct 2016
CFD	Complement factor D deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16527897, 11457876	AN_TH		Sept 2016 - Oct 2016
CFH	Haemolytic uraemic syndrome	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		12960213, 20513133	BIL, THROM, AN_TH, REN		Sept 2016 - Oct 2016
CFHR1	Haemolytic uraemic syndrome	Moderate	AR	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		19745068, 19861685	BIL, AN_TH, REN		Sept 2016 - Oct 2016
CFHR3	Haemolytic uraemic syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22626820, 16621965	BIL, AN_TH, REN		Sept 2016 - Oct 2016
CFHR4	Hemolytic-uremic syndrome, atypical, susceptibility to	Limited	UNKNOW	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		15562282, 10622723	AN_TH		Sept 2016 - Oct 2016
CFHR5	Haemolytic uraemic syndrome	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		20513133, 17000000	THROM, AN_TH, REN		Sept 2016 - Oct 2016
CFI	Haemolytic uraemic syndrome	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		16621965, 17599974	BIL, THROM, AN_TH, REN		Sept 2016 - Oct 2016
CFL2	Nemaline myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17160903, 22343409	HYPOTO		Sept 2016 - Oct 2016
CFP	Properdin deficiency, X-linked	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	3141111, 10909851,	8530058, 22229731		Sept 2016 - Oct 2016
CFTR	Cystic fibrosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10103316, 15528020	BIL, PULM, BOW		Sept 2016 - Oct 2016
CHAT	Congenital myasthenic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11172068, 3010100,	HYPOTO		Sept 2016 - Oct 2016
CHD2	Developmental delay, intellectual disability, epilepsy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24834135, 24932903	SEIZ		Sept 2016 - Oct 2016
CHD7	CHARGE syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2596527, 16207732,	HL, CHD, HYPOTO		Sept 2016 - Oct 2016
CHEK2	Breast cancer, susceptibility to	Strong	AD	LOW (A)	No	C	No	Adult-onset, Not actionable in childhood		11479295, 12454775,	22419737, 22114986		Sept 2016 - Oct 2016
CHKB	Muscular dystrophy, congenital, megaconial type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21665002, 23692895	HYPOTO		Sept 2016 - Oct 2016
CHM	Choroideremia	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	5146581, 17698759,	2220804, 23811034, 159890		Sept 2016 - Oct 2016
CHRM2	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18451336, 23743182	CM		Sept 2016 - Oct 2016
CHRNA1	Congenital myasthenic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15367858, 18707767	HYPOTO		Sept 2016 - Oct 2016
CHRNA2	Epilepsy	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		16826524, 21703448	SEIZ		Sept 2016 - Oct 2016
CHRNB1	Congenital myasthenic syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		8651643, 8872460, 2	HYPOTO		Sept 2016 - Oct 2016
CHRND	Congenital myasthenic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18398509, 16916845	HYPOTO		Sept 2016 - Oct 2016
CHRNE	Congenital myasthenic syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7531341, 7538206, 8	HYPOTO		Sept 2016 - Oct 2016
CHRNG	Pterygium syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16826520, 22167768	HYPOTO		Sept 2016 - Oct 2016
CHST3	Larsen syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18513679, 2253933,	SK, HYPOTO		Sept 2016 - Oct 2016
CHSY1	Temtamy preaxial brachydactyly syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21129728, 21129727	SK, HYPOTO		Sept 2016 - Oct 2016
CIRH1A	North American Indian childhood cirrhosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12417987, 11045837,	22916032, 18021715, 1622		Sept 2016 - Oct 2016
CISD2	Wolfram syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17846994, 10739754	HL, IEM, HYPOTO		Sept 2016 - Oct 2016
CITED2	Congenital heart defects	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16287139, 24848765	CHD		Sept 2016 - Oct 2016
CLCN1	Myotonia congenita	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11840191,17932099,	HYPOTO		Sept 2016 - Oct 2016
CLCN5	Dent disease	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8559248, 1372109, 1	REN, SK		Sept 2016 - Oct 2016
CLCN7	Osteopetrosis	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		19953639, 16234969	AN_TH, SK, SEIZ		Sept 2016 - Oct 2016
CLDN1	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	12164927, 15521008	BIL, DERM		Sept 2016 - Oct 2016
CLDN14	Hearing loss, non-syndromic, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11163249, 22246673	HL		Sept 2016 - Oct 2016
CLDN19	Hypomagnesemia 5, renal, with ocular involvement	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17033971, 7947033,	REN		Sept 2016 - Oct 2016
CLMP	Congenital short-bowel syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22155368	BOW		Sept 2016 - Oct 2016
CLN3	Ceroid lipofuscinosis, neuronal, 3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9311735, 21990111,	IEM, SEIZ		Sept 2016 - Oct 2016
CLN5	Ceroid lipofuscinosis, neuronal, 5	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9662406, 10477428,	IEM, SEIZ		Sept 2016 - Oct 2016
CLN6	Ceroid lipofuscinosis, neuronal, 6	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15996215, 11727201	IEM, SEIZ		Sept 2016 - Oct 2016
CLN8	Ceroid lipofuscinosis, neuronal, 8	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10508524, 15024724	IEM, SEIZ		Sept 2016 - Oct 2016
CLPP	Perrault syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23541340, 25956234	HL		Sept 2016 - Oct 2016
CLRN1	Usher syndrome, type 3A	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7711740, 11524702	HL		Sept 2016 - Oct 2016
CNGB3	Achromatopsia-3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10958649, 10466422,	15657609, 10888875, 2555		Sept 2016 - Oct 2016
CNTNAP2	Autism spectrum disorder	Moderate	COMPLEX	MODERATE (B)	Yes	C	No	Moderate penetrance, Not actionable in childhood		18179893, 20808228	SEIZ		Sept 2016 - Oct 2016
COCH	Deafness, non-syndromic, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9806553, 9931344, 2	HL		Sept 2016 - Oct 2016
COG4	Congenital disorder of glycosylation, type IIj	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21185756, 19494034	SEIZ, HYPOTO		Sept 2016 - Oct 2016
COG5	Congenital disorder of glycosylation, type IIi	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23430875, 23228021	SEIZ, HYPOTO		Sept 2016 - Oct 2016
COG7	Congenital disorder of glycosylation, type IIe	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15107842, 17356545	SEIZ, HYPOTO		Sept 2016 - Oct 2016
COL11A1	Stickler syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		110573014, 1528616	HL, SK		Sept 2016 - Oct 2016
COL11A2	Otospondylomegaepiphyseal dysplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7859284, 10677296,	HL, IEM, SK, DERM		Sept 2016 - Oct 2016
COL17A1	Epidermolysis bullosa, junctional, non-Herlitz type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18374450, 2135794	DERM		Sept 2016 - Oct 2016
COL1A1	Osteogenesis imperfecta, type I	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		458828, 7023758, 94	SK, HYPOTO, DERM		Sept 2016 - Oct 2016
COL1A1	Caffey disease	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		15864348, 15864348	SK, HYPOTO, DERM		Sept 2016 - Oct 2016
COL1A2	Osteogenesis imperfecta, type II	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	458828, 7023758, 21	SK, DERM		Sept 2016 - Oct 2016
COL2A1	Stickler syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16752401, 17721977	HL, IEM, SK, DERM		Sept 2016 - Oct 2016
COL3A1	Ehlers-Danlos syndrome, type IV	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8884076, 9399899, 1	DERM		Sept 2016 - Oct 2016
COL4A3	Alport syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15954103, 12028435	HL, REN		Sept 2016 - Oct 2016
COL4A4	Alport syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	24052634, 24854265	HL, REN		Sept 2016 - Oct 2016
COL4A5	Alport syndrome	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2904407, 1635357, 1	HL, REN		Sept 2016 - Oct 2016
COL5A1	Ehlers-Danlos syndrome, type I	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22696272, 8673139,	DERM		Sept 2016 - Oct 2016
COL5A2	Ehlers-Danlos syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9425231, 22696272 ,	DERM		Sept 2016 - Oct 2016
COL6A1	Ullrich congenital muscular dystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15955946, 16278855	HYPOTO		Sept 2016 - Oct 2016
COL6A2	Ullrich congenital muscular dystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8782832, 20302629,	HYPOTO		Sept 2016 - Oct 2016
COL6A3	Ullrich congenital muscular dystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10399756, 11992252	HYPOTO		Sept 2016 - Oct 2016
COL7A1	Epidermolysis bullosa dystrophica	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10827412, 9892921,	DERM		Sept 2016 - Oct 2016
COL9A1	Stickler syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Limited evidence for gene's role in disease		21421862, 16909383	HL, SK, DERM		Sept 2016 - Oct 2016
COL9A2	Stickler syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21671392	HL, SK, DERM		Sept 2016 - Oct 2016
COLQ	Congenital myasthenic syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3010100, 11865139,	HYPOTO		Sept 2016 - Oct 2016
COQ2	Coenzyme Q10 deficiency, primary, 1	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	16400613, 17855635	IEM, REN, SEIZ, HYPOTO		Sept 2016 - Oct 2016
COQ6	Nephrotic syndrome with sensorineural deafness	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21540551, 24140869	IEM, REN, HYPOTO		Sept 2016 - Oct 2016
COX4I2	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19268275	AN_TH		Sept 2016 - Oct 2016
CP	Aceruloplasminaemia	Definitive	AR	HIGH (A)	No	B	Yes	Adult-onset, Actionable in childhood		7708681, 7539672,	7755360, 19095659		Sept 2016 - Oct 2016
CPOX	Coproporphyria	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		7757079, 9843038, 7	SEIZ		Sept 2016 - Oct 2016
CPS1	Carbamoylphosphate synthetase I deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19793055, 17310273	CHD, IEM		Sept 2016 - Oct 2016
CPT1A	Carnitine palmitoyltransferase I deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7014807, 9691089, 1	COND, IEM, GLY, SEIZ		Sept 2016 - Oct 2016
CPT2	Carnitine palmitoyltransferase 2 deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12673791, 16996287	BIL, COND, IEM, GLY, SEIZ, H		Sept 2016 - Oct 2016
CPZ	Autism	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22495306, 24859339			Sept 2016 - Oct 2016
CR2	Hypogammaglobulinaemia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		26325596, 2632559	REN		Sept 2016 - Oct 2016
CREBBP	Rubinstein-Taybi syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12749047, 20717166	SEIZ		Sept 2016 - Oct 2016
CRELD1	Cardiac atrioventricular septal defect	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease		21080147, 12632326	CHD		Sept 2016 - Oct 2016
CRLF1	Crisponi syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17436252, 19012339,	24488861, 17436251		Sept 2016 - Oct 2016
CRTAP	Osteogenesis imperfecta, type VII	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12110406, 17055431	SK		Sept 2016 - Oct 2016
CRYAB	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	No	B	Yes	Moderate evidence, Actionable in childhood		16793013, 16483541	CM, HYPOTO		Sept 2016 - Oct 2016
CRYAB	Myofibrillar myopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14681890, 9731540,	CM, HYPOTO		Sept 2016 - Oct 2016
CSF1R	Leukoencephalopathy, diffuse hereditary, with spheroids	Definitive	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		22197934, 23038421	SEIZ, HYPOTO		Sept 2016 - Oct 2016
CSF2RA	Pulmonary alveolar proteinosis	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18955570, 20622029	PULM		Sept 2016 - Oct 2016
CSF2RB	Pulmonary alveolar proteinosis	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21205713, 21075760	PULM		Sept 2016 - Oct 2016
CSRP3	Cardiomyopathy, dilated, 1M	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18505755, 12507422	CM		Sept 2016 - Oct 2016
CSRP3	Cardiomyopathy, familial hypertrophic, 12	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		18505755, 12642359	CM		Sept 2016 - Oct 2016
CSTA	Exfoliative ichthyosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21944047, 2353470	DERM		Sept 2016 - Oct 2016
CSTB	Epilepsy, progressive myoclonic 1A	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8596935, 20548044, 1	SEIZ		Sept 2016 - Oct 2016
CTC1	Coats plus syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18005359, 9886310,	AN_TH, DERM		Sept 2016 - Oct 2016
CTDP1	Congenital cataracts - facial dysmorphism - neuropathy	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10439962	HYPOTO		Sept 2016 - Oct 2016
CTF1	Cardiomyopathy, dilated	Limited	UNKNOW	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		11058912	CM		Sept 2016 - Oct 2016
CTNS	Cystinosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9537412, 9792862, 1	THYR, IEM, REN		Sept 2016 - Oct 2016
CTSD	Ceroid lipofuscinosis, neuronal, 10	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16670177, 16685649	IEM, SEIZ		Sept 2016 - Oct 2016
CTSK	Pycnodysostosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10074491 , 17397052	IEM, SK		Sept 2016 - Oct 2016
CUBN	Megaloblastic anemia-1, Finnish type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22929189, 10080186	AN_TH, REN		Sept 2016 - Oct 2016

CUL7	3-M syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21364696, 21396581	SK	Sept 2016 - Oct 2016
CYBA	Chronic granulomatous disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10910929, 1415254,	BOW, AN_TH, REN	Sept 2016 - Oct 2016
CYBB	Chronic granulomatous disease	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1710153, 8634410, 1	BOW, AN_TH	Sept 2016 - Oct 2016
CYCS	Thrombocytopenia 4	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18345000, 24326104	AN_TH	Sept 2016 - Oct 2016
CYP11A1	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22968487, 19116240		Sept 2016 - Oct 2016
CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2022736, 8506298,	20089618, 23146819, 259114	Sept 2016 - Oct 2016
CYP21A2	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9556656, 9521938, 7	GLY	Sept 2016 - Oct 2016
CYP27A1	Cerebrotendinous xanthomatosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7315872, 2019602, 7	BIL, SEIZ, HYPOTO	Sept 2016 - Oct 2016
CYP27B1	Vitamin D-dependent rickets, type I	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17488797, 9837822,	9486994, 12050193 , 225881	Sept 2016 - Oct 2016
CYP4F22	Ichthyosis, congenital, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16436457, 10712205	DERM	Sept 2016 - Oct 2016
CYP7A1	Hypercholesterolemia due to cholesterol 7alpha-hydroxylase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12093894	BIL	Sept 2016 - Oct 2016
CYP7B1	Cholestasis, severe	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9802883, 18367963	BIL, HYPOTO	Sept 2016 - Oct 2016
D2HGDH	D-2-hydroxyglutaric aciduria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20020533, 16037974	SEIZ	Sept 2016 - Oct 2016
DAG1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21388311, 24361964	HYPOTO	Sept 2016 - Oct 2016
DAPK3	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
DBH	Dopamine beta-hydroxylase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15060114, 11857564	GLY	Sept 2016 - Oct 2016
DBT	Maple syrup urine disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8037208, 13813934,	IEM, SEIZ	Sept 2016 - Oct 2016
DCLRE1C	Severe combined immunodeficiency, Athabascan type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1916741, 10456341,	DERM	Sept 2016 - Oct 2016
DCTN1	Amyotrophic lateral sclerosis	Moderate	AD	LOW (B)	No	C	No	Adult-onset, Not actionable in childhood		15326253, 25109764,	25299611, 26742954	Sept 2016 - Oct 2016
DCX	Lissencephaly, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19342486, 18685874	SEIZ	Sept 2016 - Oct 2016
DCX	Lennox-Gastaut syndrome	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	20726879	SEIZ	Sept 2016 - Oct 2016
DDB2	Xeroderma pigmentosum	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20346687, 11338401	DERM	Sept 2016 - Oct 2016
DDC	Aromatic L-amino acid decarboxylase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17240182, 15079002	SEIZ	Sept 2016 - Oct 2016
DDHD1	Spastic paraplegia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24989667, 23176821	HYPOTO	Sept 2016 - Oct 2016
DDOST	Congenital disorder of glycosylation, type Ir	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22305527	SEIZ, HYPOTO	Sept 2016 - Oct 2016
DDR2	Spondylometaepiphyseal dysplasia, short limb-hand type	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19110212, 20223752	SK	Sept 2016 - Oct 2016
DECR1	2,4-Dienoyl-CoA reductase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		2332510, 19578400	GLY	Sept 2016 - Oct 2016
DES	Cardiomyopathy, dilated	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		17325244, 10430757	CM, COND, HYPOTO	Sept 2016 - Oct 2016
DES	Myopathy, myofibrillar	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14711882, 9697706,	CM, COND, HYPOTO	Sept 2016 - Oct 2016
DFNA5	Hearing loss	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9771715, 17868390,	HL	Sept 2016 - Oct 2016
DFNB31	Hearing loss	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24194196, 15841483	HL	Sept 2016 - Oct 2016
DFNB59	Hearing loss	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17373699, 17718865	HL	Sept 2016 - Oct 2016
DGKE	Haemolytic uraemic syndrome, atypical	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		23542698, 25135762	BIL, AN_TH, REN	Sept 2016 - Oct 2016
DGUOK	Mitochondrial DNA depletion syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12205643, 15883261	BIL, IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
DHCR24	Desmosterolosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11519011, 21671375	SK, HYPOTO	Sept 2016 - Oct 2016
DHCR7	Smith-Lemli-Opitz syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15954111, 9653161,	BIL, PULM, IEM, SEIZ	Sept 2016 - Oct 2016
DIABLO	Deafness, autosomal dominant	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21722859, 26969326	HL	Sept 2016 - Oct 2016
DIAPH1	Hearing loss	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		9360932, 22938506,	HL	Sept 2016 - Oct 2016
DKC1	Dyskeratosis congenita	Definitive	XLR	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood	Y	18005359, 9886310,	DERM	Sept 2016 - Oct 2016
DLC1	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24587289, 24476948	CHD	Sept 2016 - Oct 2016
DLD	Maple syrup urine disease, type III	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8037208, 13813934,	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
DLL3	Spondylocostal dysostosis, autosomal recessive, 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12746394, 12791036	SK, HYPOTO	Sept 2016 - Oct 2016
DMD	Duchenne muscular dystrophy	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19945913, 19937601	CM, HYPOTO	Sept 2016 - Oct 2016
DMD	Becker muscular dystrophy	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3384440, 19937601,	CM, HYPOTO	Sept 2016 - Oct 2016
DMD	Cardiomyopathy, dilated	Definitive	XLD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9170393, 12565910,	CM, HYPOTO	Sept 2016 - Oct 2016
DMP1	Hypophosphatemic rickets, AR	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17033625, 22695891	REN, SK	Sept 2016 - Oct 2016
DMPK	Myotonic dystrophy 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1310900, 16285929,	HYPOTO	Sept 2016 - Oct 2016
DNAAF1	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19944405, 19944400	PULM	Sept 2016 - Oct 2016
DNAAF2	Primary ciliary dyskinesia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		1905262, 24498942	PULM	Sept 2016 - Oct 2016
DNAAF3	Primary ciliary dyskinesia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22387996	PULM	Sept 2016 - Oct 2016
DNAH11	Primary ciliary dyskinesia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22184204, 24450482	PULM, CHD	Sept 2016 - Oct 2016
DNAH5	Primary ciliary dyskinesia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11788826, 16627867	PULM	Sept 2016 - Oct 2016
DNAI1	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11713099, 16858015	PULM	Sept 2016 - Oct 2016
DNAI2	Primary ciliary dyskinesia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18950741, 23261302	PULM	Sept 2016 - Oct 2016
DNAJB6	Muscular dystrophy, limb girdle	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21376592, 2236678	HYPOTO	Sept 2016 - Oct 2016
DNAJC19	3-methylglutaconic aciduria, type V	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	22797137	IEM, HYPOTO	Sept 2016 - Oct 2016
DNAJC5	Neuronal ceroid lipofuscinosis, adult-onset	Strong	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		21820099, 22073189	SEIZ	Sept 2016 - Oct 2016
DNAL1	Primary ciliary dyskinesia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21496787	PULM	Sept 2016 - Oct 2016
DNM2	Myopathy, centronuclear	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22396310, 17932957	HYPOTO	Sept 2016 - Oct 2016
DNM2	Charcot-Marie-Tooth disease, axonal, type 2M	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15731758, 19502294	HYPOTO	Sept 2016 - Oct 2016
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11102980 , 21559330		Sept 2016 - Oct 2016
DOCK8	Hyper-IgE syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20004785, 20622910	PULM	Sept 2016 - Oct 2016
DOK7	Congenital myasthenic syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18626973, 16917026	HYPOTO	Sept 2016 - Oct 2016
DOLK	Congenital disorder of glycosylation, type Im	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	23890587, 22242004	CM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
DPAGT1	Congenital disorder of glycosylation, type Ij	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22742743, 23249953	SEIZ, HYPOTO	Sept 2016 - Oct 2016
DPM1	Congenital disorder of glycosylation, type Ie	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	10642597, 10642602	SEIZ, HYPOTO	Sept 2016 - Oct 2016
DPP6	Ventricular fibrillation, paroxysmal familial, 2	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19285295	COND	Sept 2016 - Oct 2016
DPYD	Dihydropyrimidine dehydrogenase deficiency	Definitive	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	19296131, 10071185	SEIZ	Sept 2016 - Oct 2016
DSC2	Arrhythmogenic right ventricular cardiomyopathy	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		20031616, 17033975	CM, COND	Sept 2016 - Oct 2016
DSG2	Arrhythmogenic right ventricular cardiomyopathy	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		17505751, 1650573,	CM, COND	Sept 2016 - Oct 2016
DSP	Arrhythmogenic right ventricular dysplasia/cardiomyopathy	Definitive	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood	Y	12373648, 21606390	CM, COND, DERM	Sept 2016 - Oct 2016
DSP	Epidermolysis bullosa, lethal acantholytic	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16175511, 20302578	CM, COND, DERM	Sept 2016 - Oct 2016
DTHD1	Leber congenital amaurosis with myopathy	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23105016		Sept 2016 - Oct 2016
DTNA	Left ventricular noncompaction 1	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		18506004, 11238270	CM, COND, CHD	Sept 2016 - Oct 2016
DTNBP1	Hermansky-Pudlak syndrome 7	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12923531, 23364359	PULM, DERM	Sept 2016 - Oct 2016
DUOX2	Thyroid dyshormonogenesis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16134168, 15863666	THYR	Sept 2016 - Oct 2016
DUOXA2	Thyroid dyshormonogenesis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21367925, 18042646	THYR	Sept 2016 - Oct 2016
DYSF	Muscular dystrophy, limb-girdle, type 2B	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8012357, 16087766,	HYPOTO	Sept 2016 - Oct 2016
DYSF	Miyoshi muscular dystrophy 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8808603, 3942856, 1	HYPOTO	Sept 2016 - Oct 2016
ECE1	Hirschsprung disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9915973	BOW	Sept 2016 - Oct 2016
EDA	Ectodermal dysplasia, hypohidrotic	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9683615, 18510547,	DERM	Sept 2016 - Oct 2016
EDAR	Ectodermal dysplasia, hypohidrotic	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16329325, 18231121	DERM	Sept 2016 - Oct 2016
EDARADD	Ectodermal dysplasia, hypohidrotic	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17354266, 11780064	DERM	Sept 2016 - Oct 2016
EDN3	Hirschsprung disease	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	20009762, 9587491,	HL, PULM, BOW, DERM	Sept 2016 - Oct 2016
EDN3	Waardenburg syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20127975, 8630502,	HL, PULM, BOW, DERM	Sept 2016 - Oct 2016
EDNRB	Hirschsprung disease	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	17009072 , 2000976	HL, BOW, DERM	Sept 2016 - Oct 2016
EDNRB	Waardenburg syndrome	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		19764031, 20127975	HL, BOW, DERM	Sept 2016 - Oct 2016
EFEMP2	Cutis laxa, autosomal recessive, type IB	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17937443, 16685658	PULM, DERM	Sept 2016 - Oct 2016
EFHC1	Myoclonic epilepsy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15258581, 17159113	SEIZ	Sept 2016 - Oct 2016
EFTUD2	Mandibulofacial dysostosis with microcephaly	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24470203, 22305528	CHD	Sept 2016 - Oct 2016
EGR2	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10502832, 9537424,	HYPOTO	Sept 2016 - Oct 2016
EIF2AK3	Wolcott-Rallison syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19837917, 15220213	SK	Sept 2016 - Oct 2016
EIF2B1	Leukoencephalopathy with vanishing white matter	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11835386, 18263758	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ELANE	Neutropenia, congenital	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10581030, 14962902	DERM	Sept 2016 - Oct 2016
ELN	Supravalvar aortic stenosis	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8096434, 11175284,	PULM, CHD, DERM	Sept 2016 - Oct 2016
EMD	Muscular dystrophy, Emery-Dreifuss	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7894480, 10377322,	CM, COND, HYPOTO	Sept 2016 - Oct 2016
ENG	Telangiectasia, hereditary hemorrhagic, type 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16752392, 7802026,	PULM, DERM	Sept 2016 - Oct 2016
ENPP1	Arterial calcification, generalized, of infancy, 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12881724, 15605415	SK	Sept 2016 - Oct 2016
EPB42	Spherocytosis	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		1558976, 8547605, 8	BIL, AN_TH	Sept 2016 - Oct 2016
EPCAM	Lynch syndrome	Strong	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		17539898, 15872200	BOW	Sept 2016 - Oct 2016
EPHX1	Hypercholanemia, familial	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12878321	AN_TH	Sept 2016 - Oct 2016
EPM2A	Epilepsy, progressive myoclonic 2A (Lafora)	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12019207, 15781812	SEIZ	Sept 2016 - Oct 2016
ERBB3	Lethal congenital contractural syndrome 2	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	17701904, 15378541	HYPOTO	Sept 2016 - Oct 2016
ERCC1	Xeroderma pigmentosum	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21612988, 23623389	HYPOTO, DERM	Sept 2016 - Oct 2016
ERCC2	Xeroderma pigmentosum	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1372108, 1757099, 9	AN_TH, DERM	Sept 2016 - Oct 2016
ERCC3	Xeroderma pigmentosum	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16947863, 26884178	DERM	Sept 2016 - Oct 2016
ERCC4	Xeroderma pigmentosum	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	9580660, 8797827, 2	AN_TH, DERM	Sept 2016 - Oct 2016
ERCC5	Xeroderma pigmentosum	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12060391, 7951246,	DERM	Sept 2016 - Oct 2016
ERCC6	Cockayne syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	641373, 16865293, 1	SK, SEIZ, DERM	Sept 2016 - Oct 2016
ERCC8	Cockayne syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	641373, 16865293, 1	SK, SEIZ, DERM	Sept 2016 - Oct 2016
ESCO2	Roberts syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19574259, 15821733	SK	Sept 2016 - Oct 2016
ESPN	Hearing loss	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15286153, 15930085	HL	Sept 2016 - Oct 2016
ESRRB	Hearing loss	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21802533, 22951369	HL	Sept 2016 - Oct 2016
ETFA	Glutaric acidemia IIA	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1882842, 1430199, 1	IEM,GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ETFB	Glutaric acidemia IIB	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7912128, 12815589,	IEM,GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ETFDH	Glutaric acidemia IIC	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17584774, 21347544	IEM,GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ETHE1	Ethylmalonic encephalopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20978941, 14732903	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
EVC	Ellis-van Creveld syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10700184, 23220543	SK	Sept 2016 - Oct 2016
EVC2	Ellis-van Creveld syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23220543, 19810119	SK	Sept 2016 - Oct 2016
EXT1	Exostoses, multiple, type 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		77726168, 8317501,	SK, HYPOTO	Sept 2016 - Oct 2016

EXT2	Exostoses, multiple, type 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9463333, 15586175, 2	SK, HYPOTO	Sept 2016 - Oct 2016
EYA1	Branchiootorenal syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19206155, 12404110	HL, REN	Sept 2016 - Oct 2016
EYA4	Deafness, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11159937, 17568404	HL, HYPOTO	Sept 2016 - Oct 2016
EZH2	Weaver syndrome 2	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22177091, 22190405	GLY	Sept 2016 - Oct 2016
F11	Factor XI deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	3369441, 10606881,	THROM, AN_TH	Sept 2016 - Oct 2016
F2	Prothrombin deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1334372, 1421398, 1	THROM	Sept 2016 - Oct 2016
F5	Risk for deep vein thrombosis	Definitive	AR	LOW (A)	Yes	C	No	Low penetrance	Y	8164741, 8979136, 1	THROM	Sept 2016 - Oct 2016
F8	Hemophilia A	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15741993, 11857744	AN_TH	Sept 2016 - Oct 2016
F9	Hemophilia B	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9556658, 5420360, 7	THROM, AN_TH	Sept 2016 - Oct 2016
FAAH2	Autism spectrum disorder	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23352160		Sept 2016 - Oct 2016
FAH	Tyrosinemia, type I	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11209059, 15759101	BIL, THROM, IEM, GLY, REN	Sept 2016 - Oct 2016
FAM111B	Hereditary fibrosing poikiloderma with tendon contracture, myopathy, and pulmonary fibrosis	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24268661, 26471370	HYPOTO	Sept 2016 - Oct 2016
FAM126A	Hypomyelination and congenital cataract	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17683097, 21911699	SEIZ, HYPOTO	Sept 2016 - Oct 2016
FAM134B	Neuropathy, hereditary sensory and autonomic, type IIB	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19838196, 24327336	HYPOTO	Sept 2016 - Oct 2016
FAM161A	Retinal dystrophy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20705279, 20705278,	24520187, 23591405, 2258	Sept 2016 - Oct 2016
FAM20C	Osteosclerotic bone dysplasia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17924334, 19250384	SK, HYPOTO	Sept 2016 - Oct 2016
FAM58A	Syndactyly - telecanthus - anogenital and renal malformations	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18297069, 20848651	REN	Sept 2016 - Oct 2016
FANCA	Fanconi anaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15790592, 15522956	AN_TH	Sept 2016 - Oct 2016
FANCB	Fanconi anaemia	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15502827, 23613520	AN_TH	Sept 2016 - Oct 2016
FANCC	Fanconi anaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8128956, 16429406, 2	AN_TH	Sept 2016 - Oct 2016
FANCD2	Fanconi anaemia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17436244, 11239453	AN_TH	Sept 2016 - Oct 2016
FANCE	Fanconi anaemia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11001585, 17924555	AN_TH	Sept 2016 - Oct 2016
FANCF	Fanconi anaemia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10615118, 16084127	AN_TH	Sept 2016 - Oct 2016
FANCG	Fanconi anaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12552564, 11093276	AN_TH	Sept 2016 - Oct 2016
FANCI	Fanconi anaemia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17452773, 17460694	AN_TH	Sept 2016 - Oct 2016
FANCL	Fanconi anaemia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23613520, 25754594	AN_TH	Sept 2016 - Oct 2016
FANCM	Fanconi anaemia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16116422, 21681190	AN_TH	Sept 2016 - Oct 2016
FBLN5	Age-related macular degeneration	Moderate	AD	MODERATE (B)	No	C	No	Adult-onset, moderate evidence, moderate penetrance	Y	15269314, 20007835	PULM, HYPOTO	Sept 2016 - Oct 2016
FBLN5	Cutis laxa	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19401719, 12189163	PULM, HYPOTO, DERM	Sept 2016 - Oct 2016
FBN1	Marfan's syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22461464, 17568394	PULM, SK, HYPOTO, DERM	Sept 2016 - Oct 2016
FBN1	Weill-Marchesani syndrome 2, dominant	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12525539, 17718856	PULM, SK, HYPOTO, DERM	Sept 2016 - Oct 2016
FBN1	Shprintzen-Goldberg syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		8563763, 16333834,	PULM, SK, HYPOTO, DERM	Sept 2016 - Oct 2016
FBN2	Contractural arachnodactyly	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10797416, 11754102	CHD, HYPOTO, SK, DERM	Sept 2016 - Oct 2016
FGA	Afibrinogenaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10891444, 11354637	AN_TH, REN	Sept 2016 - Oct 2016
FGB	Afibrinogenaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10666208, 12393540	AN_TH	Sept 2016 - Oct 2016
FGD1	Aarskog-Scott syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20082460, 11093277	SK, SEIZ	Sept 2016 - Oct 2016
FGD4	Charcot-Marie-Tooth disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	175649172, 1756495	HYPOTO	Sept 2016 - Oct 2016
FGF3	Deafness, congenital with inner ear agenesis, microtia, and microdontia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18701883, 21480479	HL	Sept 2016 - Oct 2016
FGFR1	Kallmann syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7874169, 12627230,	SK	Sept 2016 - Oct 2016
FGFR2	Pfeiffer syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7719345, 9475591, 9	SK	Sept 2016 - Oct 2016
FGFR2	Apert syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7719344, 9700203, 1	SK	Sept 2016 - Oct 2016
FGFR2	Crouzon syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10712195, 20635358	SK	Sept 2016 - Oct 2016
FGFR2	Beare-Stevenson cutis gyrata syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8696350, 19610084,	SK	Sept 2016 - Oct 2016
FGFR2	Jackson-Weiss syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7874170, 7806229, 8	SK	Sept 2016 - Oct 2016
FGFR3	Achondroplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16140722, 20081435	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	Hypochondroplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7670477, 9450868, 1	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	Crouzon syndrome with acanthosis nigricans	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11426459, 8880573,	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	Thanatophoric dysplasia type 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2596513, 8589699, 9	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	Muenke syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9042914, 9279753, 1	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	CATSHL syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17033969, 27139183	SK, SEIZ	Sept 2016 - Oct 2016
FGFR3	LADD syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16501574, 1415342	SK, SEIZ	Sept 2016 - Oct 2016
FGG	Afibrinogenaemia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10891444, 11354637	AN_TH	Sept 2016 - Oct 2016
FH	Leiomyomatosis and renal cell cancer	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		16597677, 20618355	IEM, GLY, SEIZ, HYPOTO, DEF	Sept 2016 - Oct 2016
FH	Fumarase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9635293, 11865300, 2	IEM, GLY, SEIZ, HYPOTO, DEF	Sept 2016 - Oct 2016
FHL1	Myofibrillar myopathy	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18274675, 22094483	HYPOTO	Sept 2016 - Oct 2016
FHL1	Emery-Dreifuss muscular dystrophy	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19716112, 19687455	HYPOTO	Sept 2016 - Oct 2016
FHL2	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		25358972	CM	Sept 2016 - Oct 2016
FKBPL	Infertility	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		20210997		Sept 2016 - Oct 2016
FKRP	Muscular dystrophy, limb girdle 2I	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14523375, 1266624,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
FKRP	Muscle-eye-brain disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15121789, 11320179	SEIZ, HYPOTO	Sept 2016 - Oct 2016
FKTN	Muscular dystrophy, Fukuyama	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17878207, 10545611	SEIZ, HYPOTO	Sept 2016 - Oct 2016
FKTN	Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14627679, 10545611	SEIZ, HYPOTO	Sept 2016 - Oct 2016
FLCN	Birt-Hogg-Dube syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18505456, 15657874	PULM, DERM	Sept 2016 - Oct 2016
FLG	Ichthyosis vulgaris	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		16444271, 19037238	DERM	Sept 2016 - Oct 2016
FLNA	Otopalatodigital spectrum disorder	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1733165, 16596676	BOW, CHD, SK, SEIZ, DERM	Sept 2016 - Oct 2016
FLNC	Myofibrillar myopathy	Moderate	AD	HIGH (B)	No	C	No	Adult-onset, moderate evidence		19050726, 14711882	CM, COND, HYPOTO	Sept 2016 - Oct 2016
FMO3	Trimethylaminuria	Definitive	AR	HIGH (A)	Yes	C	No	Non-disease phenotype		9398858, 9536088,	10479479, 23791655, 16600	Sept 2016 - Oct 2016
FOXC1	Axenfeld-Rieger syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17197537, 12210347	CHD, REN	Sept 2016 - Oct 2016
FOXC2	Lymphoedema, primary	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19760751, 12114478,	22768468, 11694548, 1137	Sept 2016 - Oct 2016
FOXE1	Bamforth-Lazarus syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12165566, 16882747	THYR, IEM	Sept 2016 - Oct 2016
FOXF1	Alveolar capillary dysplasia with misalignment of pulmonary veins	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19500772, 23505205	PULM, CHD, REN	Sept 2016 - Oct 2016
FOXF2	Disorders of sex development with cleft palate	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19276632		Sept 2016 - Oct 2016
FOXH1	Congenital heart defects	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18538293, 19933292	CHD, SEIZ	Sept 2016 - Oct 2016
FOXN1	Congenital alopecia with T-cell immunodeficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	10206641, 8911612,	DERM	Sept 2016 - Oct 2016
FOXP3	IPEX syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11137993, 14671208	THYR, AN_TH, BOW	Sept 2016 - Oct 2016
FRAS1	Fraser syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17163535, 12766769	REN	Sept 2016 - Oct 2016
FREM1	Manitoba oculotrichoanal syndrome	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		21507892, 19732862	REN	Sept 2016 - Oct 2016
FREM2	Fraser syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	18203166, 16894541	REN	Sept 2016 - Oct 2016
FSCN2	Retinitis pigmentosa	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24265693		Sept 2016 - Oct 2016
FTCD	Glutamate formiminotransferase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12815595	IEM	Sept 2016 - Oct 2016
FTL	Neuroferritinopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19176363, 7669675,	SEIZ	Sept 2016 - Oct 2016
FUCA1	Fucosidosis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2012122, 7095811, 1	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
FXN	Friedreich ataxia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7397485, 2899844, 1	HYPOTO	Sept 2016 - Oct 2016
G6PC	Glycogen storage disease Ia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10447271, 10612834	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
G6PC3	Neutropaenia, congenital	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19118303, 20799326,	22050868, 22469094, 2126	Sept 2016 - Oct 2016
G6PD	Glucose-6-phosphate dehydrogenase deficiency	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7949118, 9427729, 2	BIL, AN_TH, IEM	Sept 2016 - Oct 2016
GAA	Glycogen storage disease II	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15985590, 2403755,	IEM, HYPOTO	Sept 2016 - Oct 2016
GABRA1	Epilepsy, idiopathic generalised	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11992121, 21703448	SEIZ	Sept 2016 - Oct 2016
GABRG2	Epilepsy, childhood absence with febrile seizure	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11748509, 11326274	SEIZ	Sept 2016 - Oct 2016
GALC	Krabbe disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8940268, 9272171, 7	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
GALK1	Galactokinase deficiency with cataracts	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12705493, 10521295	IEM	Sept 2016 - Oct 2016
GALNS	Mucopolysaccharidosis IVA	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9298823, 15241807,	IEM, SEIZ	Sept 2016 - Oct 2016
GALT	Galactosaemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	6262213, 10408771,	BIL, IEM, GLY	Sept 2016 - Oct 2016
GAN	Giant axonal neuropathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11062483, 12655563	SEIZ, HYPOTO	Sept 2016 - Oct 2016
GATA1	Porphyria, congenital erythropoietic	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	17148589, 25251786	AN_TH, DERM	Sept 2016 - Oct 2016
GATA1	Dyserythropoietic anemia with thrombocytopenia	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11809723, 10700180	AN_TH, DERM	Sept 2016 - Oct 2016
GATA4	Congenital heart defects	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12845333, 15863664	COND, CHD	Sept 2016 - Oct 2016
GATA5	Familial atrial fibrillation	Moderate	AD	MODERATE (B)	No	C	No	Moderate evidence, Actionable in childhood		23295592, 23175127	COND, CHD	Sept 2016 - Oct 2016
GATA6	Atrial fibrillation	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		22750565, 22257684	COND, CHD	Sept 2016 - Oct 2016
GATAD1	Cardiomyopathy, dilated, 2B	Limited	AR	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21965549	CM	Sept 2016 - Oct 2016
GBA	Gaucher disease 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18338393, 2495719, 1	BIL, AN_TH, IEM, HYPOTO	Sept 2016 - Oct 2016
GBE1	Glycogen storage disease IV	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15452297, 15019703	HYPOTO	Sept 2016 - Oct 2016
GBE1	Polyglucosan body disease, adult form	Definitive	AR	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood, nontreatable disease, but allelic dise		8494336, 9851430, 1	HYPOTO	Sept 2016 - Oct 2016
GCDH	Glutaricaciduria, type I	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8139602, 8552212, 1	IEM, GLY	Sept 2016 - Oct 2016
GCH1	Dystonia, dopa-responsive	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9778264, 11346370,	IEM	Sept 2016 - Oct 2016
GCK	Hyperinsulinemic hypoglycemia, familial	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21454522, 12941786	GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
GCLC	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		10515893, 10733484	AN_TH	Sept 2016 - Oct 2016
GCSH	Glycine encephalopathy	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	11450847, 1671321,	SEIZ	Sept 2016 - Oct 2016
GDAP1	Charcot-Marie-Tooth disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11166163, 15805163	HYPOTO	Sept 2016 - Oct 2016
GDF1	Congenital heart defects	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17924340	PULM, CHD	Sept 2016 - Oct 2016
GDNF	Hirschsprung disease	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease		8896569, 8968758, 8	PULM, BOW, REN	Sept 2016 - Oct 2016
GDNF	Central hypoventilation syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9497256, 8896569	PULM, BOW, REN	Sept 2016 - Oct 2016
GFAP	Alexander disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11138011, 15732097	SEIZ, HYPOTO	Sept 2016 - Oct 2016
GFER	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19409522, 25269795	IEM, HYPOTO	Sept 2016 - Oct 2016
GFM1	Combined oxidative phosphorylation deficiency 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15537906, 16632485	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
GFPT1	Congenital myasthenic syndrome, limb-girdle	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21310273, 23794683	HYPOTO	Sept 2016 - Oct 2016
GIPC3	Hearing loss	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21326233, 21660509	HL	Sept 2016 - Oct 2016
GJA1	Oculodentodigital dysplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12457340, 19338053	HL, CHD, SEIZ, HYPOTO, DER	Sept 2016 - Oct 2016
GJA5	Atrial fibrillation	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		16790700, 20650941	COND, CHD	Sept 2016 - Oct 2016
GJB1	Charcot-Marie-Tooth neuropathy	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1674715, 17353473, 1	HYPOTO	Sept 2016 - Oct 2016

GJB2	Deafness	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15994881, 21776002	HL, DERM	Sept 2016 - Oct 2016
GJB2	Deafness and palmoplantar keratoderma	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8736341, 12372058,	HL, DERM	Sept 2016 - Oct 2016
GJC2	Pelizaeus-Merzbacher-like disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15192806, 22669416	SEIZ, HYPOTO, DERM	Sept 2016 - Oct 2016
GLA	Fabry disease	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17371887, 12519371	CM, IEM, REN, HYPOTO	Sept 2016 - Oct 2016
GLB1	Gangliosidosis GM1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	131309, 10841810, 1	IEM, REN, SK, SEIZ	Sept 2016 - Oct 2016
GLDC	Glycine encephalopathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	6336599, 3297708, 1	SEIZ	Sept 2016 - Oct 2016
GLE1	Lethal arthrogryposis with anterior horn cell disease	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	18204449, 24961629	HYPOTO	Sept 2016 - Oct 2016
GLI2	Holoprosencephaly-9	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		21940735, 17096318	SEIZ	Sept 2016 - Oct 2016
GLI3	Greig cephalopolysyndactyly syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12794692, 18000979	SK, SEIZ	Sept 2016 - Oct 2016
GLIS3	Diabetes mellitus, neonatal, with congenital hypothyroidism	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		16715098, 21139041	THYR, REN	Sept 2016 - Oct 2016
GLRA1	Hyperekplexia, hereditary 1, autosomal dominant or recessive	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16832093, 1425575,	SEIZ	Sept 2016 - Oct 2016
GLRB	Hyperekplexia 2, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11929858, 23182654	SEIZ	Sept 2016 - Oct 2016
GLUD1	Hyperinsulinism	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10871207, 9571255,	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
GLUL	Congenital brain dysgenesis due to glutamine synthetase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		16267323, 21353613	SEIZ, HYPOTO	Sept 2016 - Oct 2016
GMPPA	Congenital disorder of glycosylation	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24035193	HYPOTO	Sept 2016 - Oct 2016
GNAS	Pseudohypoparathyroidism	Definitive	IMP	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23281139, 23884777	THYR, SK	Sept 2016 - Oct 2016
GNAS	Pseudopseudohypoparathyroidism	Definitive	IMP	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23281139, 23884777	THYR, SK	Sept 2016 - Oct 2016
GNE	Inclusion body myopathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11528398, 1247378	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
GNPTAB	Mucolipidosis II	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16630736, 16200072	AN_TH, SK, SEIZ	Sept 2016 - Oct 2016
GNPTG	Mucolipidosis III gamma	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10712439, 19370764	IEM, SK, SEIZ	Sept 2016 - Oct 2016
GNS	Mucopolysaccharidosis IIId	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12573255, 17998446	IEM, SEIZ	Sept 2016 - Oct 2016
GPC3	Simpson-Golabi-Behmel syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10814714, 16010674	IEM, GLY, SK, SEIZ	Sept 2016 - Oct 2016
GPC4	Simpson-Golabi-Behmel syndrome	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21082656, 9787072,	GLY, SK, SEIZ	Sept 2016 - Oct 2016
GPC6	Omodysplasia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19481194	SK	Sept 2016 - Oct 2016
GPD1L	Brugada syndrome	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		17967972, 12676817	COND	Sept 2016 - Oct 2016
GPHN	Hyperekplexia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12684523	SEIZ	Sept 2016 - Oct 2016
GPR143	Ocular albinism, type I	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17516023, 1355560,	DERM	Sept 2016 - Oct 2016
GPR56	Polymicrogyria, bilateral frontoparietal	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15044805, 16240336	SEIZ	Sept 2016 - Oct 2016
GPR98	Usher syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19357117, 9624053,	HL, SEIZ	Sept 2016 - Oct 2016
GPSM2	Chudley-McCullough syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22578326, 22987632	HL	Sept 2016 - Oct 2016
GPX1	Hemolytic anemia due to glutathione peroxidase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		7829093	BIL	Sept 2016 - Oct 2016
GRHL2	Hearing loss	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12393799, 23813623	HL	Sept 2016 - Oct 2016
GRHPR	Hyperoxaluria, primary, type II	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2040928, 10484776,	REN	Sept 2016 - Oct 2016
GRIN2A	Epilepsy with neurodevelopmental defects	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		20384727, 23933819	SEIZ	Sept 2016 - Oct 2016
GRXCR1	Deafness, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20137778, 25802247	HL	Sept 2016 - Oct 2016
GSS	Glutathione synthetase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11445798, 15717202	AN_TH	Sept 2016 - Oct 2016
GTF2H5	Trichothiodystrophy	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	15220921, 23562818	DERM	Sept 2016 - Oct 2016
GUCY2C	Meconium ileus	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22521417, 25370039	BOW	Sept 2016 - Oct 2016
GUSB	Mucopolysaccharidosis VII	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9099834, 8644704, 1	IEM, SK, SEIZ, HYPOTO	Sept 2016 - Oct 2016
GYG1	Glycogen storage disease XV	Moderate	AR	HIGH (B)	No	C	No	Adult-onset, moderate evidence		25272951, 20357282	COND, HYPOTO	Sept 2016 - Oct 2016
GYS2	Glycogen storage disease 0	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9691087, 12072888,	GLY, SEIZ	Sept 2016 - Oct 2016
H19	Beckwith-Wiedemann Syndrome	Definitive	IMP	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15314640, 24154661	THYR	Sept 2016 - Oct 2016
HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16176262, 1835339,	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
HADH	Hyperinsulinemic hypoglycemia, familial, 4	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21252247, 11489939	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
HADHA	Mitochondrial trifunctional protein deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10518286, 9003853,	IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
HADHB	Mitochondrial trifunctional protein deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8651282, 12754706,	IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
HAMP	Haemochromatosis	Strong	AR	LOW (A)	Yes	C	No	Low penetrance	Y	12915468, 15198949	DERM	Sept 2016 - Oct 2016
HARS	Usher syndrome type 3B	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22279524	HL	Sept 2016 - Oct 2016
HARS2	Perrault syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21464306	HL, IEM, HYPOTO	Sept 2016 - Oct 2016
HAS2	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24558368	CHD	Sept 2016 - Oct 2016
HBA1	Thalassaemia alpha	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21381239, 17129226	AN_TH	Sept 2016 - Oct 2016
HBA2	Thalassemia, alpha	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21381239, 20854116	AN_TH	Sept 2016 - Oct 2016
HBB	Beta-thalassemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11283697, 20233970	AN_TH	Sept 2016 - Oct 2016
HCCS	Microphthalmia	Moderate	XLD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		16059943, 17033964	SEIZ, HYPOTO, DERM	Sept 2016 - Oct 2016
HCN4	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12676817, 11901046	COND, SEIZ	Sept 2016 - Oct 2016
HDAC8	Cornelia de Lange syndrome-like features, ocular hypertelorism & large fontanelle	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22885700, 24403048	HYPOTO	Sept 2016 - Oct 2016
HEATR2	Primary ciliary dyskinesia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23040496, 24307375	PULM	Sept 2016 - Oct 2016
HERC2	Autism spectrum disorder	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		24927284, 23065719,	23243086	Sept 2016 - Oct 2016
HESX1	Pituitary hypoplasia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	14561704, 11748154,	15841484, 11136712, 2591	Sept 2016 - Oct 2016
HEXA	Tay-Sachs disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	3156697, 8747922, 1	IEM, SEIZ	Sept 2016 - Oct 2016
HEXB	Sandhoff disease, infantile, juvenile, and adult forms	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2525553, 2973515, 2	IEM, SEIZ	Sept 2016 - Oct 2016
HFE	Hemochromatosis	Definitive	AR	LOW (A)	Yes	C	No	Low penetrance	Y	10401000, 9341868,	12241803, 8696333, 17042	Sept 2016 - Oct 2016
HFE2	Haemochromatosis	Strong	AR	LOW (A)	Yes	C	No	Low penetrance	Y	14647275, 14982873,	15461631, 14982873	Sept 2016 - Oct 2016
HGD	Alkaptonuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9154114, 10482952,	21720873, 19862842, 9529	Sept 2016 - Oct 2016
HGF	Deafness, autosomal recessive	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19576567	HL	Sept 2016 - Oct 2016
HGSNAT	Mucopolysaccharidosis IIIC	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19479962, 16960811	IEM, SEIZ	Sept 2016 - Oct 2016
HIBCH	Neurodegeneration, progressive infantile	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17160907, 25591832,	24299452, 25251209, 2616	Sept 2016 - Oct 2016
HINT1	Axonal neuropathy with neuromyotonia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22961002, 24105373	HYPOTO	Sept 2016 - Oct 2016
HK1	Hemolytic anemia due to hexokinase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12393545, 7655856,	BIL, AN_TH	Sept 2016 - Oct 2016
HLCS	Holocarboxylase synthetase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16134170, 11585745	IEM, HYPOTO	Sept 2016 - Oct 2016
HMBS	Porphyria, acute intermittent	Definitive	AD	LOW (A)	Yes	C	No	Low penetrance		12372055, 2789372,	BOW	Sept 2016 - Oct 2016
HMGCL	3-hydroxy-3-methylglutaric aciduria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	3128690, 3063529, 8	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
HNF1B	Renal cysts and diabetes syndrome	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		15930087, 22706971	BIL, CHD, REN	Sept 2016 - Oct 2016
HNF4A	Hypoglycaemia, hyperinsulinaemic	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		20164212, 23348805	GLY, SEIZ	Sept 2016 - Oct 2016
HOMEZ	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23574532	CHD	Sept 2016 - Oct 2016
HOXA1	Athabaskan brainstem dysgenesis syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16155570	CHD, SEIZ	Sept 2016 - Oct 2016
HPD	Tyrosinemia, type III	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		9343288, 7774914, 1	IEM, SEIZ	Sept 2016 - Oct 2016
HPRT1	Lesch-Nyhan syndrome 1	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1266847, 2071157, 1	REN	Sept 2016 - Oct 2016
HPS1	Hermansky-Pudlak syndrome 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10768343, 9497254,	PULM, DERM	Sept 2016 - Oct 2016
HPS3	Hermansky-Pudlak syndrome 3	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11455388, 11590544	PULM, DERM	Sept 2016 - Oct 2016
HPS4	Hermansky-Pudlak syndrome 4	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12664304, 11836498	PULM, DERM	Sept 2016 - Oct 2016
HPS5	Hermansky-Pudlak syndrome 5	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12548288, 15296495	PULM, DERM	Sept 2016 - Oct 2016
HPS6	Hermansky-Pudlak syndrome 6	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19843503, 12548288	PULM, DERM	Sept 2016 - Oct 2016
HRAS	Costello syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8882404, 16170316,	CHD, IEM, SEIZ, HYPOTO, DE	Sept 2016 - Oct 2016
HSD17B10	17-beta-hydroxysteroid dehydrogenase X deficiency	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12696021, 11102558	IEM	Sept 2016 - Oct 2016
HSD17B3	Pseudohermaphroditism, male, with gynecomastia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8075637, 8550739, 9	THYR	Sept 2016 - Oct 2016
HSD17B4	D-bifunctional protein deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16385454, 11165012	HL, SEIZ, HYPOTO	Sept 2016 - Oct 2016
HSD3B7	3 beta-hydroxysteroid dehydrogenase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12679481, 17645593	BIL	Sept 2016 - Oct 2016
HSPB8	Charcot-Marie-Tooth disease, axonal, type 2L	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15122253, 15565283	HYPOTO	Sept 2016 - Oct 2016
HSPG2	Schwartz-Jampel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16927315, 11279527	SK, HYPOTO	Sept 2016 - Oct 2016
HTRA1	CARASIL syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11889251, 19387015,	26063658, 21115960, 2450	Sept 2016 - Oct 2016
HYDIN	Primary ciliary dyskinesia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24450482, 23022101	PULM	Sept 2016 - Oct 2016
HYLS1	Hydrolethalus syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	15843405, 18648327		Sept 2016 - Oct 2016
IDS	Mucopolysaccharidosis II	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1303211, 8281149, 8	CHD, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
IDUA	Mucopolysaccharidosis Ih	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7550242, 1301941, 2	IEM, SK, SEIZ	Sept 2016 - Oct 2016
IFT122	Cranioectodermal dysplasia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20493458, 23826986	BIL, REN, SK, DERM	Sept 2016 - Oct 2016
IFT43	Cranioectodermal dysplasia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21378380, 26489029	BIL, SK, DERM	Sept 2016 - Oct 2016
IFT80	Asphyxiating thoracic dystrophy 2	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17468754, 2333910	PULM, SK	Sept 2016 - Oct 2016
IGBP1	Agenesis of the corpus callosum - intellectual deficit - coloboma - micrognathia	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	14556245		Sept 2016 - Oct 2016
IGF1	Insulin-like growth factor deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	15769976, 20668042,	14684690, 8857020, 24243	Sept 2016 - Oct 2016
IGHMBP2	Spinal muscular atrophy with respiratory distress	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14681881, 17431882	HYPOTO	Sept 2016 - Oct 2016
IGSF1	Central hypothyroidism and testicular enlargement	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23143598, 23966245	THYR	Sept 2016 - Oct 2016
IKBKAP	Dysautonomia, familial	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11179008, 22975760	HYPOTO	Sept 2016 - Oct 2016
IKBKG	Incontinentia pigmenti 1	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11590134,10756353,	AN_TH, SEIZ, DERM	Sept 2016 - Oct 2016
IL10RA	Inflammatory bowel disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19890111, 25373860	BOW	Sept 2016 - Oct 2016
IL10RB	Inflammatory bowel disease	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		25373860, 24216686	BOW	Sept 2016 - Oct 2016
IL2RG	Severe combined immunodeficiency, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15032591, 7668284,	8093460, 2896355	Sept 2016 - Oct 2016
ILDR1	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21255762, 24768815	HL	Sept 2016 - Oct 2016
ILK	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		17646580, 21252143	CM	Sept 2016 - Oct 2016
INSR	Leprechaunism	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2569023, 8105179, 8	GLY	Sept 2016 - Oct 2016
INVS	Nephronophthisis 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12872123, 9792867,	BIL, REN	Sept 2016 - Oct 2016
IQCB1	Senior-Loken syndrome 5	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15723066, 21866095	REN	Sept 2016 - Oct 2016
IRF6	Popliteal pterygium syndrome	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		19449419, 12219090	DERM	Sept 2016 - Oct 2016
IRF6	van der Woude syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19449419, 11920830	DERM	Sept 2016 - Oct 2016
IRS1	Diabetes mellitus, noninsulin dependent	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		8104271, 10206679,	14633864, 11775217, 9588	Sept 2016 - Oct 2016
ISCU	Myopathy with defiency of succinate dehydrogenase	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18304497, 19567699	IEM, HYPOTO	Sept 2016 - Oct 2016
ISL1	Diabetes, type 2	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		10969846		Sept 2016 - Oct 2016
ISPD	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22522421, 22522420	SEIZ, HYPOTO	Sept 2016 - Oct 2016
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22512483, 23114595	REN, PULM, DERM	Sept 2016 - Oct 2016

ITGA6	Epidermolysis bullosa, junctional, with pyloric stenosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	18348258, 9185503,	DERM	Sept 2016 - Oct 2016
ITGA7	Congenital muscular dystrophy with integrin deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9590299, 23800289	HYPOTO	Sept 2016 - Oct 2016
ITGB4	Epidermolysis bullosa, junctional, with pyloric atresia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18348258, 9185503,	REN, DERM	Sept 2016 - Oct 2016
IVD	Isovaleric acidemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2063866, 15486829,	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
IYD	Thyroid dyshormonogenesis	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18434651, 18765512	THYR	Sept 2016 - Oct 2016
JAG1	Alagille syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16575836, 11180599	BIL, CHD	Sept 2016 - Oct 2016
JAK3	SCID, autosomal recessive, T-negative/B-positive type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9354668, 11668610,	11668621, 10982185, 15644	Sept 2016 - Oct 2016
JPH2	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17509612, 17476457	CM, COND	Sept 2016 - Oct 2016
JUP	Arrhythmogenic right ventricular dysplasia 12	Strong	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood		17924338, 25087486	CM, COND, DERM	Sept 2016 - Oct 2016
JUP	Naxos disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2945574, 11874502,	CM, COND, DERM	Sept 2016 - Oct 2016
KANSL1	Koolen-De Vries syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22544363, 22544367	THYR, SEIZ	Sept 2016 - Oct 2016
KARS	Charcot-Marie-Tooth disease, recessive intermediate	Moderate	AR	HIGH (B)	No	C	No	Moderate evidence for gene's role in disease		20920668, 25476837	HL, HYPOTO	Sept 2016 - Oct 2016
KARS	Hearing loss	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23768514	HL, HYPOTO	Sept 2016 - Oct 2016
KAT6B	Genitopatellar syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22265014, 22265017	THYR, SEIZ	Sept 2016 - Oct 2016
KBTBD13	Nemaline myopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12805120, 21109227	HYPOTO	Sept 2016 - Oct 2016
KCNA1	Episodic ataxia type 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19307729, 9600245 ,	SEIZ	Sept 2016 - Oct 2016
KCNA5	Atrial fibrillation	Strong	AD	HIGH (A)	No	B	Yes	Moderate penetrance, actionable in childhood		16772329, 19343045	COND	Sept 2016 - Oct 2016
KCND3	Brugada syndrome	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21349352, 22840528	COND	Sept 2016 - Oct 2016
KCNE1	Long QT syndrome-5	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate evidence, actionable in childhood		14760488, 19716085	COND, HL	Sept 2016 - Oct 2016
KCNE1	Jervell and Lange-Nielsen syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14228001, 9020846,	COND	Sept 2016 - Oct 2016
KCNE1L	Atrial fibrillation	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		18313602	COND	Sept 2016 - Oct 2016
KCNE2	Long QT syndrome-6	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, actionable in childhood		14760488, 16922724	THYR, COND	Sept 2016 - Oct 2016
KCNE3	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12676817, 11901046	COND	Sept 2016 - Oct 2016
KCNH2	Long QT syndrome-2	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, actionable in childhood		14760488, 9753711,	COND, CHD, SEIZ	Sept 2016 - Oct 2016
KCNJ1	Bartter syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9002665, 10611379,	COND, REN, SEIZ	Sept 2016 - Oct 2016
KCNJ11	Hyperinsulinemic hypoglycemia, familial	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16357843, 15448107	GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
KCNJ18	Hypokalaemic periodic paralysis	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease		20074522, 21665951	HYPOTO	Sept 2016 - Oct 2016
KCNJ2	Andersen cardiodysrhythmic periodic paralysis	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16217063, 16419128	COND	Sept 2016 - Oct 2016
KCNJ5	Long QT syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20560207, 24574546	COND	Sept 2016 - Oct 2016
KCNJ8	Sudden infant death syndrom	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21215473, 23465283	COND	Sept 2016 - Oct 2016
KCNQ1	Long QT syndrome-1	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, actionable in childhood	Y	9753711, 15051636,	THYR, COND, HL	Sept 2016 - Oct 2016
KCNQ1	Jervell and Lange-Nielsen syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14228001, 9020846,	THYR, COND, HL	Sept 2016 - Oct 2016
KCNQ1OT1	Beckwith-Wiedemann syndrome	Moderate	IMP	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15372379, 21920939	THYR	Sept 2016 - Oct 2016
KCNQ2	Epilepsy, benign neonatal	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		9425895, 14534157,	SEIZ	Sept 2016 - Oct 2016
KCNQ3	Epilepsy, benign neonatal	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		9425900, 14534157,	SEIZ	Sept 2016 - Oct 2016
KCNQ4	Deafness, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10369879, 16596322	HL	Sept 2016 - Oct 2016
KCTD7	Epilepsy, progressive myoclonic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22693283, 22638565	SEIZ	Sept 2016 - Oct 2016
KDM5B	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
KDM6A	Kabuki syndrome 2	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23913813, 23076834	SEIZ	Sept 2016 - Oct 2016
KIAA1279	Goldberg-Shprintzen megacolon syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15883926, 23427148	BOW, SEIZ	Sept 2016 - Oct 2016
KIF1B	Charcot-Marie-Tooth disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		11389829, 25802885	HYPOTO	Sept 2016 - Oct 2016
KIF21A	Fibrosis of extraocular muscles, congenital	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18332320, 15827546	HYPOTO	Sept 2016 - Oct 2016
KIF22	Spondyloepimetaphyseal dysplasia with joint laxity, type 2	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22152677, 22152678	SK	Sept 2016 - Oct 2016
KIT	Piebaldism	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7529964, 15737214,	BOW	Sept 2016 - Oct 2016
KLF1	Anemia, dyserythropoietic congenital, type IV	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21055716, 24443441	AN_TH	Sept 2016 - Oct 2016
KLHL40	Nemaline myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23746549	HYPOTO	Sept 2016 - Oct 2016
KLHL41	Nemaline myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24268659	HYPOTO	Sept 2016 - Oct 2016
KMT2D	Kabuki syndrome 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23913813, 20711175	CHD, SEIZ	Sept 2016 - Oct 2016
KPTN	Macrocephaly, neurodevelopmental delay, and seizures	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24239382, 25847626	SEIZ	Sept 2016 - Oct 2016
KRAS	Noonan syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1705663, 19396835,	CHD, AN_TH, SEIZ, DERM	Sept 2016 - Oct 2016
KRT14	Epidermolysis bullosa simplex	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3372762, 17039244,	DERM	Sept 2016 - Oct 2016
KRT16	Pachyonychia congenita	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7539673, 11886499,	DERM	Sept 2016 - Oct 2016
KRT17	Pachyonychia congenita	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9008238, 9767294, 7	DERM	Sept 2016 - Oct 2016
KRT18	Cirrhosis, cryptogenic	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	12724528, 9011570,	BOW	Sept 2016 - Oct 2016
KRT5	Epidermolysis bullosa simplex	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3372762, 21375516,	DERM	Sept 2016 - Oct 2016
KRT6A	Pachyonychia congenita	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		6829608, 7545493, 1	DERM	Sept 2016 - Oct 2016
KRT6B	Pachyonychia congenita	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		9618173, 24354895,	DERM	Sept 2016 - Oct 2016
KRT8	Cirrhosis, cryptogenic	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16143128, 22419260	BOW	Sept 2016 - Oct 2016
L1CAM	X-linked hydrocephalus syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1303258, 7562969, 1	BOW, SEIZ, HYPOTO	Sept 2016 - Oct 2016
LAMA2	Muscular dystrophy, congenital merosin-deficient	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16216942, 7550355,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
LAMA3	Epidermolysis bullosa, junctional	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11810295, 16473856	PULM, DERM	Sept 2016 - Oct 2016
LAMA4	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		17646580, 26406308	CM	Sept 2016 - Oct 2016
LAMB2	Pierson syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16097004, 18672223,	20507940, 20556798, 1827	Sept 2016 - Oct 2016
LAMB3	Epidermolysis bullosa, junctional	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8824879, 7698759, 9	DERM	Sept 2016 - Oct 2016
LAMC2	Epidermolysis bullosa, junctional	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8012393, 10660342,	DERM	Sept 2016 - Oct 2016
LAMP2	Danon disease	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8504498, 17899313,	CM, IEM, HYPOTO	Sept 2016 - Oct 2016
LARGE	Walker-Warburg syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17878207, 17436019	SEIZ, HYPOTO	Sept 2016 - Oct 2016
LARS	Infantile liver failure syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22607940, 25476837	IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
LARS2	Perrault syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23541342, 26657938	HL	Sept 2016 - Oct 2016
LBR	Pelger-Huet anomaly	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12118250, 23824842	BIL, SK, SEIZ	Sept 2016 - Oct 2016
LBR	Reynolds syndrome	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		20522425	BIL, SK, SEIZ	Sept 2016 - Oct 2016
LDB3	Myofibrillar myopathy	Strong	AD	HIGH (A)	No	C	No	Adult-onset, not actionable in childhood		15668942, 21676617	CM, COND, HYPOTO	Sept 2016 - Oct 2016
LDLR	Hypercholesterolemia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3924410, 11810272,	10206683, 20809525, 94849	Sept 2016 - Oct 2016
LEPR	Obesity, morbid, due to leptin receptor deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17229951, 9537324,	THYR	Sept 2016 - Oct 2016
LGI1	Epilepsy, familial temporal lobe, 1	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		15079011, 17562837	SEIZ	Sept 2016 - Oct 2016
LHB	Hypogonadism	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		1727547, 15602022,	17761593, 22723313	Sept 2016 - Oct 2016
LHFPL5	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16459341, 16752389	HL	Sept 2016 - Oct 2016
LHX3	Pituitary hormone deficiency, combined	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22238406, 17327381	HL	Sept 2016 - Oct 2016
LIFR	Stuve-Wiedemann syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14740318, 20447141	SK	Sept 2016 - Oct 2016
LIG4	Severe combined immunodeficiency with sensitivity to ionizing radiation	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16357942, 26172957	SEIZ	Sept 2016 - Oct 2016
LIPA	Wolman syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8146180, 10562460,	BIL, IEM	Sept 2016 - Oct 2016
LITAF	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8264707, 19705173,	HYPOTO	Sept 2016 - Oct 2016
LMBRD1	Methylmalonic aciduria and homocystinuria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19136951, 21303734	THROM, IEM, SEIZ	Sept 2016 - Oct 2016
LMNA	Emery-Dreifuss muscular dystrophy 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10739764, 10939567	CM, COND, SK, HYPOTO	Sept 2016 - Oct 2016
LMNA	Dilated cardiomyopathy	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		10580077, 12920062	CM, COND, SK, HYPOTO	Sept 2016 - Oct 2016
LMNA	Charcot-Marie-Tooth disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15965218, 11799477	CM, COND, SK, HYPOTO	Sept 2016 - Oct 2016
LMNB2	Lipodystrophy, partial	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16826530, 22768673		Sept 2016 - Oct 2016
LMOD3	Nemaline myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		25250574	HYPOTO	Sept 2016 - Oct 2016
LMX1B	Nail patella syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9323941, 9618165, 1	REN	Sept 2016 - Oct 2016
LOXHD1	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19732867, 21465660	HL	Sept 2016 - Oct 2016
LPIN2	Majeed syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15994876, 17330256	DERM	Sept 2016 - Oct 2016
LPP	Tetralogy of Fallot	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20949626	CHD	Sept 2016 - Oct 2016
LRP2	Donnai-Barrow syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17632512, 18553518	CHD, REN, SEIZ	Sept 2016 - Oct 2016
LRP4	Cenani-Lenz syndactyly syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20381006, 24924585	THYR, REN, SK	Sept 2016 - Oct 2016
LRP5	Osteoporosis-pseudoglioma syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11719191, 16252235	HL, SK	Sept 2016 - Oct 2016
LRP5	Osteopetrosis, autosomal dominant	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12579474, 12054167	HL, SK	Sept 2016 - Oct 2016
LRPPRC	Leigh syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21266382, 12529507	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
LRRC6	Primary ciliary dyskinesia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23891469, 23122589	PULM	Sept 2016 - Oct 2016
LRRK2	Parkinson disease	Strong	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		17914064, 22170881,	16251215, 16157909, 1742	Sept 2016 - Oct 2016
LRSAM1	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24894446, 22781092	HYPOTO	Sept 2016 - Oct 2016
LRTOMT	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18953341, 22903915	HL	Sept 2016 - Oct 2016
LTBP4	Cutis laxa, autosomal recessive, type IC	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22829427, 19836010	PULM, BOW, DERM	Sept 2016 - Oct 2016
LUM	Amyotrophic lateral sclerosis	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21220648		Sept 2016 - Oct 2016
LYST	Chediak-Higashi syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8751864, 13465231,	DERM	Sept 2016 - Oct 2016
LYZ	Amyloidosis, systemic	Moderate	AD	HIGH (B)	No	C	No	Adult-onset, Not actionable in childhood		8464497, 17269695,	REN	Sept 2016 - Oct 2016
MAFB	Multicentric carpotarsal osteolysis syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22387013, 23956186	REN, SK	Sept 2016 - Oct 2016
MAGI2	Infantile spasms	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18565486	SEIZ	Sept 2016 - Oct 2016
MAN2B1	Mannosidosis, alpha	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22161967, 9915946,	IEM	Sept 2016 - Oct 2016
MAP2K1	Cardiofaciocutaneous syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16439621, 17366577	CHD, AN_TH, SEIZ, DERM	Sept 2016 - Oct 2016
MAP2K2	Cardiofaciocutaneous syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17366577, 19156172	CHD, AN_TH, SEIZ, DERM	Sept 2016 - Oct 2016
MAPK10	Epileptic encephalopathy	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	16249883, 23329067	SEIZ	Sept 2016 - Oct 2016
MAPT	Dementia, frontotemporal, with or without parkinsonism	Strong	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		11708988, 19884572,	8940276	Sept 2016 - Oct 2016
MARVELD2	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17186462, 23967202	HL	Sept 2016 - Oct 2016
MAT1A	Methionine adenosyltransferase deficiency	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		7573050, 7560086, 1	THROM, IEM	Sept 2016 - Oct 2016
MATN4	Multiple anomalies	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		25558065		Sept 2016 - Oct 2016
MBTPS2	Ichthyosis follicularis, alopecia & photophobia	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19361614, 23316014	DERM	Sept 2016 - Oct 2016
MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency	Definitive	AR	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		11406611, 9187484,	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency	Definitive	AR	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood	Y	11181649, 11406611	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MCEE	Methylmalonyl-CoA epimerase deficiency	Moderate	AR	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		16752391, 16697227	THROM, IEM, SEIZ	Sept 2016 - Oct 2016
MCFD2	Factor V and Factor VIII deficiency, combined	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12717434, 18391077	THROM	Sept 2016 - Oct 2016
MCOLN1	Mucolipidosis IV	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10973263, 11317355	IEM, SEIZ	Sept 2016 - Oct 2016
MCPH1	Microcephaly 1, primary, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16211557, 20978018	SEIZ	Sept 2016 - Oct 2016
MECP2	Rett syndrome	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10508514, 12655490	SEIZ	Sept 2016 - Oct 2016
MED12	Intellectual disability	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17369503, 17036352	SEIZ, HYPOTO	Sept 2016 - Oct 2016
MED13L	Transposition of great arteries	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		14638541	CHD	Sept 2016 - Oct 2016

MED20	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
MED25	Charcot-Marie-Tooth disease	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19290556, 26257172	HYPOTO	Sept 2016 - Oct 2016
MEFV	Mediterranean fever, familial	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7726228, 19863562,	BOW, REN	Sept 2016 - Oct 2016
MEGF10	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22101682, 22371254	HYPOTO	Sept 2016 - Oct 2016
MEN1	Multiple endocrine neoplasia I	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9215690, 9215689, 17623761, 12112656, 100904		Sept 2016 - Oct 2016
MESP2	Spondylocostal dysostosis, autosomal recessive 2	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15122512, 18485326	SK	Sept 2016 - Oct 2016
MFN2	Charcot-Marie-Tooth disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18458227, 9409358,	IEM, HYPOTO	Sept 2016 - Oct 2016
MFSD8	Ceroid lipofuscinosis, neuronal	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17564970, 21990111	IEM, SEIZ	Sept 2016 - Oct 2016
MGAT2	CDG syndrome type IIa	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	8808595, 11228641,	HYPOTO	Sept 2016 - Oct 2016
MGP	Keutel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9916809, 15810001,	SK	Sept 2016 - Oct 2016
MIB1	Left ventricular noncompaction	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23314057	CM	Sept 2016 - Oct 2016
MIR96	Hearing loss	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19363479, 22038834	HL	Sept 2016 - Oct 2016
MITF	Waardenburg syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8589691 , 9279758, 8	HL, DERM	Sept 2016 - Oct 2016
MKKS	Bardet-Biedl syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10973251, 11179009	REN	Sept 2016 - Oct 2016
MKS1	Meckel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	6486167, 17377820,	BIL, REN, SK	Sept 2016 - Oct 2016
MLC1	Megalencephalic leukoencephalopathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11254442, 11935341	SEIZ, HYPOTO	Sept 2016 - Oct 2016
MLH1	Lynch syndrome	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		17539898, 15872200, 19861671, 8574961, 16479		Sept 2016 - Oct 2016
MLPH	Griscelli syndrome type 3	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12897212, 21883982	DERM	Sept 2016 - Oct 2016
MLYCD	Malonyl-CoA decarboxylase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10417274, 12955715	IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MMAA	Methylmalonic aciduria, vitamin B12-responsive	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12438653, 15523652	THROM, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12471062, 16410054	THROM, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16311595, 20631720	THROM, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MMADHC	Methylmalonic aciduria and homocystinuria, cblD type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18385497, 19058814	THROM, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MOCS1	Molybdenum cofactor deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9731530, 9921896, 1	SEIZ	Sept 2016 - Oct 2016
MOCS2	Molybdenum cofactor deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12754701, 21031595	SEIZ	Sept 2016 - Oct 2016
MOGS	Glucosidase 1 deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	10788335, 26805780	SEIZ, HYPOTO	Sept 2016 - Oct 2016
MPDU1	Congenital disorder of glycosylation, type If	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11733564, 11733556	SEIZ, HYPOTO	Sept 2016 - Oct 2016
MPI	Congenital disorder of glycosylation 1b	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10980531, 15771971	GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
MPL	Amegakaryocytic thrombocytopaenia, congenital	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11133753, 16470591	AN_TH	Sept 2016 - Oct 2016
MPV17	Mitochondrial DNA depletion syndrome, hepatic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18695062, 16582910	BIL, IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
MPZ	Charcot-Marie-Tooth disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8264707, 19705173,	HYPOTO	Sept 2016 - Oct 2016
MRPS16	Mitochondrial respiratory chain disorder	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	15505824	IEM, HYPOTO	Sept 2016 - Oct 2016
MRPS22	Mitochondrial respiratory chain disorder	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17873122, 21189481	IEM, HYPOTO	Sept 2016 - Oct 2016
MSH2	Lynch syndrome	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		8261515, 17539898, 10480359, 15872200, 85749		Sept 2016 - Oct 2016
MSH6	Lynch syndrome	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		9354786, 9307272, 16479259, 12112654, 104803		Sept 2016 - Oct 2016
MSRB3	Deafness, autosomal recessive	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21185009, 24949729	HL	Sept 2016 - Oct 2016
MSX2	Parietal foramina 1	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16222674, 10767351	SEIZ	Sept 2016 - Oct 2016
MT-ND1	Leber hereditary optic neuropathy	Definitive	MITOCHO	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		1928099, 1732158, 1	HYPOTO	Sept 2016 - Oct 2016
MT-ND4	Leber hereditary optic neuropathy	Definitive	MITOCHO	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		1937476, 8457609, 1	HYPOTO	Sept 2016 - Oct 2016
MT-ND6	Leber hereditary optic neuropathy	Definitive	MITOCHO	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		1634041, 9012411, 1	HYPOTO	Sept 2016 - Oct 2016
MTHFR	Homocystinuria due to MTHFR deficiency	Definitive	AR	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood	Y	1866027, 10679944,	THROM	Sept 2016 - Oct 2016
MTM1	Myotubular myopathy, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10502779, 8544184,	HYPOTO	Sept 2016 - Oct 2016
MTO1	Hypertrophic cardiomyopathy & lactic acidosis	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23929671 , 22608499, 25552653, 25058219		Sept 2016 - Oct 2016
MTR	Methylmalonic aciduria and homocystinuria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12068375, 8968736 ,	THROM, IEM, SEIZ	Sept 2016 - Oct 2016
MTRR	Methylmalonic aciduria and homocystinuria	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12555939, 10484769	THROM, IEM, SEIZ	Sept 2016 - Oct 2016
MTTP	Abetalipoproteinaemia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10679949, 8533758, 8361539, 23043934, 77822		Sept 2016 - Oct 2016
MUC5B	Pulmonary fibrosis, idiopathic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21506741	PULM	Sept 2016 - Oct 2016
MURC	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21642240, 23299917	CM	Sept 2016 - Oct 2016
MUSK	Congenital myasthenic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15496425, 20371544	HYPOTO	Sept 2016 - Oct 2016
MUT	Methylmalonic aciduria, mut(0) type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1970180, 6132336, 2	THROM, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
MUTYH	MUTYH-associated polyposis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease (Alth	Y	15690400, 12853198, 12606733, 15931596, 1962		Sept 2016 - Oct 2016
MVK	Hyperimmunoglobulin D and periodic fever syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10369261, 11313768	DERM	Sept 2016 - Oct 2016
MYBPC1	Distal arthrogrypsis type I	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20045868, 23657818	HYPOTO	Sept 2016 - Oct 2016
MYBPC3	Cardiomyopathy, familial hypertrophic	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		7493025, 7493026, 1	CM	Sept 2016 - Oct 2016
MYBPC3	Cardiomyopathy, dilated	Conflicting	AD	UNKNOWN	Yes	C	No	Disputed evidence for gene's role in disease		20215591, 23281406	CM	Sept 2016 - Oct 2016
MYCN	Feingold syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15821734, 18470948	REN, SK	Sept 2016 - Oct 2016
MYH11	Aortic aneurysm, familial thoracic 4	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		16444274, 15998682	CHD	Sept 2016 - Oct 2016
MYH14	Deafness, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15015131, 16222661	HL	Sept 2016 - Oct 2016
MYH2	Proximal myopathy and ophthalmoplegia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20418530, 23388406	HYPOTO	Sept 2016 - Oct 2016
MYH3	Arthrogrypsis, distal	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16642020, 23401156	HYPOTO	Sept 2016 - Oct 2016
MYH6	Atrial septal defect	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		15735645, 20656787	CM, CHD	Sept 2016 - Oct 2016
MYH6	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		15998695, 20215591	CM	Sept 2016 - Oct 2016
MYH6	Cardiomyopathy, familial hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		11815426, 26656175	CM	Sept 2016 - Oct 2016
MYH7	Cardiomyopathy, familial hypertrophic	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		12351586, 23598715	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Cardiomyopathy, dilated	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		23274168, 21127202	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Laing distal myopathy	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16103042, 12975303	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Left ventricular noncompaction	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		23794396, 18506004	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Myopathy, myosin storage	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15699387, 16684601	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Scapuloperoneal syndrome, myopathic type	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17336526	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH7	Congenital fiber type disproportion	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20733148, 21288719	CM, COND, CHD, HYPOTO	Sept 2016 - Oct 2016
MYH9	Macrothrombocytopenia and progressive sensorineural deafness	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11776386, 25077172	HL, AN_TH, REN	Sept 2016 - Oct 2016
MYL2	Cardiomyopathy, familial hypertrophic, 10	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9535554, 24111713,	CM	Sept 2016 - Oct 2016
MYL3	Cardiomyopathy, familial hypertrophic, 8	Strong	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood		16267253, 23283745	CM	Sept 2016 - Oct 2016
MYLK	Aortic aneurysm, familial thoracic 7	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		21055718, 25907466	CM	Sept 2016 - Oct 2016
MYLK2	Cardiomyopathy, hypertrophic	Limited	UNKNOW	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		11733062, 24082139	CM	Sept 2016 - Oct 2016
MYO15A	Sensorineural hearing loss	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17546645, 17851452	HL	Sept 2016 - Oct 2016
MYO1C	Sensorineural hearing loss	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		25342930, 19027848	HL	Sept 2016 - Oct 2016
MYO1E	Focal segmental glomerulosclerosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21756023, 23595123	REN	Sept 2016 - Oct 2016
MYO1F	Sensorineural hearing loss	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19027848	HL	Sept 2016 - Oct 2016
MYO3A	Sensorineural hearing loss	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23967202, 23990876	HL	Sept 2016 - Oct 2016
MYO5A	Griscelli syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	9207796, 12897212,	DERM	Sept 2016 - Oct 2016
MYO6	Deafness	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12687499, 23767834	HL	Sept 2016 - Oct 2016
MYO7A	Usher syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11391666, 16679490	HL	Sept 2016 - Oct 2016
MYOM1	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21256114, 26656175	CM	Sept 2016 - Oct 2016
MYOT	Myofibrillar myopathy	Strong	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		14711882, 15111675	HYPOTO	Sept 2016 - Oct 2016
MYOZ2	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17347475, 22987565	CM	Sept 2016 - Oct 2016
MYPN	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22286171, 18006477	CM	Sept 2016 - Oct 2016
MYPN	Cardiomyopathy, hypertrophic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22286171, 18006477	CM	Sept 2016 - Oct 2016
NAA10	N-terminal acetyltransferase deficiency	Moderate	XLD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21700266, 25489052	COND	Sept 2016 - Oct 2016
NAA15	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
NAGA	N-acetylgalactosaminidase alpha deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8782044, 2243144, 1	IEM	Sept 2016 - Oct 2016
NAGLU	Sanfilippo syndrome type B	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9950362, 10094189, 9	IEM, SEIZ	Sept 2016 - Oct 2016
NAGS	N-acetylglutamate synthetase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12754705, 12594532	IEM, SEIZ	Sept 2016 - Oct 2016
NBN	Nijmegen breakage syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9590181, 9590180, 1	AN_TH	Sept 2016 - Oct 2016
NCF1	Chronic granulomatous disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8634410, 16972229,	AN_TH	Sept 2016 - Oct 2016
NCF2	Chronic granulomatous disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20167518, 10598813	BOW, AN_TH	Sept 2016 - Oct 2016
NCF4	Chronic granulomatous disease	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19692703	BOW, AN_TH	Sept 2016 - Oct 2016
NDP	Norrie disease	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10484772, 8807344,	HL, SEIZ	Sept 2016 - Oct 2016
NEB	Nemaline myopathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12899878, 15336686	HYPOTO	Sept 2016 - Oct 2016
NEBL	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	No	C	No	Moderate evidence for gene's role in disease		20951326, 23632046	CM	Sept 2016 - Oct 2016
NEDD4L	Epilepsy, photosensitive generalised	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17331106	SEIZ	Sept 2016 - Oct 2016
NEFL	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8264707, 19705173,	HYPOTO	Sept 2016 - Oct 2016
NEK1	Short rib-polydactyly syndorme, type II	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21211617, 22795106	REN, SK	Sept 2016 - Oct 2016
NEK8	Nephronophthisis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18199800, 26862157	REN	Sept 2016 - Oct 2016
NEU1	Sialidosis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11063730, 10767332	IEM, SEIZ	Sept 2016 - Oct 2016
NEUROG3	Diarrhea 4, malabsorptive, congenital	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16855267, 21378176, 21490072, 24134759		Sept 2016 - Oct 2016
NEXN	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19881492, 24503780	CM, CHD	Sept 2016 - Oct 2016
NEXN	Cardiomyopathy, familial hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		20970104, 26265630	CM, CHD	Sept 2016 - Oct 2016
NF1	Neurofibromatosis, type 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2491776, 4633999, 2	CHD, SEIZ, DERM	Sept 2016 - Oct 2016
NF2	Neurofibromatosis 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8081368, 12011146,	DERM	Sept 2016 - Oct 2016
NFATC1	Congenital heart disease	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23226213, 25205790	CHD	Sept 2016 - Oct 2016
NGLY1	Developmental delay, multifocal epilepsy & abnormal liver function	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22581936, 24651605	SEIZ	Sept 2016 - Oct 2016
NHEJ1	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16439204, 20597108	SEIZ	Sept 2016 - Oct 2016
NHLRC1	Myoclonic epilepsy of Lafora	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15781812, 12958597	SEIZ	Sept 2016 - Oct 2016
NHP2	Dyskeratosis congenita	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18005359, 9886310	DERM	Sept 2016 - Oct 2016
NIN	Seckel syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22933543	THYR	Sept 2016 - Oct 2016
NIPAL4	Ichthyosis, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15317751, 17557927	DERM	Sept 2016 - Oct 2016
NIPBL	Cornelia de Lange syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15146186, 20824775	AN_TH, SK, SEIZ	Sept 2016 - Oct 2016
NKX2-1	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18957494, 22825795	THYR, PULM, GLY	Sept 2016 - Oct 2016
NKX2-5	Congenital heart disease	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		17891520, 19073351	THYR, COND, CHD	Sept 2016 - Oct 2016

NKX3-2	Spondylo-megaepiphyseal-metaphyseal dysplasia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20004766, 22791571	SK	Sept 2016 - Oct 2016
NLGN3	Autism	Strong	COMPLEX	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		160880825, 1041729	SEIZ	Sept 2016 - Oct 2016
NLGN4X	Autism	Strong	COMPLEX	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		160880825, 1041729	SEIZ	Sept 2016 - Oct 2016
NLRP7	Hydatidiform mole	Definitive	AR	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		16462743, 17579354,	19066229, 16462743, 2165	Sept 2016 - Oct 2016
NME8	Ciliary dyskinesia, primary	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17360648	PULM	Sept 2016 - Oct 2016
NOG	Symphalangism, proximal, 1A	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10080184, 11846737,	11545688	Sept 2016 - Oct 2016
NOP10	Dyskeratosis congenita	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18005359, 9886310	DERM	Sept 2016 - Oct 2016
NOTCH1	Aortic valve disease	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease		18593716, 17662764	CHD	Sept 2016 - Oct 2016
NOTCH2	Hajdu-Cheney syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21378985, 23389697	BIL, CHD, REN, SK	Sept 2016 - Oct 2016
NOTCH3	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9818928, 9388399, 1	SEIZ	Sept 2016 - Oct 2016
NPC1	Niemann-Pick disease type C1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12555942, 12955717	BIL, IEM, SEIZ	Sept 2016 - Oct 2016
NPC2	Niemann-Pick disease type C2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11125141, 17470133	BIL, SEIZ	Sept 2016 - Oct 2016
NPHP1	Nephronophthisis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9598719, 9856524, 9	BIL, REN, SEIZ	Sept 2016 - Oct 2016
NPHP3	Nephronophthisis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19177160, 23188109	BIL, REN, SEIZ	Sept 2016 - Oct 2016
NPHP4	Nephronophthisis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15776426, 23559409	BIL, REN, SEIZ	Sept 2016 - Oct 2016
NPHS1	Congenital nephrotic syndrome, Finnish type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12495287, 11317351	REN	Sept 2016 - Oct 2016
NPPA	Atrial fibrillation	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19646991, 18614783	COND	Sept 2016 - Oct 2016
NR0B1	Congenital adrenal hypoplasia	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18762570, 9709929,	GLY, SEIZ	Sept 2016 - Oct 2016
NR1H4	Cholestasis, infantile	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		21633855, 26888176	BIL	Sept 2016 - Oct 2016
NRG1	Hirschsprung disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22574178, 23400839	BOW	Sept 2016 - Oct 2016
NRXN1	Autism	Strong	COMPLEX	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		160880825, 1041729	SEIZ	Sept 2016 - Oct 2016
NSD1	Sotos syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11896389, 12807965	THYR, GLY, SEIZ	Sept 2016 - Oct 2016
NSDHL	CHILD syndrome	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12966526, 10710235	SK, SEIZ, DERM	Sept 2016 - Oct 2016
NSDHL	CK syndrome	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21129721, 25900314	SK, SEIZ, DERM	Sept 2016 - Oct 2016
NTRK1	Congenital insensitivity to pain with anhidrosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10982191, 18077166	SEIZ, HYPOTO	Sept 2016 - Oct 2016
NTRK1	Medullary thyroid carcinoma, familial	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		10443680	SEIZ, HYPOTO	Sept 2016 - Oct 2016
NUB1	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
NUP155	Atrial fibrillation	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19070573	COND	Sept 2016 - Oct 2016
NUP62	Striatonigral degeneration, infantile	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16786527		Sept 2016 - Oct 2016
OBSL1	3-M syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21364696, 21396581	SK	Sept 2016 - Oct 2016
OCA2	Albinism, oculocutaneous	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7762554, 23504663,	DERM	Sept 2016 - Oct 2016
OCRL	Lowe oculocerebrorenal syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11146467, 9430698,	IEM, REN, SEIZ, HYPOTO, DE	Sept 2016 - Oct 2016
OFD1	Oral-facial-digital syndrome	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1941964, 17963220,	REN, SEIZ	Sept 2016 - Oct 2016
OPA1	Optic atrophy 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9006432, 17306754,	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
OPA3	3-methylglutaconic aciduria, type III	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11668429, 12126933	IEM, HYPOTO	Sept 2016 - Oct 2016
OPA3	Optic atrophy 3 with cataract	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		18496845, 15342707	IEM, HYPOTO	Sept 2016 - Oct 2016
ORC1	Meier-Gorlin syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11477602, 21358631	PULM, HYPOTO	Sept 2016 - Oct 2016
ORC4	Meier-Gorlin syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21358632, 21358631	PULM, HYPOTO	Sept 2016 - Oct 2016
ORC6	Meier-Gorlin syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21358632, 22333897	PULM, HYPOTO	Sept 2016 - Oct 2016
OSMR	Amyloidosis, primary cutaneous	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18179886, 20507362,	19690585, 19466957	Sept 2016 - Oct 2016
OSTM1	Osteopetrosis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12627228, 23685543	SK	Sept 2016 - Oct 2016
OTC	Ornithine transcarbamylase deficiency	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2012137, 9831349, 1	IEM, SEIZ	Sept 2016 - Oct 2016
OTOA	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11972037, 23173898	HL	Sept 2016 - Oct 2016
OTOF	Deafness, autosomal recessive	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12114484, 10192385	HL	Sept 2016 - Oct 2016
OTOG	Deafness, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23122587, 24378291	HL	Sept 2016 - Oct 2016
OTOGL	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23122586, 23850727	HL	Sept 2016 - Oct 2016
OTUD4	Hypogonadotropic hypogonadism, ataxia & dementia	Limited	AR-DIGEN	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23656588		Sept 2016 - Oct 2016
P2RX2	Hearing loss	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23345450, 24211385	HL	Sept 2016 - Oct 2016
PABPN1	Oculopharyngeal muscular dystrophy	Strong	AR	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		15725589, 10719989	HYPOTO	Sept 2016 - Oct 2016
PAH	Phenylketonuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9399896 , 1671852, 7	IEM	Sept 2016 - Oct 2016
PAK3	Mental retardation syndrome, X-linked	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10946356, 9731525,	SEIZ, DERM	Sept 2016 - Oct 2016
PALB2	Breast cancer	Definitive	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		17200668, 21618343	AN_TH	Sept 2016 - Oct 2016
PANK2	Neurodegeneration with brain iron accumulation 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11479594, 22221393	SEIZ	Sept 2016 - Oct 2016
PAX3	Waardenburg syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8799378, 11683776,	HL, DERM	Sept 2016 - Oct 2016
PAX6	Aniridia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16712695, 10234503	SEIZ	Sept 2016 - Oct 2016
PAX8	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15547625, 11344199	THYR, BIL, IEM	Sept 2016 - Oct 2016
PC	Pyruvate carboxylase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9585612, 9585002, 1	IEM, GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PCCA	Propionicacidemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19238581, 17051315	AN_TH, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
PCCB	Propionicacidemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2249848, 22033733,	AN_TH, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
PCDH15	Usher syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18719945, 21436283	HL	Sept 2016 - Oct 2016
PCNT	Microcephalic osteodysplastic primordial dwarfism type 2	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18174396, 21270239	SK	Sept 2016 - Oct 2016
PCSK9	Hypercholesterolemia	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		14727179, 16211558,	17316651, 2596202, 16909	Sept 2016 - Oct 2016
PDE11A	Adrenocortical hyperplasia	Moderate	AD	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease, low pentrance		16767104, 20351491,	18559625, 21047926	Sept 2016 - Oct 2016
PDE4D	Acrodysostosis 2, with or without hormone resistance	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22464252, 23033274	THYR	Sept 2016 - Oct 2016
PDHA1	Pyruvate dehydrogenase deficiency	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12163191, 9187674,	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PDHX	Pyruvate dehydrogenase complex deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16904023, 11935326	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PDLIM3	Cardiomyopathy, dilated	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		17254821	CM	Sept 2016 - Oct 2016
PDP1	Pyruvate dehydrogenase phosphatase deficiency	Limited	AR	UNKNOWN	No	C	No	Limited evidence for gene's role in disease	Y	19184109, 15855260	IEM, HYPOTO	Sept 2016 - Oct 2016
PDSS1	Deafness - encephaloneuropathy - obesity - valvulopathy Neonatal	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	17332895	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PDSS2	Leigh syndrome with nephropathy and COQ10 deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	17186472	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX1	Zellweger syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16141001, 9539740,	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX10	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21031596, 19127411	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX11B	Peroxisome biogenesis disorder	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22581968, 26233629	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX12	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21031596, 14571262	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX13	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17041890, 21031596	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX14	Zellweger syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21031596, 15146459	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX16	Zellweger syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20647552, 20681997	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX19	Zellweger syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21031596, 20683989	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX2	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14630978, 10652207	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX26	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15542397, 21031596	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX3	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10958759, 21031596	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX5	Zellweger syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18712838, 21031596	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX6	Zellweger syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19877282, 10408779	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX7	Refsum disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11781871	BIL, IEM, SK, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PEX7	Rhizomelic chondrodysplasia punctata	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9090381, 9090382, 1	BIL, IEM, SK, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PFKM	Glycogen storage disease 7	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7513946, 8659544, 8	BIL, IEM, HYPOTO	Sept 2016 - Oct 2016
PHF6	Borjeson-Forssman-Lehmann syndrome	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12415272, 15994862	SEIZ	Sept 2016 - Oct 2016
PHKA1	Phosphorylase kinase deficiency	Moderate	XLR	LOW (B)	Yes	C	No	Moderate evidence for gene's role in disease		7874115, 22238410,	GLY, HYPOTO	Sept 2016 - Oct 2016
PHKA2	Phosphorylase kinase deficiency	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21646031, 17689125	GLY, HYPOTO	Sept 2016 - Oct 2016
PHKB	Phosphorylase kinase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12825073, 9215682,	GLY, HYPOTO	Sept 2016 - Oct 2016
PHKG2	Phosphorylase kinase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9384616, 8896567, 1	GLY, HYPOTO	Sept 2016 - Oct 2016
PHOX2A	Fibrosis of extraocular muscles, congenital	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		11600883, 14597037	HYPOTO	Sept 2016 - Oct 2016
PHOX2B	Central hypoventilation syndrome	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		12640453, 16873766	PULM, BOW	Sept 2016 - Oct 2016
PHYH	Refsum disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9326939, 9326940, 1	IEM, HYPOTO	Sept 2016 - Oct 2016
PIEZO2	Arthrogryposis, distal, type 5	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23487782, 24726473	HYPOTO	Sept 2016 - Oct 2016
PIGA	Epileptic encephalopathy, early-onset	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24706016, 22305531	AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PINK1	Parkinson disease 6, early onset	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18524835, 15596610,	22644621, 15349870, 1508	Sept 2016 - Oct 2016
PITX2B	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
PITX2C	Atrial fibrillation	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		23611745, 24333117	COND, CHD	Sept 2016 - Oct 2016
PKD1	Polycystic kidney disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9005987, 1605247, 1	REN	Sept 2016 - Oct 2016
PKD2	Polycystic kidney disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8650545, 22508176,	REN	Sept 2016 - Oct 2016
PKHD1	Polycystic kidney and hepatic disease	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16133180, 19021613	BIL, REN	Sept 2016 - Oct 2016
PKLR	Pyruvate kinase deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8616073, 16704447,	BIL, AN_TH	Sept 2016 - Oct 2016
PKP2	Arrhythmogenic right ventricular dysplasia 9	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		15489853, 24125834	CM, COND, HYPOTO	Sept 2016 - Oct 2016
PLA2G6	Infantile neuroaxonal dystrophy 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10227637, 20584031	SEIZ, HYPOTO	Sept 2016 - Oct 2016
PLCE1	Nephrotic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20591883, 25349199	REN	Sept 2016 - Oct 2016
PLEC	Epidermolysis bullosa simplex	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14675180, 23289980	HYPOTO, DERM	Sept 2016 - Oct 2016
PLEC	Muscular dystrophy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21109228, 25987458	HYPOTO, DERM	Sept 2016 - Oct 2016
PLG	Plasminogen deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16849641, 10233898	THROM, REN	Sept 2016 - Oct 2016
PLN	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	No	B	Yes	Moderate evidence, Actionable in childhood		12610310, 12639993	CM, COND	Sept 2016 - Oct 2016
PLN	Cardiomyopathy, familial hypertrophic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21167350, 16829191	CM, COND	Sept 2016 - Oct 2016
PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10874315, 21699693	DERM	Sept 2016 - Oct 2016
PLOD2	Bruck syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15523624, 22689593	SK	Sept 2016 - Oct 2016
PLP1	Pelizaeus-Merzbacher disease	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1384324, 8012387, 7	SEIZ, HYPOTO	Sept 2016 - Oct 2016
PLP1	Spastic paraplegia 2, X-linked	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3840606, 7522741, 1	SEIZ, HYPOTO	Sept 2016 - Oct 2016
PMM2	Congenital disorder of glycosylation, type Ia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12409504, 10571956	GLY, SEIZ, HYPOTO	Sept 2016 - Oct 2016
PMP22	Charcot-Marie-Tooth disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8264707, 19705173,	HYPOTO	Sept 2016 - Oct 2016
PMS2	Lynch syndrome	Strong	AD	MODERATE (A)	No	C	No	Adult-onset, Not actionable in childhood		17539898, 15872200,	19861671, 8574961, 16479	Sept 2016 - Oct 2016
PNKD	Paroxysmal nonkinesiogenic dyskinesia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15262732, 19124534	DERM	Sept 2016 - Oct 2016
PNKP	Microcephaly - seizures - developmental delay	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20118933, 24965255	SEIZ	Sept 2016 - Oct 2016
PNPLA1	Ichthyosis, autosomal recessive congenital	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22246504, 24344921	DERM	Sept 2016 - Oct 2016
PNPO	Epileptic encephalopathy, neonatal	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15772097, 18024216	SEIZ	Sept 2016 - Oct 2016
PODXL	Focal and segmental glomerulosclerosis	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		24048372		Sept 2016 - Oct 2016
POLG	POLG-Related Ataxia Neuropathy Spectrum Disorders	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14694057, 12565911	BIL, BOW, IEM, SEIZ, HYPOT	Sept 2016 - Oct 2016

POLH	Xeroderma pigmentosum	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20346687, 11338401	DERM	Sept 2016 - Oct 2016
POMC	Proopiomelanocortin deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23649472, 21860632	THYR, GLY, DERM	Sept 2016 - Oct 2016
POMGNT1	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17878207, 11709191	SEIZ, HYPOTO	Sept 2016 - Oct 2016
POMGNT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18195152, 22419172	SEIZ, HYPOTO	Sept 2016 - Oct 2016
POMT1	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15792865, 1671722(	SEIZ, HYPOTO	Sept 2016 - Oct 2016
POMT1	Walker-Warburg syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16575835, 15637732	SEIZ, HYPOTO	Sept 2016 - Oct 2016
POMT2	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17923109, 17878207	SEIZ, HYPOTO	Sept 2016 - Oct 2016
POR	Disordered steroidogenesis with and without Antley-Bixler syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16467261, 19258400	SK	Sept 2016 - Oct 2016
PORCN	Focal dermal hypoplasia	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17546030, 17546031	DERM	Sept 2016 - Oct 2016
POU1F1	Pituitary hormone deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	1509263, 15928241,	THYR	Sept 2016 - Oct 2016
POU3F4	Deafness, X-linked	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7839145, 8872461, 2(	HL	Sept 2016 - Oct 2016
POU4F3	Deafness, autosomal dominant	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9506947, 18228599,	HL	Sept 2016 - Oct 2016
PPOX	Porphyria variegata	Definitive	AD	LOW (A)	Yes	C	No	Low penetrance		10486317, 8673113,	SEIZ, DERM	Sept 2016 - Oct 2016
PPT1	Neuronal ceroid lipofuscinosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9664077, 11589012,	IEM, SEIZ	Sept 2016 - Oct 2016
PQBP1	Mental retardation	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14634649, 20950397,	16493439, 24781215, 1502	Sept 2016 - Oct 2016
PRDM16	Left ventricular noncompaction	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23768516, 24387995	CM	Sept 2016 - Oct 2016
PREPL	Hypotonia - cystinuria syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16385448, 22796000	HYPOTO	Sept 2016 - Oct 2016
PRF1	Hemophagocytic lymphohistiocytosis, familial, 2	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17873118, 14757862	BIL, AN_TH, SEIZ	Sept 2016 - Oct 2016
PRICKLE1	Epilepsy, progressive myoclonic 1B	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18976727	SEIZ	Sept 2016 - Oct 2016
PRKAG2	Cardiomyopathy, hypertrophic	Strong	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood	Y	1182799, 15673802,	CM, COND	Sept 2016 - Oct 2016
PRKAG2	Wolff-Parkinson-White syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11586982, 11407343	CM, COND	Sept 2016 - Oct 2016
PRKAG2	Glycogen storage disease of heart, lethal congenital	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		6424667	CM, COND	Sept 2016 - Oct 2016
PRKAR1A	Carney complex	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18241045, 11115848	THYR, SK, DERM	Sept 2016 - Oct 2016
PRKCSH	Polycystic liver disease	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		12577059, 20095989	BIL	Sept 2016 - Oct 2016
PROC	Thrombophilia due to protein C deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10669160, 7482420,	THROM	Sept 2016 - Oct 2016
PRODH	Hyperprolinemia, type I	Moderate	AR	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		11510941, 17412540	SEIZ	Sept 2016 - Oct 2016
PROKR2	Hypogonadotropic hypogonadism	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17054399, 22773735,	20022991, 23533228, 1882	Sept 2016 - Oct 2016
PROP1	Pituitary hormone deficiency, combined, 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11549703, 9920061,	THYR, GLY	Sept 2016 - Oct 2016
PROS1	Protein S deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8943854, 9241758, 7(	THROM	Sept 2016 - Oct 2016
PRPS1	Arts syndrome	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	22546954, 17701896	HL, REN, HYPOTO	Sept 2016 - Oct 2016
PRPS1	Charcot-Marie-Tooth disease	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17701900, 24285972	HL, REN, HYPOTO	Sept 2016 - Oct 2016
PRRX1	Agnathia-otocephaly complex	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23444262, 22674740,	21294718, 22211708	Sept 2016 - Oct 2016
PRX	Charcot-Marie-Tooth disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11157804, 24011642	HYPOTO	Sept 2016 - Oct 2016
PSAP	Metachromatic leukodystrophy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10196694, 17616409	IEM, HYPOTO	Sept 2016 - Oct 2016
PSAT1	Phosphoserine aminotransferase deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17436247, 25152457	SEIZ	Sept 2016 - Oct 2016
PSEN1	Alzheimer disease, type 3	Definitive	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		7596406, 1411576, 7550356, 8634711, 11524469		Sept 2016 - Oct 2016
PSEN2	Alzheimer disease, type 4	Strong	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		7638621, 7651536 , 10631141, 14681895, 117232		Sept 2016 - Oct 2016
PTCH1	Nevoid basal cell carcinoma syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8352281, 16301862,	CHD, SEIZ, DERM	Sept 2016 - Oct 2016
PTEN	Cowden disease	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17526800, 9140396,	BOW, DERM	Sept 2016 - Oct 2016
PTEN	Bannayan-Riley-Ruvalcaba syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17526800, 12844284	BOW, DERM	Sept 2016 - Oct 2016
PTH1R	Metaphyseal chondrodysplasia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8703170, 15240651,	REN, SK	Sept 2016 - Oct 2016
PTPN11	Noonan syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12161596, 16523510	CM, CHD, AN_TH, SK, SEIZ, D	Sept 2016 - Oct 2016
PTRF	Lipodystrophy, congenital generalized, type 4	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19726876, 20684003	COND	Sept 2016 - Oct 2016
PTS	Hyperphenylalaninemia, BH4-deficient, A	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8178819, 9222757, 1(	IEM	Sept 2016 - Oct 2016
PVRL1	Cleft lip / palate	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17089422, 16674562,	11559849	Sept 2016 - Oct 2016
PYGL	Glycogen storage disease VI	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17705025, 9529348,	GLY, HYPOTO	Sept 2016 - Oct 2016
QDPR	Dihydropteridine reductase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2116088, 9744478, 1(	IEM, SEIZ	Sept 2016 - Oct 2016
RAB10	Congenital heart disease	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23665959	CHD	Sept 2016 - Oct 2016
RAB23	Carpenter syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20358613, 17503333,	21412941, 23599695, 2516	Sept 2016 - Oct 2016
RAB27A	Griscelli syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10835631, 19953648	DERM	Sept 2016 - Oct 2016
RAB3GAP1	Warburg micro syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15696165, 23420520	SEIZ	Sept 2016 - Oct 2016
RAB3GAP2	Warburg micro syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	23420520, 20967465	SEIZ	Sept 2016 - Oct 2016
RAB7A	Charcot-Marie-Tooth disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11389829, 15455439	HYPOTO	Sept 2016 - Oct 2016
RAD51B	Breast and/or ovarian cancer	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		26261251, 24139550,	27149063	Sept 2016 - Oct 2016
RAF1	Noonan syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17603482, 17603483	CM, CHD, AN_TH, SEIZ, HYP	Sept 2016 - Oct 2016
RAG1	Omenn syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11313270, 11133745	DERM	Sept 2016 - Oct 2016
RAG2	Omenn syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	25869295, 11313270	DERM	Sept 2016 - Oct 2016
RAI1	Smith-Magenis syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12652298, 15565467	IEM, SEIZ	Sept 2016 - Oct 2016
RAI1	Potocki-Lupski syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20188345, 19015223		Sept 2016 - Oct 2016
RANGRF	Brugada syndrome	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21447824, 26173111	COND	Sept 2016 - Oct 2016
RAPSN	Congenital myasthenic syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14504330, 12730725	HYPOTO	Sept 2016 - Oct 2016
RASA1	Capillary malformation-arteriovenous malformation	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14639529, 18446851	SEIZ, DERM	Sept 2016 - Oct 2016
RB1	Retinoblastoma	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2594929, 2521957, 8364580, 12541220, 1588404		Sept 2016 - Oct 2016
RBM20	Cardiomyopathy, dilated, 1DD	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		19712804, 20590677	CM	Sept 2016 - Oct 2016
RBM8A	Thrombocytopaenia-absent radius syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22366785, 17236129	AN_TH	Sept 2016 - Oct 2016
RDX	Deafness, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17226784, 19215054	HL	Sept 2016 - Oct 2016
RECQL4	Baller-Gerold syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18716613, 15964893	SK, DERM	Sept 2016 - Oct 2016
RECQL4	Rapadilino syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1481838, 12952869,	SK, DERM	Sept 2016 - Oct 2016
RECQL4	Rothmund-Thomson syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12835862, 10319867	SK, DERM	Sept 2016 - Oct 2016
RELN	Lissencephaly syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17431900, 10973257	SEIZ	Sept 2016 - Oct 2016
REN	Renal tubular dysgenesis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2359105, 22095942,	AN_TH, REN	Sept 2016 - Oct 2016
RET	Multiple endocrine neoplasia IIA	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19469690, 8632274,	PULM, BOW, REN	Sept 2016 - Oct 2016
RET	Multiple endocrine neoplasia IIB	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19469690, 8632274,	PULM, BOW, REN	Sept 2016 - Oct 2016
RFX6	Diabetes, neonatal, with intestinal atresia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20148032, 24411943	BOW	Sept 2016 - Oct 2016
RHAG	Rh-deficiency syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		8563755, 9746795, 1(	BIL, AN_TH	Sept 2016 - Oct 2016
RMRP	Cartilage-hair hypoplasia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16838329, 12107819	SK, DERM	Sept 2016 - Oct 2016
RNASEH2A	Aicardi-Goutieres syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17846997, 23592335	THYR, AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
RNASEH2B	Aicardi-Goutieres syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16845400, 17846997	THYR, AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
RNASEH2C	Aicardi-Goutieres syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16845400, 17846997	THYR, AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
ROR2	Robinow syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10932186, 17256787	SK	Sept 2016 - Oct 2016
ROR2	Brachydactyly, type B1	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10700182, 19461659	SK	Sept 2016 - Oct 2016
RPGR	Retinitis pigmentosa	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8673101, 8817343, 1(	PULM	Sept 2016 - Oct 2016
RPGRIP1L	Meckel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17558409, 17960139	REN, SK, SEIZ	Sept 2016 - Oct 2016
RPGRIP1L	Joubert syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17558407, 23351400	REN, SK, SEIZ	Sept 2016 - Oct 2016
RPL11	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10541318, 19061985	AN_TH	Sept 2016 - Oct 2016
RPL35A	Diamond-Blackfan anemia	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		16990592, 10590074	AN_TH	Sept 2016 - Oct 2016
RPL5	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19061985, 20960466	AN_TH	Sept 2016 - Oct 2016
RPS10	Diamond-Blackfan anemia	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20116044, 25946618	AN_TH	Sept 2016 - Oct 2016
RPS15	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20960466, 16990592	AN_TH	Sept 2016 - Oct 2016
RPS17	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20960466, 16990592	AN_TH	Sept 2016 - Oct 2016
RPS19	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9988267, 16990592,	AN_TH	Sept 2016 - Oct 2016
RPS24	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17186470, 19689926	AN_TH	Sept 2016 - Oct 2016
RPS26	Diamond-Blackfan anemia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20960466, 20116044	AN_TH	Sept 2016 - Oct 2016
RPS6KA3	Coffin-Lowry syndrome	Definitive	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16879200, 17100996	SEIZ	Sept 2016 - Oct 2016
RPS7	Diamond-Blackfan anemia	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20960466, 23718193	AN_TH	Sept 2016 - Oct 2016
RRM2B	Mitochondrial DNA depletion syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19667227, 17486094	BOW	Sept 2016 - Oct 2016
RS1	Retinoschisis, X linked	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9618178, 9326935, 12928282		Sept 2016 - Oct 2016
RSPH4A	Ciliary dyskinesia, primary	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19200523, 23798057	PULM	Sept 2016 - Oct 2016
RSPH9	Ciliary dyskinesia, primary	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19200523, 23993197	PULM	Sept 2016 - Oct 2016
RUNX2	Cleidocranial dysostosis	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16575894, 7747775,	SK	Sept 2016 - Oct 2016
RYR1	Central core disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20583297, 16084090	HYPOTO	Sept 2016 - Oct 2016
RYR1	Centronuclear myopathy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21062345, 20839240	HYPOTO	Sept 2016 - Oct 2016
RYR1	Malignant hyperthermia	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		12124989, 14732627	HYPOTO	Sept 2016 - Oct 2016
RYR1	Congenital fiber type disproportion	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		16084090	HYPOTO	Sept 2016 - Oct 2016
RYR1	Multiminicore disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12112081, 16380615	HYPOTO	Sept 2016 - Oct 2016
RYR2	Arrhythmogenic right ventricular dysplasia 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11159936, 25041964	CM, COND	Sept 2016 - Oct 2016
RYR2	Ventricular tachycardia, catecholaminergic polymorphic	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11157710, 17875969	CM, COND	Sept 2016 - Oct 2016
SACS	Spastic ataxia Charlevoix-Saguenay type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20876471, 18398442, 8472930		Sept 2016 - Oct 2016
SALL1	Townes-Brocks syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14755477, 16892410	REN, SK	Sept 2016 - Oct 2016
SAMHD1	Aicardi-Goutieres syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19525956, 24300241	THYR, AN_TH, SEIZ	Sept 2016 - Oct 2016
SBDS	Shwachman-Bodian-Diamond syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12496757, 21536732	AN_TH, SK	Sept 2016 - Oct 2016
SC5D	Lathosterolosis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12189593, 12812989	BIL	Sept 2016 - Oct 2016
SCN11A	Episodic pain syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		24207120, 24036948	SEIZ	Sept 2016 - Oct 2016
SCN1A	Dravet syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18930999, 24168886	SEIZ	Sept 2016 - Oct 2016
SCN1B	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18464934, 12676817	COND	Sept 2016 - Oct 2016
SCN2B	Atrial fibrillation	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		9070470, 19808477,	COND	Sept 2016 - Oct 2016
SCN3B	Brugada syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		12676817, 11901046	COND	Sept 2016 - Oct 2016
SCN4A	Hyperkalemic periodic paralysis, type 2	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1651050, 15596759	HYPOTO	Sept 2016 - Oct 2016
SCN4A	Hypokalemic periodic paralysis, type 2	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		15557532, 10599760	SEIZ	Sept 2016 - Oct 2016
SCN4B	Long QT syndrome	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		17592081, 20226894	COND	Sept 2016 - Oct 2016
SCN5A	Long QT syndrome	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9753711, 15051636,	CM, COND	Sept 2016 - Oct 2016
SCN5A	Brugada syndrome	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		12417552, 12676817	CM, COND	Sept 2016 - Oct 2016

SCNN1A	Pseudohypoaldosteronism	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16207733, 8589714,	PULM, REN	Sept 2016 - Oct 2016
SCNN1B	Pseudohypoaldosteronism	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12107247, 16207733	PULM, REN	Sept 2016 - Oct 2016
SCNN1B	Liddle syndrome	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		8524790, 21956615,	PULM, REN	Sept 2016 - Oct 2016
SCNN1G	Pseudohypoaldosteronism	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	8640238, 7550319, 1	PULM, REN	Sept 2016 - Oct 2016
SCO1	Hepatic failure, early onset, and neurologic disorder	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19295170, 11013136	IEM, HYPOTO	Sept 2016 - Oct 2016
SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12538779, 12020273	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SCP2	Leukoencephalopathy - dystonia - motor neuropathy	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16685654	SEIZ, HYPOTO	Sept 2016 - Oct 2016
SDHAF2	Hereditary Paraganglioma-Pheochromocytoma Syndromes	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence for gene's role in disease, Actionable in childhood		19628817, 20071235,	21348866, 24414418, 2224	Sept 2016 - Oct 2016
SDHB	Hereditary Paraganglioma-Pheochromocytoma Syndromes	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		18057081, 15328326,	14685938, 19454582, 1935	Sept 2016 - Oct 2016
SDHC	Hereditary Paraganglioma-Pheochromocytoma Syndromes	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		11062460, 19351833,	19454582, 16249420	Sept 2016 - Oct 2016
SDHD	Hereditary Paraganglioma-Pheochromocytoma Syndromes	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10657297, 17557926	, 15328326, 1200081, 19802	Sept 2016 - Oct 2016
SEC63	Polycystic liver disease	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		15133510, 20095989	BIL	Sept 2016 - Oct 2016
SEMA3A	Kallmann syndrome 1	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22927827, 22416012	BOW	Sept 2016 - Oct 2016
SEPN1	Muscular dystrophy, rigid spine	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10665485, 9585610,	HYPOTO	Sept 2016 - Oct 2016
SEPN1	Myopathy, congenital, with fiber-type disproportion	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16365872	HYPOTO	Sept 2016 - Oct 2016
SEPT9	Amyotrophy, hereditary neuralgic	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11523561, 19139049	HYPOTO	Sept 2016 - Oct 2016
SERPINA1	Antitrypsin alpha 1 deficiency	Definitive	AR	LOW (A)	Yes	C	No	Low penetrance	Y	18515255, 2227940,	BIL, PULM	Sept 2016 - Oct 2016
SERPINB6	Deafness, autosomal recessive	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20451170, 25719458	HL	Sept 2016 - Oct 2016
SERPINC1	Thrombophilia due to antithrombin III deficiency	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		9031473, 21264449,	THROM	Sept 2016 - Oct 2016
SERPIND1	Heparin cofactor 2 deficiency	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		8902986, 8562924, 1	THROM	Sept 2016 - Oct 2016
SETBP1	Schinzel-Giedion syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20436468, 25217958	SEIZ	Sept 2016 - Oct 2016
SETX	Ataxia-ocular apraxia 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14770181, 17159128	SEIZ	Sept 2016 - Oct 2016
SFTPA2	Pulmonary fibrosis, idiopathic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19100526, 20466729	PULM	Sept 2016 - Oct 2016
SFTPB	Surfactant metabolism dysfunction, pulmonary	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10571948, 10712351	PULM	Sept 2016 - Oct 2016
SFTPC	Interstitial lung disease	Definitive	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	18383112, 19443464	PULM	Sept 2016 - Oct 2016
SGCA	Muscular dystrophy, limb-girdle, type 2D	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9032047, 7663524, 8	HYPOTO	Sept 2016 - Oct 2016
SGCB	Muscular dystrophy, limb-girdle, type 2E	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7581449, 18285821,	HYPOTO	Sept 2016 - Oct 2016
SGCD	Cardiomyopathy, dilated	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		10974018, 24503780	CM, HYPOTO	Sept 2016 - Oct 2016
SGCD	Muscular dystrophy, limb-girdle, type 2F	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10069710, 8841194,	CM, HYPOTO	Sept 2016 - Oct 2016
SGCG	Muscular dystrophy, limb-girdle, type 2C	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7668303, 11053682,	HYPOTO	Sept 2016 - Oct 2016
SGSH	Mucopolysaccharidisis type IIIA (Sanfilippo A)	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11182930, 9401012,	IEM, SEIZ	Sept 2016 - Oct 2016
SH2D1A	Lymphoproliferative syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20926771, 10556288,	9771704, 10549287, 11049	Sept 2016 - Oct 2016
SH3BP2	Cherubism	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11381256, 10364528	SK	Sept 2016 - Oct 2016
SH3TC2	Charcot-Marie-Tooth disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21291453, 19272779	HYPOTO	Sept 2016 - Oct 2016
SHANK3	Phelan-McDermid syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17173049, 21779178	SEIZ	Sept 2016 - Oct 2016
SHH	Holoprosencephaly-3	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21940735, 8896572,	SK, SEIZ	Sept 2016 - Oct 2016
SHOC2	Noonan-like syndrome with loose anagen hair	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19684605, 22528146	SEIZ, DERM	Sept 2016 - Oct 2016
SIL1	Marinesco-Sjogren syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	14012309, 17309654	HYPOTO	Sept 2016 - Oct 2016
SIX1	Branchiootorenal syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15141091, 18330911	HL, REN	Sept 2016 - Oct 2016
SIX2	Renal hypodysplasia	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18305125, 24429398	REN	Sept 2016 - Oct 2016
SIX3	Holoprosencephaly-2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21940735, 18791198	SEIZ	Sept 2016 - Oct 2016
SIX5	Branchiootorenal syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17357085	HL, REN	Sept 2016 - Oct 2016
SKI	Shprintzen-Goldberg syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23103230, 15884042	CHD, SK, HYPOTO	Sept 2016 - Oct 2016
SLC11A2	Anemia, hypochromic microcytic	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		15459009, 16439678	AN_TH	Sept 2016 - Oct 2016
SLC12A1	Bartter syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9585600, 18391953,	REN	Sept 2016 - Oct 2016
SLC12A3	Gitelman syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8528245, 12112667,	REN	Sept 2016 - Oct 2016
SLC12A5	Febrile seizures	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24668262, 24928908	SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8292134, 12368912,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC16A1	Monocarboxylate transporter 1 deficiency	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		25390740	GLY, HYPOTO	Sept 2016 - Oct 2016
SLC16A12	Cataract, juvenile with microcornea and renal glucosuria	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18304496, 21778275	REN	Sept 2016 - Oct 2016
SLC16A2	Allan-Herndon-Dudley syndrome	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	23568789, 18398436	THYR, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC17A5	Sialic acid storage disorder, infantile	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10581036, 10947946	BIL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10391221, 10874303	AN_TH	Sept 2016 - Oct 2016
SLC19A3	Basal ganglia disease, biotin-responsive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20065143, 15871139	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC22A5	Carnitine deficiency, systemic primary	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9826541, 10072434,	IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
SLC25A12	Hypomyelination, global cerebral	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19641205, 24515575	SK, SEIZ	Sept 2016 - Oct 2016
SLC25A13	Citrullinemia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8105687, 16059747,	BIL, IEM, GLY, SEIZ	Sept 2016 - Oct 2016
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18978333, 1432421,	IEM, SEIZ	Sept 2016 - Oct 2016
SLC25A20	Carnitine-acylcarnitine translocase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8054358, 15363639,	COND, IEM, HYPOTO	Sept 2016 - Oct 2016
SLC25A22	Early myoclonic encephalopathy	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	15592994, 19780765	SEIZ	Sept 2016 - Oct 2016
SLC25A38	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19412178, 21393332	AN_TH	Sept 2016 - Oct 2016
SLC25A4	Progressive external ophthalmoplegia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10926541, 10364542	IEM, HYPOTO	Sept 2016 - Oct 2016
SLC26A2	Achondrogenesis 1B	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	7977372, 8528239, 2	SK	Sept 2016 - Oct 2016
SLC26A3	Chloride diarrhea, congenital, Finnish type	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8896562, 11524734,	REN	Sept 2016 - Oct 2016
SLC26A4	Pendred syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11317356, 23918157	THYR, HL	Sept 2016 - Oct 2016
SLC27A4	Ichthyosis prematurity syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14715877, 19631310	DERM	Sept 2016 - Oct 2016
SLC27A5	Bile acid amidation defect	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22089923	BIL	Sept 2016 - Oct 2016
SLC2A1	GLUT1 deficiency syndrome 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9462754, 10980529,	SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC2A10	Arterial tortuosity syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16550171, 17935213	HYPOTO	Sept 2016 - Oct 2016
SLC33A1	Congenital cataracts, hearing loss and low serum copper and ceruloplasmin	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22243965	HYPOTO	Sept 2016 - Oct 2016
SLC33A1	Spastic paraplegia, autosomal dominant	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19061983, 25402622	HYPOTO	Sept 2016 - Oct 2016
SLC34A2	Pulmonary alveolar microlithiasis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16960801, 17095743	PULM	Sept 2016 - Oct 2016
SLC34A3	Hypophosphatemic rickets with hypercalciuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16358214, 16358215	REN, SK	Sept 2016 - Oct 2016
SLC35A1	CDG syndrome type IIf	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	23873973, 15576474	AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC35A2	Early-onset epileptic encephalopathy	Moderate	XLD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		24115232, 23561849	HYPOTO	Sept 2016 - Oct 2016
SLC35C1	Congenital disorder of glycosylation 2c	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	11326280, 23806237	SEIZ, HYPOTO	Sept 2016 - Oct 2016
SLC35D1	Schneckenbecken dysplasia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19508970, 17952091	SK, HYPOTO	Sept 2016 - Oct 2016
SLC37A4	Glycogen storage disease Ib	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10482962, 9758626,	BOW, IEM, GLY, SEIZ, HYPOT	Sept 2016 - Oct 2016
SLC39A4	Acrodermatitis enteropathica	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12068297, 11254458	DERM	Sept 2016 - Oct 2016
SLC3A1	Cystinuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15635077, 8792820,	REN	Sept 2016 - Oct 2016
SLC41A1	Parkinson disease, idiopathic	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21812739, 20683486,	24565865	Sept 2016 - Oct 2016
SLC45A2	Oculocutaneous albinism, type IV	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11574907, 23504663	DERM	Sept 2016 - Oct 2016
SLC46A1	Folate malabsorption, hereditary	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11804211, 17446347	AN_TH, SEIZ	Sept 2016 - Oct 2016
SLC4A1	Spherocytosis	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8640229, 7530501, 1	BIL, AN_TH, REN	Sept 2016 - Oct 2016
SLC4A10	Epilepsy & mental retardation	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21204806, 18413482	SEIZ	Sept 2016 - Oct 2016
SLC4A11	Corneal endothelial dystrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17220209, 16767101	HL	Sept 2016 - Oct 2016
SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		10545938, 15930088	REN	Sept 2016 - Oct 2016
SLC5A2	Renal glucosuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12436245, 18622023	REN	Sept 2016 - Oct 2016
SLC5A5	Thyroid dyshormonogenesis 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15863666, 9171822,	THYR	Sept 2016 - Oct 2016
SLC6A19	Hartnup disorder	Definitive	AR	LOW (A)	Yes	C	No	Low penetrance		15286788, 18484095	REN, DERM	Sept 2016 - Oct 2016
SLC6A2	Orthostatic intolerance	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		10684912, 23580201	COND	Sept 2016 - Oct 2016
SLC6A5	Hyperekplexia 3	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16751771, 22700964	SEIZ	Sept 2016 - Oct 2016
SLC6A8	Creatine deficiency syndrome, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21910234, 22281021	SEIZ	Sept 2016 - Oct 2016
SLC7A7	Lysinuric protein intolerance	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10080183, 18716612	AN_TH, IEM, REN	Sept 2016 - Oct 2016
SLC7A9	Cystinuria	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12820697, 11157794	REN	Sept 2016 - Oct 2016
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		18784102, 25296721	REN	Sept 2016 - Oct 2016
SLC9A6	Christianson syndrome	Strong	XLD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21932316, 18342287	SEIZ	Sept 2016 - Oct 2016
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic	Moderate	AR-DIGEN	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22232210, 25546334	BIL	Sept 2016 - Oct 2016
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic	Moderate	AR-DIGEN	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22232210, 25546334	BIL	Sept 2016 - Oct 2016
SLCO2A1	Hypertrophic osteoarthropathy, primary, autosomal recessive 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22197487, 22553128	DERM	Sept 2016 - Oct 2016
SMAD1	Pulmonary arterial hypertension	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21898662	PULM	Sept 2016 - Oct 2016
SMAD3	Loeys-Dietz syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22167769, 21217753	DERM	Sept 2016 - Oct 2016
SMAD4	Juvenile polyposis syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9582123, 10764709,	PULM, BOW	Sept 2016 - Oct 2016
SMAD6	Cardiovascular malformation, congenital	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22275001	CHD	Sept 2016 - Oct 2016
SMAD9	Pulmonary arterial hypertension	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19211612, 21898662	PULM	Sept 2016 - Oct 2016
SMARCAL1	Schimke immunoosseous dysplasia	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11799392, 17089404	AN_TH, REN, SK	Sept 2016 - Oct 2016
SMC1A	Cornelia de Lange syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16604071, 19701948	AN_TH, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SMN1	Spinal muscular atrophy	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12815596, 8787675,	HYPOTO	Sept 2016 - Oct 2016
SMO	Medulloblastoma	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		24651015		Sept 2016 - Oct 2016
SMPD1	Niemann-Pick disease, type A	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19405096, 12369017	BIL, AN_TH, IEM, SEIZ	Sept 2016 - Oct 2016
SMPD1	Niemann-Pick disease, type B	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2023926, 19405096,	BIL, AN_TH, IEM, SEIZ	Sept 2016 - Oct 2016
SMPX	Deafness, X-linked	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21549336, 21549342	HL	Sept 2016 - Oct 2016
SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	15968592, 23231787	SEIZ, HYPOTO, DERM	Sept 2016 - Oct 2016
SNTA1	Long QT syndrome	Moderate	AD	HIGH (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		19684871, 18591664	COND	Sept 2016 - Oct 2016
SOD1	Amyotrophic lateral sclerosis	Definitive	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		8446170, 9365366, 7	647793, 7951252, 14506936	Sept 2016 - Oct 2016
SOX10	Shah-Waardenburg syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20127975, 21898658	HL, BOW, HYPOTO, DERM	Sept 2016 - Oct 2016
SOX18	Hypotrichosis-lymphedema-telangiectasia syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12740761, 24167460	DERM	Sept 2016 - Oct 2016
SOX9	Campomelic dysplasia	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7485151, 8411055, 1	SK	Sept 2016 - Oct 2016
SP110	Hepatic venoocclusive disease with immunodeficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16648851, 22621957	AN_TH	Sept 2016 - Oct 2016

SP7	Osteogenesis imperfecta, type XII	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20579626	SK	Sept 2016 - Oct 2016
SPEG	Centronuclear myopathy with dilated cardiomyopathy	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		25087613	HYPOTO	Sept 2016 - Oct 2016
SPINK5	Netherton syndrome; Netherton syndrome 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7822652, 10835624	DERM	Sept 2016 - Oct 2016
SPR	Sepiapterin reductase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11443547, 22291068	IEM, SEIZ	Sept 2016 - Oct 2016
SPRED1	Legius syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22753041, 21649642	SEIZ, DERM	Sept 2016 - Oct 2016
SPTA1	Elliptocytosis	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1679439, 1878597, 2	BIL, AN_TH	Sept 2016 - Oct 2016
SPTB	Spherocytosis	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		2961992, 1486040, 8	BIL, AN_TH	Sept 2016 - Oct 2016
SPTLC1	Neuropathy, hereditary sensory and autonomic, type IA	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19651702, 8673084	HYPOTO	Sept 2016 - Oct 2016
SPTLC2	Neuropathy, hereditary sensory and autonomic, type IC	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20920666, 26392352	HYPOTO	Sept 2016 - Oct 2016
SRCAP	Floating-Harbor syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22265015, 23621943	SEIZ	Sept 2016 - Oct 2016
ST14	Ichthyosis hypotrichosis syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		17273967, 18445049	DERM	Sept 2016 - Oct 2016
ST3GAL5	Amish infantile epilepsy syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	15502825, 22990144	SEIZ, HYPOTO	Sept 2016 - Oct 2016
STAC3	Myopathy, Native American	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23736855	HYPOTO	Sept 2016 - Oct 2016
STAR	Congenital lipoid adrenal hyperplasia,	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	8948562, 16968793,	11061515	Sept 2016 - Oct 2016
STAT3	Hyper-IgE recurrent infection syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17881745, 17676033	PULM, DERM	Sept 2016 - Oct 2016
STK11	Peutz-Jeghers syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15121768, 10874301	DERM	Sept 2016 - Oct 2016
STRA6	Microphthalmia, syndromic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17273977, 19309693	PULM, REN	Sept 2016 - Oct 2016
STRC	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22147502, 23208854	HL	Sept 2016 - Oct 2016
STS	Ichthyosis, X-linked	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7546451, 10922387,	REN, DERM	Sept 2016 - Oct 2016
STX11	Hemophagocytic lymphohistiocytosis, familial, 4	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16582076, 20486178	SEIZ	Sept 2016 - Oct 2016
STXBP1	Epileptic encephalopathy, early infantile	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20887364, 24623842	SEIZ, HYPOTO	Sept 2016 - Oct 2016
STXBP2	Hemophagocytic lymphohistiocytosis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19804848, 20798128	AN_TH	Sept 2016 - Oct 2016
SUCLA2	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with methylmalonic aciduria)	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17287286, 15877282	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SUCLG1	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20693550, 17668387	IEM, SEIZ	Sept 2016 - Oct 2016
SUOX	Sulphite oxidase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12112661, 9428520	SEIZ	Sept 2016 - Oct 2016
SURF1	Leigh syndrome, due to COX deficiency	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9837813, 9843204, 1	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
SYNE4	Hearing loss	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23348741	HL	Sept 2016 - Oct 2016
SYT14	Spinocerebellar ataxia, autosomal recessive 11	Limited	AR	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		21835308		Sept 2016 - Oct 2016
TAB2	Congenital heart disease, nonsyndromic	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		20493459, 25516202	CHD	Sept 2016 - Oct 2016
TARDBP	Amyotrophic lateral sclerosis type 10	Strong	AD	HIGH (A)	No	C	No	Adult-onset, Not actionable in childhood		22539580, 18372902,	19224587, 18396105, 1830	Sept 2016 - Oct 2016
TAT	Tyrosinemia, type II	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1356171, 1357662, 1	IEM, DERM	Sept 2016 - Oct 2016
TAZ	Barth syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15098233, 8042670,	THYR, CM, IEM, HYPOTO	Sept 2016 - Oct 2016
TBC1D24	Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20727515, 24291220	HL	Sept 2016 - Oct 2016
TBCE	Hypoparathyroidism retardation dysmorphism syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	12389028, 16938882	SK	Sept 2016 - Oct 2016
TBX1	DiGeorge syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11748311, 14585638	CHD, SK, SEIZ	Sept 2016 - Oct 2016
TBX20	Congenital heart disease	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		17668378, 18834961	CHD	Sept 2016 - Oct 2016
TBX5	Holt-Oram syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		5904863, 12818525,	CHD, SK	Sept 2016 - Oct 2016
TCAP	Cardiomyopathy, dilated	Moderate	AR	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		12507402, 9245996,	CM, HYPOTO	Sept 2016 - Oct 2016
TCAP	Muscular dystrophy, limb-girdle, type 2G	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10655062, 25724973	CM, HYPOTO	Sept 2016 - Oct 2016
TCIRG1	Osteopetrosis, infantile malignant	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22231430, 11532986	SK	Sept 2016 - Oct 2016
TCOF1	Treacher Collins syndrome 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9042910, 22317976,	HL	Sept 2016 - Oct 2016
TCTN1	Joubert syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21725307, 22693042	REN	Sept 2016 - Oct 2016
TCTN3	Joubert syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22883145, 25118024	SK	Sept 2016 - Oct 2016
TECTA	Deafness	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10987647, 21520338	HL	Sept 2016 - Oct 2016
TERC	Dyskeratosis congenita	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		18005359, 9886310,	PULM, AN_TH, DERM	Sept 2016 - Oct 2016
TERT	Dyskeratosis congenita	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		18042801, 17785587	PULM, AN_TH, DERM	Sept 2016 - Oct 2016
TFAP2A	Branchiooculofacial syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		19764023, 1456287,	REN	Sept 2016 - Oct 2016
TFAP2B	Char syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10368122, 10802654	CHD	Sept 2016 - Oct 2016
TFG	Hereditary motor and sensory neuropathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22883144, 23553329	HYPOTO	Sept 2016 - Oct 2016
TFR2	Hemochromatosis type 3	Strong	AR	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood	Y	10802645, 21524769	DERM	Sept 2016 - Oct 2016
TG	Thyroid dyshormonogenesis 3	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8626865, 16720658,	THYR	Sept 2016 - Oct 2016
TGFB1	Camurati-Engelmann disease	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		11810278, 7917133,	SK	Sept 2016 - Oct 2016
TGFB3	Arrhythmogenic right ventricular dysplasia	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		15639475, 24238504	COND	Sept 2016 - Oct 2016
TGFBR1	Loeys-Dietz syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16928994, 15731757	DERM	Sept 2016 - Oct 2016
TGFBR2	Loeys-Dietz syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15731757, 17470566	DERM	Sept 2016 - Oct 2016
TGFBR3	Premature ovarian failure	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		16613887, 22036907		Sept 2016 - Oct 2016
TGIF1	Holoprosencephaly-4	Strong	AD	MODERATE (A)	Yes	C	No	Moderate penetrance, Not actionable in childhood		21940735, 10835638	SEIZ	Sept 2016 - Oct 2016
TGM1	Ichthyosis, congenital, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19434086, 10712205	DERM	Sept 2016 - Oct 2016
TGM5	Peeling skin syndrome, acral type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22622422, 24628291	DERM	Sept 2016 - Oct 2016
TH	Tyrosine hydroxylase deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	19491146, 18058633	IEM, GLY	Sept 2016 - Oct 2016
THBD	Haemolytic uraemic syndrome	Moderate	AD	MODERATE (B)	Yes	C	No	Moderate evidence for gene's role in disease		16621965, 23314101	BIL, THROM, AN_TH, REN	Sept 2016 - Oct 2016
THBS1	Pulmonary hypertension	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22198906	PULM	Sept 2016 - Oct 2016
THRA	Hypothyroidism, congenital, nongoitrous, 6	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22168587, 24969835	THYR	Sept 2016 - Oct 2016
THRB	Thyroid hormone resistance	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7838159, 8040303, 9	THYR	Sept 2016 - Oct 2016
TIMM8A	Mohr-Tranebjaerg syndrome	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15037720, 8841189,	HL, IEM, HYPOTO	Sept 2016 - Oct 2016
TINF2	Dyskeratosis congenita	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		1866989, 18252230,	AN_TH, DERM	Sept 2016 - Oct 2016
TJP2	Hypercholanemia, familial	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		12704386, 24614073	BIL, AN_TH	Sept 2016 - Oct 2016
TK2	Mitochondrial DNA depletion syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16908738, 15907288	IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
TMC1	Deafness	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11850618, 16134132	HL	Sept 2016 - Oct 2016
TMC8	Epidermodysplasia verruciformi	Limited	AR	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		12426567, 17368633,	22158547	Sept 2016 - Oct 2016
TMEM216	Joubert syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20036350, 20512146	BIL, REN, SK, SEIZ	Sept 2016 - Oct 2016
TMEM216	Meckel syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		6486167, 17377820	BIL, REN, SK, SEIZ	Sept 2016 - Oct 2016
TMEM237	Joubert syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22152675, 22693042	REN	Sept 2016 - Oct 2016
TMEM43	Arrhythmogenic right ventricular dysplasia 5	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		18313022, 23812740	CM, COND	Sept 2016 - Oct 2016
TMEM67	Joubert syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17160906, 21068128	REN, SK, SEIZ	Sept 2016 - Oct 2016
TMEM67	Meckel syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		6486167, 17377820,	REN, SK, SEIZ	Sept 2016 - Oct 2016
TMIE	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12145746, 19438934	HL	Sept 2016 - Oct 2016
TMPO	Cardiomyopathy, dilated	Conflicting	AD	UNKNOWN	No	C	No	Disputed evidence for gene's role in disease		16247757	CM	Sept 2016 - Oct 2016
TMPRSS3	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11907649, 11137999	HL	Sept 2016 - Oct 2016
TNFRSF11B	Paget disease	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12189164, 14672344	SK	Sept 2016 - Oct 2016
TNFSF11	Osteopetrosis, autosomal recessive 2	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17632511, 23762088	CHD, SK	Sept 2016 - Oct 2016
TNNC1	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		15542288, 20215591	CM	Sept 2016 - Oct 2016
TNNI2	Distal arthrogryposis syndrome 2b	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12592607, 16924011	HYPOTO	Sept 2016 - Oct 2016
TNNI3	Cardiomyopathy, dilated	Moderate	AD	MODERATE (B)	Yes	B	Yes	Moderate evidence, Actionable in childhood		19590045, 15607392	CM	Sept 2016 - Oct 2016
TNNI3	Familial hypertrophic cardiomyopathy	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9241277, 20624503,	CM	Sept 2016 - Oct 2016
TNNT1	Nemaline myopathy, Amish type	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10952871, 24689076	HYPOTO	Sept 2016 - Oct 2016
TNNT2	Cardiomyopathy, dilated	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		20083571, 15542288	CM	Sept 2016 - Oct 2016
TNNT2	Familial hypertrophic cardiomyopathy	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		8205619, 7898523, 1	CM	Sept 2016 - Oct 2016
TNNT3	Arthyrgryposis, distal	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		25337069, 12865991	HYPOTO	Sept 2016 - Oct 2016
TNXB	Ehlers-Danlos syndrome due to tenascin X deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		9288108, 12865992,	AN_TH, DERM	Sept 2016 - Oct 2016
TP53	Li-Fraumeni syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1978757, 15695383,	15381368, 11498785, 2259	Sept 2016 - Oct 2016
TPM1	Cardiomyopathy, hypertrophic	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		9714088, 7898523, 8	CM	Sept 2016 - Oct 2016
TPM2	Nemaline myopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17846275, 11738357	HYPOTO	Sept 2016 - Oct 2016
TPM2	Arthrogrypsosis multiplex congenita, distal	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		17339586, 12592607	HYPOTO	Sept 2016 - Oct 2016
TPM3	Nemaline myopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12196661, 17376686	HYPOTO	Sept 2016 - Oct 2016
TPM3	Congenital fiber-type disproportion myopathy	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15857933, 18300303	HYPOTO	Sept 2016 - Oct 2016
TPO	Thyroid dyshormonogenesis 2A	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9024270, 8964831, 1	THYR	Sept 2016 - Oct 2016
TPP1	Neuronal ceroid lipofuscinosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10330339, 12414822	IEM, SEIZ	Sept 2016 - Oct 2016
TPRN	Deafness, autosomal recessive	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20170898, 20170899	HL	Sept 2016 - Oct 2016
TRAPPC2	Spondyloepiphyseal dysplasia tarda	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11349230, 10431248	SK	Sept 2016 - Oct 2016
TRDN	Catecholaminergic polymorphic ventricular tachycardia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22422768, 24025405	CM, COND	Sept 2016 - Oct 2016
TREX1	Aicardi-Goutieres syndrome 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16845398, 20799324	THYR, AN_TH, HYPOTO	Sept 2016 - Oct 2016
TRH	Thyrotropin-releasing hormone deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		3938792, 6432893	THYR	Sept 2016 - Oct 2016
TRHR	Thyrotropin-releasing hormone resistance, generalized	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		9141550, 26735259	THYR	Sept 2016 - Oct 2016
TRIM32	Muscular dystrophy, limb-girdle, type 2H	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		11822024, 17994549	REN, HYPOTO	Sept 2016 - Oct 2016
TRIM37	Mulibrey nanism syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	10888877, 15108285,	17100991, 23385855, 1275	Sept 2016 - Oct 2016
TRIOBP	Deafness, autosomal recessive	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16385458, 16385457	HL	Sept 2016 - Oct 2016
TRIP11	Achondrogenesis type 1A	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20089971, 23956106	SK	Sept 2016 - Oct 2016
TRMU	Liver failure, transient infantile	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21169334, 21680270	BIL, IEM, GLY, HYPOTO	Sept 2016 - Oct 2016
TRPM2	ALS and Parkinson's disease	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		19004782		Sept 2016 - Oct 2016
TRPM4	Cardiac conduction disease	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21887725, 20562447	COND	Sept 2016 - Oct 2016
TSC1	Tuberous sclerosis 1	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9242607, 9328481, 1	PULM, REN, SEIZ	Sept 2016 - Oct 2016
TSC2	Tuberous sclerosis 2	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8269512, 17120248,	PULM, REN, SEIZ	Sept 2016 - Oct 2016
TSEN54	Pontocerebellar hypoplasia type 4	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18711368, 20952379	SEIZ, HYPOTO	Sept 2016 - Oct 2016
TSFM	Combined oxidative phosphorylation deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	17033963, 22499341	IEM	Sept 2016 - Oct 2016
TSHB	Hypothyroidism, congenital, nongoitrous 4	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9589689, 15292359,	THYR	Sept 2016 - Oct 2016
TSHR	Hypothyroidism	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12050212, 20718767	THYR	Sept 2016 - Oct 2016
TSPEAR	Sensorineural deafness	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22678063, 26969326	HL	Sept 2016 - Oct 2016
TSPYL1	Sudden infant death with dysgenesis of the testes syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	15273283		Sept 2016 - Oct 2016
TTC21B	Bardet-Biedl syndrome	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		21258341, 22773737	REN	Sept 2016 - Oct 2016
TTC37	Trichohepatoenteric syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20176027, 21120949	BOW	Sept 2016 - Oct 2016
TTC7A	Immunodeficiency, combined, with intestinal atresias	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23830146, 24417819	BOW	Sept 2016 - Oct 2016
TTN	Centronuclear myopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	23975875, 24395473	CM, HYPOTO	Sept 2016 - Oct 2016

TTN	Cardiomyopathy, dilated	Definitive	AD	MODERATE (A)	No	B	Yes	Moderate penetrance, Actionable in childhood		22335739, 24119082	CM, HYPOTO	Sept 2016 - Oct 2016
TTPA	Ataxia with isolated vitamin E deficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9463307, 7719340, 8	602747, 10360777, 9270601	Sept 2016 - Oct 2016
TTR	Amyloidosis, hereditary, transthyretin-related	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		1626556, 1301926, 9	CM	Sept 2016 - Oct 2016
TUBA8	Polymicrogyria with optic nerve hypoplasia	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19896110	SEIZ	Sept 2016 - Oct 2016
TWIST1	Saethre-Chotzen syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9259286, 9585583,	SK	Sept 2016 - Oct 2016
TYMP	Mitochondrial DNA depletion syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		12177387, 9924029,	BOW	Sept 2016 - Oct 2016
TYR	Albinism, oculocutaneous 1	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		13680365, 23504663	DERM	Sept 2016 - Oct 2016
UBA1	Spinal muscular atrophy, X-linked infantile	Moderate	XLR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	18179898, 23518311	HYPOTO	Sept 2016 - Oct 2016
UBR1	Johanson-Blizzard syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16311597, 24599544	THYR	Sept 2016 - Oct 2016
UCP2	Hyperinsulinism	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		19065272, 23275527	GLY, SEIZ	Sept 2016 - Oct 2016
UGT1A1	Crigler-Najjar syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		7989595, 19830808,	BIL	Sept 2016 - Oct 2016
UGT1A4	Crigler-Najjar syndrome	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		8280139	BIL	Sept 2016 - Oct 2016
UGT1A5	UDP glucuronosyltransferase deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16786511	BIL	Sept 2016 - Oct 2016
UMOD	Nephropathy	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		14569098, 21868615,	12471200, 12629136, 1946	Sept 2016 - Oct 2016
UNC13D	Hemophagocytic lymphohistiocytosis, familial, 3	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16528436, 16278825	SEIZ	Sept 2016 - Oct 2016
UQCRB	Mitochondrial complex III deficiency	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	12709789, 25446085	IEM, HYPOTO	Sept 2016 - Oct 2016
UQCRQ	Mitochondrial complex III deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	18439546	IEM, HYPOTO	Sept 2016 - Oct 2016
UROD	Porphyria, hepatoerythropoietic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		3821794, 1905636, 1	DERM	Sept 2016 - Oct 2016
UROS	Porphyria, congenital erythropoietic	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9092747, 1737856, 7	AN_TH, DERM	Sept 2016 - Oct 2016
USH1C	Usher syndrome 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	11139240, 10973248	HL	Sept 2016 - Oct 2016
USH1G	Usher syndrome 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	12588794, 16283141	HL	Sept 2016 - Oct 2016
USH2A	Usher syndrome 2	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	16963483, 22135276	HL	Sept 2016 - Oct 2016
VAMP1	Spastic ataxia	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		22958904	HYPOTO	Sept 2016 - Oct 2016
VCAN	Wagner syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		22739342, 21738396	CHD	Sept 2016 - Oct 2016
VCL	Cardiomyopathy, dilated	Strong	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		11815424, 24503780	CM	Sept 2016 - Oct 2016
VCP	Inclusion body myopathy with early-onset paget disease and frontotemporal dementia	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		16247064, 21145000	HYPOTO	Sept 2016 - Oct 2016
VDR	Vitamin D-dependent rickets	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	2849209, 2557627, 8	796143, 15190891, 9005998	Sept 2016 - Oct 2016
VHL	von Hippel-Lindau syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8758207, 8493574, 2	AN_TH	Sept 2016 - Oct 2016
VIPAS39	Arthrogryposis, renal dysfunction and cholestasis	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20190753, 22753090	BIL, REN, HYPOTO, DERM	Sept 2016 - Oct 2016
VLDLR	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	22973972, 16080122	SEIZ	Sept 2016 - Oct 2016
VPS13A	Choreoacanthocytosis	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		15918062, 12404112	SEIZ, HYPOTO	Sept 2016 - Oct 2016
VPS13B	Cohen syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	18655112, 11477603	SEIZ	Sept 2016 - Oct 2016
VPS33B	Arthrogryposis renal dysfunction cholestasis syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15052268, 16896922	BIL, REN	Sept 2016 - Oct 2016
VPS53	Progressive cerebello-cerebral atrophy	Limited	AR	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		24577744		Sept 2016 - Oct 2016
VSX1	Keratoconus	Moderate	AD	MODERATE (B)	No	C	No	Moderate evidence for gene's role in disease		11978762, 19956409,	25963163, 21976959, 1862	Sept 2016 - Oct 2016
VWF	von Willebrand disease	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood		7468655, 20409624,	AN_TH	Sept 2016 - Oct 2016
WAS	Wiskott-Aldrich syndrome	Definitive	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	17250667, 9445409,	AN_TH	Sept 2016 - Oct 2016
WDR19	Nephronophthisis	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		23559409, 25726036	BIL, REN, SK	Sept 2016 - Oct 2016
WDR35	Cranioectodermal dysplasia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		20817137, 22486404	BIL, RE, SK, DERM	Sept 2016 - Oct 2016
WDR36	Glaucoma	Limited	AD	UNKNOWN	No	C	No	Limited evidence for gene's role in disease		15677485, 18172102,	16723468, 24825108	Sept 2016 - Oct 2016
WDR62	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20890279, 20729831	SEIZ	Sept 2016 - Oct 2016
WFS1	Wolfram syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		10521293, 21446023	HL, IEM, SEIZ, HYPOTO	Sept 2016 - Oct 2016
WNK1	Neuropathy, hereditary sensory and autonomic, type I	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16534117, 15911806	REN, HYPOTO	Sept 2016 - Oct 2016
WNT10A	Ectodermal dysplasia	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20979233, 24043634,	17847007	Sept 2016 - Oct 2016
WNT3	Tetra-amelia, autosomal recessive	Limited	AR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease	Y	14872406	SK	Sept 2016 - Oct 2016
WNT5A	Robinow syndrome	Moderate	AD	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		19918918, 24716670	SK	Sept 2016 - Oct 2016
WNT7A	Ulna and fibula absence of with severe limb deficiency	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease	Y	16826533, 21271649,	23266637, 20949531	Sept 2016 - Oct 2016
WRAP53	Dyskeratosis congenita	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		18005359, 9886310	DERM	Sept 2016 - Oct 2016
WRN	Werner syndrome	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9048918, 9288107, 9	DERM	Sept 2016 - Oct 2016
WT1	Wilms tumor, type 1	Definitive	AD	MODERATE (A)	Yes	B	Yes	Moderate penetrance, Actionable in childhood	Y	10475544, 2848758,	REN	Sept 2016 - Oct 2016
WT1	Denys-Drash syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9607189, 2172500, 1	REN	Sept 2016 - Oct 2016
WT1	Frasier syndrome	Definitive	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		9475094, 9607189, 9	REN	Sept 2016 - Oct 2016
XPA	Xeroderma pigmentosum	Definitive	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	20346687, 11338401	DERM	Sept 2016 - Oct 2016
XPC	Xeroderma pigmentosum	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		20346687, 11338401	DERM	Sept 2016 - Oct 2016
YARS2	Myopathy, lactic acidosis, and sideroblastic anemia	Moderate	AR	HIGH (B)	Yes	C	No	Moderate evidence for gene's role in disease		22504945, 24344687	AN_TH, IEM, HYPOTO	Sept 2016 - Oct 2016
ZAP70	ZAP70-related severe combined immunodeficiency	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		8124727, 8202712, 8	AN_TH	Sept 2016 - Oct 2016
ZEB2	Mowat-Wilson syndrome	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		23466526, 19215041	SEIZ	Sept 2016 - Oct 2016
ZFPM2	Tetralogy of Fallot	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		14517948, 20807224,	24469719, 25025186	Sept 2016 - Oct 2016
ZIC2	Holoprosencephaly-5	Strong	AD	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease		21940735, 19177455	SEIZ	Sept 2016 - Oct 2016
ZIC3	Heterotaxy	Strong	XLR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	9354794, 14681828,	CHD	Sept 2016 - Oct 2016
ZMPSTE24	Restrictive dermopathy	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	15843403, 16297189	SK, HYPOTO	Sept 2016 - Oct 2016
ZNF252P	Hypothyroidism	Limited	AD	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		23295299	THYR	Sept 2016 - Oct 2016
ZNF469	Brittle cornea syndrome	Strong	AR	HIGH (A)	Yes	A	Yes	Strong evidence for highly penetrant childhood-onset disease	Y	21664999, 23680354,	18452888, 19661234	Sept 2016 - Oct 2016
ZNF674	Mental retardation	Limited	XLR	UNKNOWN	Yes	C	No	Limited evidence for gene's role in disease		16385466, 23871722		Sept 2016 - Oct 2016

AR, autosomal recessive; AD, autosomal dominant; XLR, X-linked recessive;XLR, X-linked dominant; yrs; years; BabySeq gene panels: AN_TH, anemia-thrombocytopenia; BIL, hyperbilirubinemia; BOW, bowel dysfunction; CHD, congenital heart disease; CM, cardiomyopathy; COND, cardiac conduction disease; DERM, dermatologic disease; GLY, hypoglycinemia; HL, hearing loss; HYPOTO, hypotonia; IEM, inborn errors of metabolism; REN, renal disorder; RESP, respiratory disorder; SEIZ, seizure; SK, skeletal dysplasia; THROM, thrombophilia; THYR, hypothyroidism.

Example genes in category A

	Gene	Disease	Gene	Disease
Genes associated with diseases that present at birth	CHD7	Charge syndrome	MYH3	Arthrogryposis, distal
	EDA	Ectodermal dysplasia, hypohidrotic	NIPBL	Cornelia de Lange syndrome
	FGFR3	Achondroplasia	PEX1	Zellweger syndrome
	HRAS	Costello syndrome	SHH	Holoprosencephaly
	MTM1	Myotubular myopathy	TBX5	Holt-Oram syndrome
Genes associated with diseases that are currently tested in conventional NBS and are likely to benefit from early treatment	BTD	Biotinidase deficiency	GALT	Galactosemia
	CBS	Homocystinuria	GJB2	Hearing loss
	CFTR	Cystic fibrosis	IVD	Isovaleric acidemia
	FAH	Tyrosinemia type I	PAH	Phenylketonuria
	GAA	Glycogen storage disease type II (Pompe disease)	OTC	Ornithine transcarbamylase deficiency
Genes associated with diseases that may benefit from early treatment but currently are not tested in conventional NBS	ABCD1	X-linked adrenoleukodystrophy	GALC	Krabbe disease
	ATP7B	Wilson disease	GBA	Gaucher disease
	DHCR7	Smith-Lemli-Opitz syndrome	IDUA	Mucopolysaccharidosis I
	DMD	Duchenne muscular dystrophy	MPI	Congenital disorder of glycosylation type Ib
	ELANE	Congenital neutropenia	ZAP70	Severe combined immunodeficiency
Genes associated with diseases that present during childhood and currently lack effective treatment opportunities	APTX	Ataxia with oculomotor apraxia and hypoalbuminemia	MECP2	Rett syndrome
	COL7A1	Epidermolysis bullosa dystrophica	NEB	Nemaline myopathy
	ERCC6	Cockayne syndrome	POMT1	Walker-Warburg syndrome
	IKBKAP	Familial dysautonomia	RAI1	Smith Magenis syndrome
	LMNA	Emery-Dreifuss muscular dystrophy	TPP1	Neuronal ceroid lipofuscinosis

NBS, newborn screening.