Gene	Gene MIM	Phenotype	Phenotype MIM	Inheritance	Outcome Considered	Age of Onset	Intervention Considered	Age of Intervention Implementation	Severity	Likelihood	Efficacy	Acceptability	Knowledge	Total	CATEGORY	RUSP Condition	BabySeq Category
45, X	N/A	Turner syndrome	N/A	Multiple and/or complex pattern	Heart disease	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	3	3	3	13	**1**		
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, 3	610921	AR	Respiratory distress / failure	Birth	Referral to specialists for surveillance and early management	Neonatal	3	3	1	1	3	11	**2**		A
ABCC8	600509	Hyperinsulinemic hypoglycemia, familial 1	256450	AR	Hyperinsulinemic hypoglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	3	14	**1**		A
ABCC8	600509	Diabetes mellitus, transient and permanent neonatal	125853	AD	Hyperglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	3	3	2	12	**1**		
ABCC9	601439	Cantu Syndrome / Cardiomyopathy, dilated, 1O	608569	AD	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	2	3	1	3	2	11	**1**		B
ABCD1	300371	Adrenoleukodystrophy	300100	X-linked	Neurodegeneration	Childhood	Referral to specialists for surveillance and early management	Neonatal	2	2	2	1	2	9	**1**		A
ABCG5	605459	Sitosterolemia	210250	AR	Coronary artery disease	Childhood	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	2	11	**1**		A
ACADM	607008	Acyl-CoA Dehydrogenase, Medium Chain, Deficiency of	201450	AR	Hypoglycemia - profound	Infant	Referral to specialists for surveillance and dietary management	Neonatal	3	3	3	3	3	15	**1**	Core	A
ACADS	606885	Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency - Infantile	201470	AR	Hypoglycemia	Variable	Referral to specialists for surveillance and dietary management	Neonatal	1	0	3	3	0	7	**2**	Secondary	C
ACADSB	600301	2-Methylbutyrylglycinuria	610006	AR	Hypoglycemia - profound	Infant	Referral to specialists for surveillance and dietary management	Infant	2	0	3	3	1	9	**2**		C
ACADVL	609575	VLCAD deficiency	201475	AR	Hypoglycemic crises	Variable	Referral to specialists for surveillance and dietary management	Neonatal	3	3	3	3	3	15	**1**	Core	A
ACAT1	607809	Alpha-Methylacetoacetic Aciduria	203750	AR	Severe metabolic acidosis	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	3	2	13	**1**	Core	A
ACTA2	102620	Familial Thoracic Aortic Aneurysms	611788	AD	Aortic dissection	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	2	3	2	12	**1**		B
ACTC1	102540	Cardiomyopathy (Hypertrophic 11, dilated, 1R / Left Ventricular Noncompaction 4)	612098	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		B
ACTG1	102560	Baraitser-Winter syndrome 2	614583	AD	Intellectual disability and developmental delay	Infant	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	1	6	**2**		A
ACTG1	102560	Deafness, autosomal dominant 20/26	604717	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	2	12	**1**		A
ACTN1	102575	Bleeding disorder, platelet-type, 15	615193	AD	Bleeding	Birth	Referral to specialists for surveillance and supportive care	N / A	0	2	0	0	0	2	**2**		A
ACTN2	102573	Cardiomyopathy, Familial Hypertrophic, 23, w/ or w/o LVNC / dilated 1AA w/ or w/o LVNC	612158	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	2	2	3	2	11	**1**		B
ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2	600376	AD	Syndromic manifestations	Adolescent-Adulthood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
ADA	608958	Severe Combined Immunodeficiency due to ADA Deficiency	102700	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	3	14	**1**		A
ADCK3	606980	Coenzyme Q10 deficiency, primary, 4	612016	AR	Neurologic involvement	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	1	3	1	9	**2**		
ADCY1	103072	?Deafness, autosomal recessive 44	610154	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Birth	1	3	2	3	0	9	**2**		
ADGRV1	602851	Usher Syndrome, type 2C	605472	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	3	12	**1**		
ADK	102750	Hypermethioninemia due to adenosine kinase deficiency	614300	AR	Hyperinsulinism	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	1	10	**1**	Secondary	A
AGA	613228	Aspartylglucosaminuria	208400	AR	Neurodegeneration	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	1	1	2	9	**2**		A
AGL	610860	Glycogen Storage Disease III	232400	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	2	3	3	12	**1**		A
AHCY	180960	Hypermethioninemia with deficiency of S-Adenosylhomocysteine Hydrolase	613752	AR	Intellectual disability and psychomotor delay	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	3	0	10	**2**	Secondary	
AIFM1	300169	Deafness, X-linked 5 / Cowchock Syndrome	300614	X-linked	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	1	3	1	9	**2**		
AIP	605555	Pituitary adenoma, growth hormone-secreting / Pituitary adenoma, ACTH-secreting / Pituitary adenoma, prolactin-secreting	102200, 219090, 600634	AD	Pituitary adenoma	Variable	Referral to specialists for surveillance and/or preemptive management	Childhood	1	2	3	3	3	12	**1**		B
AK2	103020	Reticular Dysgenesis	267500	AR	Immunodeficiency - combined	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	1	2	2	11	**1**		
AKAP9	604001	Long QT Syndrome 11	611820	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Childhood	3	0	2	2	0	7	**2**		C
ALB	103600	Analbuminemia	616000	AR	Mild edema, hypotension, and fatigue	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	2	3	1	9	**2**		A
ALB	103600	Familial dysalbuminemic hyperthyroxinemia	615999	AD	Dysalbuminemic hyperthyroxinemia	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	1	3	3	2	10	**1**		
ALDH3A2	609523	Sjogren-Larsson Syndrome	270200	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	Infant	1	3	0	0	2	6	**2**		A
ALDH7A1	107323	Pyridoxine-dependent epilepsy	266100	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and dietary management	Neonatal	1	3	2	3	3	12	**1**		
ALDOB	612724	Fructose intolerance	229600	AR	Fructose intolerance manifestations	Infant	Referral to specialists for surveillance and dietary management	Infant	2	3	3	3	3	14	**1**		A
ALMS1	606844	Alstrom Syndrome	203800	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	1	3	3	12	**1**		A
ANK2	106410	Long QT Syndrome 4	600919	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	2	2	2	10	**1**		B
ANKRD1	609599	Cardiomyopathy	N/A	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Childhood	2	3	2	3	1	11	**2**		B
ANO6	608663	Scott syndrome	262890	AR	Bleeding and hemorrhagic diathesis	Adolescent	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	0	10	**2**		
AP3B1	603401	Hermansky-Pudlak Syndrome 2	608233	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Neonatal	0	3	3	3	1	10	**2**		A
APC	611731	Familial Adenomatous Polyposis	175100	AD	Hereditary cancer predisposition	Adolescent	Referral to specialists for surveillance and/or preemptive management	Childhood	2	3	3	2	3	13	**1**		A
APOB	107730	Familial hypercholesterolemia due to ligand-defective APOB	144010	AD	Hypercholesterolemia and cardiovascular disease	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
APOL1	603743	{Susceptibility to end-stage renal disease, nondiabetic / Susceptibility to glomerulosclerosis, focal segmental, 4}	612551	AR	Kidney disease	Variable	Referral to specialists for surveillance and early management	Variable	1	1	1	3	2	8	**2**		
APTX	606350	Coenzyme Q10 deficiency, secondary	612016	AR	Neurologic involvement	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	1	3	1	9	**2**		A
ARG1	608313	Argininemia	207800	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	2	2	2	10	**1**	Secondary	A
ARHGEF6	300267	Mental Retardation, X-linked 46	300436	X-linked	Intellectual disability	Childhood	Referral to specialists for surveillance and supportive care	Neonatal	1	3	0	0	1	5	**2**		
ARMC4	615408	Ciliary dyskinesia, primary, 23	615451	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		A
ARSA	607574	Metachromatic Leukodystrophy	250100	AR	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Variable	2	3	1	1	2	9	**2**		A
ARSB	611542	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	253200	AR	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
ASAH1	613468	Spinal Muscular Atrophy with Progressive Myoclonic Epilepsy	159950	AR	Muscle weakness	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	1	6	**2**		
ASCL1	100790	Central hypoventilation syndrome / Haddad syndrome	209880	AD	Hypoventilation	Birth	Referral to specialists for surveillance and early management	Neonatal	3	0	3	2	0	8	**2**		C
ASL	608310	Argininosuccinic aciduria	207900	AR	Hyperammonemic crisis and chronic liver disease	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	3	12	**1**	Core	A

ASS1	603570	Citrullinemia	215700	AR	Hyperammonemic crisis	Infant	Referral to specialists for surveillance and dietary management	Neonatal	3	3	2	2	2	12	**1**	Core	A
ATP6V1B1	192132	Renal tubular acidosis with deafness	267300	AR	Renal tubular acidosis	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	2	12	**1**		A
ATP7A	300011	Menkes Disease	309400	X-linked	Neurologic involvement	Birth	Referral to specialists for surveillance and early management	Neonatal	3	3	1	3	2	12	**1**		A
ATP7B	606882	Wilson Disease	277900	AR	Liver cirrhosis	Variable	Referral to specialists for surveillance and dietary management	Neonatal	2	2	2	3	3	12	**1**		A
BAG3	603883	Cardiomyopathy (Dilated, 1HH)	613881	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		B
BCHE	177400	Increased sensitivity to choline ester anesthesia	N/A	AR	Prolonged apnea	Variable	Referral to specialists for surveillance and early management	Neonatal	0	0	3	3	3	9	**2**		
BCKDHA	608348	Maple syrup urine disease, type Ia, Ib, and type II	248600	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	3	12	**1**	Core	A
BCKDHB	248611	Maple syrup urine disease, type Ib	248600	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	3	12	**1**	Core	A
BCS1L	603647	Leigh syndrome / GRACILE syndrome / Mitochondrial complex III deficiency, nuclear type 1	256000	AR	Mitochondrial manifestations and lactic acidosis	Birth	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		A
BDNF	113505	Central hypoventilation syndrome	209880	AD	Hypoventilation	Birth	Referral to specialists for surveillance and early management	Neonatal	3	0	3	2	0	8	**2**		C
BLK	191305	MODY, type XI (MODY11)	613375	AD	Diabetes mellitus	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	3	0	0	1	5	**4**		
BLM	604610	Bloom syndrome	210900	AR	Hereditary cancer predisposition	Childhood	Referral to specialists for surveillance and/or preemptive management	Infant	2	3	1	1	2	9	**2**		A
BLOC1S3	609762	Hermansky-Pudlak Syndrome 8	614077	AR	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	0	9	**2**		C
BLOC1S6	604310	Hermansky-Pudlak syndrome, 9	614171	AR	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	0	9	**2**		C
BMPR1A	601299	Polyposis, juvenile intestinal	174900	AD	Gastrointestinal cancer	Variable	Referral to specialists for surveillance and/or preemptive management	Infant	2	3	2	2	2	11	**1**		A
BMPR2	600799	Pulmonary hypertension, familial primary, 1, with or without HHT / fenfluramine or dexfenfluramine-associated / Pulmonary venoocclusive disease 1	178600 / 265450	AD	Pulmonary hypertension	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	1	2	3	10	**1**		B
BRCA1	113705	Hereditary Breast and Ovarian Cancer	604370	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	2	3	3	13	**3**		C
BRCA2	600185	Fanconi anemia, complementation group D1	605724	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
BRCA2	600185	Hereditary Breast and Ovarian Cancer	612555	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	2	3	3	13	**3**		C
BRIP1	605882	Fanconi Anemia complementation group J	609054	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	1	10	**1**		
BRIP1	605882	Breast Cancer, early-onset	114480	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	2	2	3	2	11	**3**		
BSND	606412	Bartter syndrome type 4A (recessive)	602522	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
BTD	609019	Biotinidase Deficiency	253260	AR	Developmental delay	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**	Core	A
C21ORF59	615494	Ciliary dyskinesia, primary, 26	615500	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
CACNA1C	114205	Arrhythmia (Brugada Syndrome 3, Timothy syndrome)	601005	AD	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	2	11	**1**		B
CACNA1D	114206	Sinoatrial Node Dysfunction and Deafness	614896	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**2**		C
CACNA1S	114208	Malignant hyperthermia susceptibility 5	601887	AD	Anesthesia-induced malignant hyperthermia	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	1	3	3	2	11	**1**		C
CACNB2	600003	Brugada Syndrome 4	611876	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	2	11	**1**		C
CALM1	114180	Arrhythmia (Long QT Syndrome 14 / Ventricular tachycardia, catecholaminergic polymorphic, 4)	616247	AD	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	2	2	2	12	**1**		
CALM2	114182	Long QT Syndrome 15	616249	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	2	11	**1**		
CALR3	611414	?Cardiomyopathy, Familial Hypertrophic, 19	613875	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		
CARD11	607210	Immunodeficiency 11	615206	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	3	3	2	1	12	**1**		
CASQ2	114251	Ventricular tachycardia, catecholaminergic polymorphic, 2 (recessive)	611938	AR	Arrhythmia and severe cardiac events	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	2	11	**1**		A
CASQ2	114251	Ventricular Tachycardia, catecholaminergic polymorphic, 2	611938	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	0	9	**1**		
CASR	601199	Hyperparathyroidism, neonatal severe	239200	AR	Hypercalcemia - severe	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	2	13	**1**		
CASR	601199	Hypocalcemia, AD, w/ or w/o Bartter syndrome	601198	AD	Hypocalcemia	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	1	11	**1**		
CATSPER2	607249	Sensorineural Deafness and Male Infertility	611102	AR	Male infertility	Variable	Referral to specialists for surveillance and early management	Neonatal	0	3	2	2	1	8	**2**		
CAV1	601047	Pulmonary hypertension, primary, 3 / ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome / ?Lipodystrophy, congenital generalized, type 3	615343	AD	Pulmonary hypertension	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	1	2	0	8	**2**		
CAV3	601253	Long QT Syndrome 9	611818	AD	Arrhythmia and severe cardiac events	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	3	0	2	2	0	7	**2**		C
CBS	613381	Homocystinuria, B6-responsive and nonresponsive types	236200	AR	Thrombosis	Childhood	Referral to specialists for surveillance and dietary management	Infant	2	3	3	2	3	13	**1**	Core	A
CCDC103	614677	Ciliary dyskinesia, primary, 17	614679	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		C
CCDC114	615038	Ciliary dyskinesia, primary, 20	615067	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		
CCDC151	615956	Ciliary dyskinesia, primary, 30	616037	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		
CCDC39	613798	Ciliary dyskinesia, primary, 14	613807	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	3	11	**1**		A
CCDC40	613799	Ciliary dyskinesia, primary, 15	613808	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		A
CCDC50	611051	Deafness, autosomal dominant 44	607453	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	1	11	**2**		C
CCDC65	611088	Ciliary dyskinesia, primary, 27	615504	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
CCNO	607752	Ciliary dyskinesia, primary, 29	615872	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
CD247	186780	Immunodeficiency due to defect in CD3-zeta	610163	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	0	3	2	1	8	**2**		
CD36	173510	Platelet Glycoprotein IV Deficiency	608404	AR	Bleeding	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	0	0	2	2	2	6	**2**		C
CD3D	186790	Immunodeficiency 19	615617	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	1	12	**1**		
CD3E	186830	Immunodeficiency 18 (Severe Combined Immunodeficiency)	615615	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	1	12	**1**		
CD3G	186740	Immunodeficiency 17, CD3 gamma deficient	615607	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	0	3	2	0	8	**2**		
CD40LG	300386	Immunodeficiency, X-linked, with hyper-IgM	308230	X-linked	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	2	2	2	11	**1**		A

CDC73	607393	Hyperparathyroidism, familial primary	145000	AD	Parathyroid carcinoma	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	1	3	3	3	2	12	**1**		
CDH1	192090	Hereditary diffuse gastric cancer	137215	AD	Gastric cancer	Adulthood	Referral to specialists for surveillance and/or preemptive management	Variable	2	3	3	0	3	11	**3**		B
CDH23	605516	Deafness, autosomal recessive 12 (DFNB12)	601386	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
CDH23	605516	Usher Syndrome, type 1D	601067	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	3	13	**1**		A
CDH23 / PCDH15	605514	Usher Syndrome, type ID / F Digenic	601067	Multiple and/or complex pattern	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	1	11	**2**		
CDKN2A	600160	Pancreatic cancer / melanoma syndrome	606719	AD	Melanoma	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	2	3	2	2	3	12	**1**		B
CEACAM16	614591	?Deafness, autosomal dominant 4B	614614	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**2**		C
CEL	114840	MODY, type VIII (MODY8)	609812	AD	Diabetes mellitus	Adult	Referral to specialists for surveillance and preemptive management	Childhood	1	3	2	2	1	9	**2**		
CENPF	600236	Stromme Syndrome	243605	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	Neonatal	2	3	0	0	1	6	**2**		
CFTR	602421	Cystic Fibrosis	219700	AR	Pulmonary disease	Infant	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
CIB2	605564	Usher Syndrome, type IJ	614869	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	2	12	**1**		
CIB2	605564	Deafness, autosomal recessive 48	609439	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		
CISD2	611507	Wolfram syndrome 2	604928	AR	Diabetes mellitus / insipidus	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	2	2	12	**1**		C
CLCNKB	602023	Bartter syndrome, type 3	607364	AR	Hypotension	Variable	Referral to specialists for surveillance and preemptive management	Variable	2	3	2	3	2	12	**1**		
CLDN14	605608	Deafness, autosomal recessive 29	614035	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
CLN3	607042	Ceroid lipofuscinosis, neuronal, 3	204200	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	3	8	**2**		A
CLN5	608102	Ceroid lipofuscinosis, neuronal, 5 (CLN5)	256731	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
CLN6	606725	Ceroid lipofuscinosis, neuronal, 6	601780	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8 (CLN8), Northern Epilepsy Variant	610003	AR	Epileptic seizures and intellectual disability	Adulthood	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	3	7	**4**		
CLN8	607837	Ceroid lipofuscinosis, neuronal, 8 (CLN8)	600143	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
CLPP	601119	Deafness, autosomal recessive 81 (aka Perrault syndrome 3)	614129	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
CLRN1	606397	Usher Syndrome, type 3A	276902	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	2	11	**1**		A
COCH	603196	Deafness, autosomal dominant 9 (Non-syndromic deafness, dominant)	601369	AD	Communication deficits and vestibular involvement	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	3	3	3	2	12	**3**		A
COL11A1	120280	Fibrochondrogenesis 1	228520	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		
COL11A1	120280	Stickler syndrome, type II / Marshall syndrome	604841	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and preemptive management	Infant	2	3	3	3	2	13	**1**		A
COL11A2	120290	Otospondylomegaepiphyseal Dysplasia	215150	Multiple and/or complex pattern	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
COL11A2	120290	Stickler Syndrome, Type III	184840	Multiple and/or complex pattern	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
COL11A2	120290	Deafness, autosomal recessive 53	609706	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
COL11A2	120290	Deafness, autosomal dominant 13 (DFNA13)	601868	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Childhood	0	3	3	3	1	10	**2**		
COL1A1	120150	Osteogenesis Imperfecta, type I	166200	AD	Osteogenesis imperfecta manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	2	3	11	**1**		
COL1A1	120150	Caffey Disease	114000	AD	Hyperostosis and pain	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		C
COL1A2	120160	Ehlers-Danlos syndrome, cardiac valvular form	225320	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Childhood	2	3	1	2	1	9	**2**		
COL1A2	120160	Osteogenesis imperfecta, type I & type III	166200	AD	Osteogenesis imperfecta manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	2	3	11	**1**		A
COL2A1	120140	Stickler syndrome, type I / Epiphyseal dysplasia, multiple, with myopia and deafness / Kniest dysplasia / Spondyloperipheral dysplasia	108300	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	3	13	**1**		A
COL3A1	120180	Ehlers-Danlos Syndrome Type IV - Vascular Type	130050	AD	Arterial dissection and organ rupture	Variable	Referral to specialists for surveillance and preemptive management	Childhood	3	3	1	2	2	11	**1**		A
COL4A3	120070	Alport syndrome, autosomal recessive	203780	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		A
COL4A4	120131	Alport syndrome, autosomal recessive	203780	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		A
COL4A5	303630	Alport Syndrome	301050	X-linked	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		A
COL9A1	614134	Stickler syndrome, type IV	614134	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
COQ6	614647	Coenzyme Q10 deficiency, primary, 6	614650	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	1	11	**1**		C
CORO1A	605000	Immunodeficiency 8	615401	AR	Recurrent infection	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	2	1	11	**1**		
CP	117700	Hemosiderosis, systemic, due to aceruloplasminemia	604290	AR	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	2	3	1	10	**3**		B
CPS1	608307	Carbamoylphosphate synthetase I deficiency	237300	AR	Hyperammonemic crisis	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	2	3	13	**1**		A
CPT1A	600528	CPT deficiency, hepatic, type IA	255120	AR	Hypoketotic hypoglycemia	Infant	Referral to specialists for surveillance and dietary management	Infant	3	2	2	3	2	12	**1**	Secondary	A
CPT2	600650	Myopathy due to CPTII Deficiency	255110	AR	Myopathy and rhabdomyolysis	Childhood	Referral to specialists for surveillance and dietary management	Infant	2	3	3	3	2	13	**1**	Secondary	A
CRYAB	123590	Cardiomyopathy, dilated, 1II	615184	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		B
CSF2RA	306250	Surfactant metabolism dysfunction, pulmonary, 4 / Familial pulmonary alveolar proteinosis	300770	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Childhood	1	3	3	2	1	10	**1**		A
CSF2RB	138981	Surfactant metabolic dysfunction, pulmonary, 5 / Familial pulmonary alveolar proteinosis	614370	AR	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Childhood	1	3	3	2	0	9	**2**		C
CSRP3	600824	Cardiomyopathy (Hypertrophic 12, ?dilated, 1M)	612124	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	1	11	**1**		B
CTNNA3	607667	Arrhythmogenic right ventricular dysplasia 13	615616	AD	Arrhythmia and severe cardiac events	Adolescent	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		
CTNS	606272	Cystinosis	219800	AR	Kidney failure	Childhood	Referral to specialists for surveillance and early management	Neonatal	2	3	3	3	3	14	**1**		A
CTSA	613111	Galactosialidosis	256540	AR	Cardiac involvement	Childhood	Referral to specialists for surveillance and supportive care	N / A	3	2	0	0	1	6	**2**		
CTSD	116840	Ceroid lipofuscinosis, neuronal, 10 (CLN10)	610127	AR	Neurodegeneration and retinal degeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		A
CTSK	601105	Pycnodysostosis	265800	AR	Short stature	Variable	Referral to specialists for surveillance and early management	Variable	0	3	2	3	2	10	**2**		A

CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	202010	AR	Androgen excess and hypertension	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	2	11	**1**		A
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	201910	AR	Salt-wasting crises	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	3	14	**1**		A
CYP27B1	609506	Vitamin D-dependent rickets, type I	264700	AR	Bone disease	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
CYRL1	609877	Nonsyndromic hearing loss	N/A	AR	Communication deficits	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
DBT	248610	Maple syrup urine disease, type II	248600	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	3	12	**1**	Core	A
DCLRE1C	605988	Severe Combined Immunodeficiency, Athabascan Type	602450	AR	Immunodeficiency - combined	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	2	13	**1**		A
DES	125660	Cardiomyopathy (Myopathy myofibrillar 1, dilated 1I)	601419	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		A,B
DFNA5	608798	Deafness, autosomal dominant 5	600994	AD	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Childhood	0	3	3	3	2	11	**2**		A
DFNB59 / PJVK	610219	Non-Syndromic Deafness, Recessive	610220	AR	Communication deficits with or without auditory neuropathy spectrum disorder	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
DHCR7	602858	Smith-Lemli-Opitz syndrome	270400	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	Neonatal	2	3	0	0	3	8	**2**		A
DIABLO	605219	Deafness, autosomal dominant 64	614152	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	1	0	3	3	0	7	**2**		C
DIAPH1	602121	Deafness, autosomal dominant 1	124900	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**2**		C
DKC1	300126	Dyskeratosis congenita, X-linked	305000	X-linked	Bone marrow failure	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	1	2	2	10	**2**		B
DLD	238331	Dihydrolipoamide dehydrogenase deficiency	246900	AR	Mitochondrial encephalopathy and Leigh syndrome manifestations	Neonatal	Referral to specialists for surveillance and supportive care	Neonatal	3	3	0	0	1	7	**2**		A
DMD	300377	Cardiomyopathy, Dilated, 3B	302045	X-linked	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	1	11	**2**		B
DNAAF1	613190	Ciliary dyskinesia, primary, 13	613193	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		A
DNAAF2	612517	Ciliary dyskinesia, primary, 10	612518	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		C
DNAAF3	614566	Ciliary Dyskinesia, Primary, 2	606763	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		C
DNAAF5	614864	Ciliary dyskinesia, primary, 18	614874	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
DNAH1	603332	?Ciliary dyskinesia, primary, 37	617577	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	0	2	2	0	5	**2**		
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus	611884	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	3	11	**1**		A
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	608644	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	3	11	**1**		A
DNAH8	603337	Ciliary dyskinesia, primary, 1	244400	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	0	2	2	0	5	**2**		
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus (Kartagener Syndrome)	244400	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	3	11	**1**		A
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	612444	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		C
DNAJB13	610263	Ciliary dyskinesia, primary, 34	617091	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
DNAJC5	611203	Ceroid lipofuscinosis, neuronal, 4, Parry type (CLN4B)	162350	AD	Neurodegeneration and retinal degeneration	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	3	8	**4**		C
DNAL1	610062	Ciliary dyskinesia, primary, 16	614017	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		C
DNMT3B	602900	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	242860	AR	Immunodeficiency	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	2	2	12	**1**		A
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive	243700	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	2	2	11	**1**		A
DRC1	615288	Ciliary dyskinesia, primary, 21	615294	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
DSC2	125645	Arrhythmogenic right ventricular cardiomyopathy 11	610476	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	2	2	2	1	9	**1**		B
DSG2	125671	Cardiomyopathy (ARVC 10, Dilated, 1BB)	610193	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Childhood	3	2	2	3	2	12	**1**		B
DSP	125647	Cardiomyopathy (Dilated with woolly hair and keratoderma / Arrhthmogenic Right Ventricular Cardiomyopathy 8) and related phenotypes	605676	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Childhood-Adolescence	Referral to specialists for surveillance and early management	Childhood	2	2	2	3	3	12	**1**		B
DSPP	125485	Deafness, autosomal dominant 36, with dentinogenesis	605594	AD	Dentinogenesis imperfecta	Infant	Referral to specialists for surveillance and supportive care	Infant	1	2	0	0	2	5	**2**		
DTNBP1	607145	Hermansky-Pudlak Syndrome 7	614076	AR	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	0	9	**4**		C
DUOX2	606759	Thyroid dyshormonogenesis 6	607200	AR	Hypothyroidism	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
DUOXA2	612772	Thyroid dyshormonogenesis 5	274900	AR	Hypothyroidism	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		C
DYX1C1	608706	Ciliary dyskinesia, primary, 25	615482	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
EDN3	131242	Waardenburg syndrome, type 4B	613265	Multiple and/or complex pattern	Syndromic manifestations	Birth	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	3	2	2	12	**1**		
EDN3	606118	Waardenburg syndrome, type 4B (Dominant)	613265	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	2	3	2	2	11	**1**		C
EDN3	131242	Central hypoventilation syndrome / Waardenburg syndrome, type 4B, (Hirschsprung disease, susceptibility to, 4)	209880	AD	Hypoventilation	Birth	Referral to specialists for surveillance and early management	Neonatal	3	0	3	2	0	8	**2**		
EDNRB	131244	Waardenburg syndrome, type 4A	277580	Multiple and/or complex pattern	Syndromic manifestations	Birth	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	3	2	2	12	**1**		C
EDNRB	606682	Waardenburg syndrome, type 4A (Dominant)	277580	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	2	3	2	2	11	**1**		
EFEMP2	604633	Cutis laxa, AR, type IB	614437	AR	Emphysema and cutis laxa	Birth	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		C
ELMOD2	610196	Pulmonary Fibrosis	610196	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	0	2	**4**		
ELN	130160	Cutis Laxa, AD 1 / Supravalvar aortic stenosis	185500	AD	Heart failure	Childhood	Referral to specialists for surveillance and early management	Neonatal	2	3	3	3	2	13	**1**		A
ELN	130160	Cutis Laxa, AD 1	123700	AD	Cutis laxa	Birth	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	2	6	**2**		
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1	187300	AD	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Infant	2	2	2	3	3	12	**1**		A
EPCAM	185535	Colorectal cancer, hereditary nonpolyposis, type 8	613244	Multiple and/or complex pattern	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	3	13	**3**		C
ERCC4	133520	Fanconi anemia, complementation group Q	615272	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**		
ESPN	606351	Deafness, autosomal recessive 36	609006	AR	Communication deficits with or without vestibular involvement	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
ESPN	606351	Deafness, neurosensory, without vestibular involvement, autosomal dominant	609006	AD	Communication deficits without vestibular involvement	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	0	3	3	0	7	**2**		
ESRRB	602167	Non-Syndromic Deafness, Recessive 35	608565	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A

ETFA	608053	Glutaric acidemia IIA	231680	AR	Metabolic crisis	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	3	12	**1**	Secondary	A
ETFA	608053	Glutaric acidemia IIA (severe forms)	231680	AR	Hypoglycemia and neonatal acidosis	Neonatal	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**	Secondary	
ETFB	130410	Glutaric acidemia IIB	231680	AR	Metabolic crisis	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	3	12	**1**	Secondary	A
ETFB	130410	Glutaric acidemia IIB (severe forms)	231680	AR	Hypoglycemia and neonatal acidosis	Neonatal	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**	Secondary	
ETFDH	231675	Glutaric acidemia IIC	231680	AR	Metabolic crisis	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	3	12	**1**	Secondary	A
ETFDH	231675	Glutaric acidemia IIC (severe forms)	231680	AR	Hypoglycemia and neonatal acidosis	Neonatal	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**	Secondary	
ETFDH	231675	Coenzyme Q10 Deficiency, secondary	N/A	AR	Myopathy - isolated	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	3	3	1	11	**1**		
EYA1	601653	Branchiootorenal syndrome 1, with or w/o cataracts / Anterior segment anomalies with or w/o cataracts	113650	AD	Syndromic manifestations	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	2	11	**1**		A
EYA4	603550	Non-Syndromic Deafness, Dominant 10	601316	AD	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**3**		A
EYA4	603550	Cardiomyopathy, dilated, 1J	605362	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		
F10	613872	Factor X deficiency	227600	AR	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		
F11	264900	Factor XI deficiency, autosomal recessive	612416	Multiple and/or complex pattern	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Variable	0	3	3	3	2	11	**1**		A
F11	264900	Factor XI deficiency, autosomal dominant	612416	AD	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	1	3	2	1	7	**2**		
F12	610619	Factor XII deficiency	234000	AR	Clotting	Variable	Referral to specialists for surveillance and supportive care	N / A	0	0	0	0	1	1	**4**		
F12	610619	Angioedema, hereditary, type III	610618	AD	Swelling and respiratory compromise	Variable	Referral to specialists for surveillance and preemptive management	Variable	2	2	2	3	1	10	**1**		
F13A1	134570	Factor XIIIA deficiency	613225	AR	Intracranial hemorrhage	Infant	Referral to specialists for surveillance and early management	Neonatal	2	2	3	3	2	12	**1**		
F13B	134580	Factor XIIIB deficiency	613235	AR	Intracranial hemorrhage	Neonatal	Referral to specialists for surveillance and early management	Neonatal	2	2	3	3	2	12	**1**		
F2	176930	Prothrombin Deficiency, congenital (Dysprothrombinemia, Hypoprothrombinemia)	613679	AR	Intracranial hemorrhage	Birth	Referral to specialists for surveillance and early management	Neonatal	2	2	3	3	2	12	**1**		A
F5	612309	Factor V deficiency	227400	AR	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	2	12	**1**		
F5	612309	Thrombophilia due to activated protein C resistance (Homozygote)	188055	AD	Thrombosis	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	2	3	2	3	2	12	**1**		C
F7	613878	Factor VII deficiency	227500	AR	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	2	3	3	3	13	**1**		
F8	300841	Hemophilia A	306700	X-linked	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	3	3	3	14	**1**		A
F9	300746	Hemophilia B	306900	X-linked	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	3	3	3	14	**1**		A
F9	300746	Thrombophilia, X-linked, due to factor IX defect	300807	X-linked	Deep vein thrombosis	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	0	2	3	1	8	**2**		
FAH	613871	Tyrosinemia, type I	276700	AR	Liver failure and hepatocellular carcinoma	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	3	2	3	13	**1**	Core	A
FANCA	607139	Fanconi anemia, complementation group A	227650	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
FANCB	300515	Fanconi anemia, complementation group B	300514	X-linked	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
FANCC	613899	Fanconi anemia, complementation group C	227645	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
FANCD2	613984	Fanconi anemia, complementation group D2	227646	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
FANCE	613976	Fanconi anemia, complementation group E	600901	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		C
FANCF	613897	Fanconi anemia, complementation group F	603467	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		C
FANCG	602956	Fanconi anemia, complementation group G	614082	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
FANCI	611360	Fanconi anemia, complementation group I	609053	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
FANCL	608111	Fanconi anemia, complementation group L	614083	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	1	10	**1**		C
FANCM	609644	Fanconi-Anemia-Associated Polypeptide	N/A	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**		C
FBLN5	604580	Cutis laxa, AR, type IA	219100	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and supportive care	Neonatal	2	3	0	0	2	7	**2**		A
FBN1	134797	Marfan syndrome and other aortopathy syndromes	154700	AD	Aortic dissection	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		A
FBP1	611570	Fructose bisphosphatase deficiency	229700	AR	Hypoglycemia and metabolic acidosis	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	3	2	13	**1**		
FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia	610706	AR	Communication deficits	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
FGFR3	134934	CATSHL syndrome	610474	Multiple and/or complex pattern	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**2**		C
FGFR3	134934	Muenke syndrome	602849	AD	Craniosynostosis	Neonatal	Referral to specialists for surveillance and early management	Infant	1	3	3	2	3	12	**1**		A
FH	136850	Fumarase deficiency	606812	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
FH (Dominant)	136850	Leiomyomatosis and renal cell cancer	150800	AD	Renal cancer	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	2	2	3	2	11	**3**		C
FKTN	607440	Cardiomyopathy, dilated, 1X	611615	AR	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	0	0	2	7	**2**		
FLCN	607273	Birt-Hogg-Dube syndrome	135150	AD	Renal cancer	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	1	2	2	3	2	10	**4**		A
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency	613068	AR	Neurodegeneration	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
FOXE1	602617	Bamforth-Lazarus syndrome	241850	AR	Hypothyroidism	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	1	11	**1**		C
FOXF1	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins	265380	AD	Pulmonary hypertension and congenital alveolar capillary dysplasia	Birth	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
FUCA1	612280	Fucosidosis	230000	AR	Neurodegeneration and psychomotor impairment	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	2	1	2	10	**2**		A
G6PC	613742	Glycogen Storage Disease 1a	232200	AR	Severe hypoglycemia	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	2	3	13	**1**		A
G6PD	305900	Hemolytic anemia due to G6PD deficiency	300908	X-linked	Hemolytic anemia	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	1	2	3	3	3	12	**1**		A
GAA	606800	Glycogen storage disease II (GSD2)	232300	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Infant	3	3	3	2	3	14	**1**	Core	A
GALC	606890	Krabbe Disease	245200	AR	Neurodegeneration	Birth	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
GALE	606953	Galactose epimerase deficiency	230350	AR	Galactosemia	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	0	2	2	1	6	**2**	Secondary	

GALK1	604313	Galactokinase Deficiency with Cataracts	230200	AR	Vision impairment	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	3	3	3	13	**1**	Secondary	A
GALNS	612222	Mucopolysaccharidosis IVA	253000	AR	Skeletal abnormalities	Infant	Referral to specialists for surveillance and early management	Neonatal	1	3	1	2	2	9	**1**		A
GALT	606999	Galactosemia	230400	AR	Liver failure or E. coli sepsis	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	2	3	13	**1**	Core	A
GAMT	601240	Cerebral creatine deficiency syndrome 2; Guanidinoacetate methyltransferase deficiency	612736	AR	Encephalopathy	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	1	10	**1**		
GAS8	605178	Ciliary dyskinesia, primary, 33	616726	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
GATA1	305371	Anemia, with or without neutropenia and/or platelet abnormalities	300835	X-linked	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	1	10	**1**		A
GATA2	137295	Immunodeficiency 21	137295	AD	Immunodeficiency and myeloid malignancies	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	3	2	2	12	**1**		
GATA3	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	146255	AD	Syndromic manifestations	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	2	12	**1**		
GATAD1	614518	?Cardiomyopathy, dilated, 2B	614672	AR	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		C
GATM	602360	Cerebral creatine deficiency syndrome 3; L-arginine:glycine amidinotransferase deficiency	612718	AR	Encephalopathy	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	1	10	**1**		
GBA	606463	Gaucher Disease, Type I	230800	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	3	13	**1**		A
GBA	606463	Gaucher Disease, all other types (perinatal lethal, type II, type III, type IIIC)	230800	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		
GBE1	607839	Glycogen Storage Disease IV	232500	AR	Neurodegeration and psychomotor impairment	Neonatal	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		A
GCDH	608801	Glutaricaciduria, type I	231670	AR	Metabolic crisis	Variable	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	3	3	13	**1**	Core	A
GCH1	600225	GTPCH1 deficiency (biopterin deficiency)	233910	AR	Neurologic involvement	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	3	3	1	11	**1**	Secondary	
GCH1	600225	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia	128230	AD	Dystonia	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	3	13	**1**		B
GCK	138079	Diabetes mellitus, permanent neonatal	606176	AR	Hyperglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
GCK	138079	MODY, type II (MODY2)	125851	AD	Diabetes mellitus	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	0	3	0	0	3	6	**2**		
GDF2	605120	Telangiectasia, hereditary hemorrhagic, type 5	615506	AD	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Childhood	2	0	2	2	0	6	**2**		
GDNF	600837	Central hypoventilation syndrome	209880	AD	Hypoventilation	Birth	Referral to specialists for surveillance and early management	Neonatal	3	0	3	2	0	8	**2**		C
GGCX	137167	Vitamin K-dependent coagulation defect	277450	AR	Bleeding	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
GH1	139250	Growth hormone deficiency, isolated, type IA (recessive)	262400	AR	Postnatal growth hormone deficiency and hypoglycemia	Infant	Referral to specialists for surveillance and early management	Neonatal	1	3	2	3	3	12	**1**		
GH1	139250	Growth hormone deficiency, isolated, type IA (dominant)	173100	AD	Postnatal growth hormone deficiency and hypoglycemia	Infant	Referral to specialists for surveillance and early management	Neonatal	1	3	2	3	3	12	**1**		
GIF	609342	Intrinsic factor deficiency	261000	AR	Pernicious anemia	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
GIPC3	608792	Deafness, Autosomal Recessive 15	601869	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
GJA5	121013	Atrial fibrillation, familial, 11	614049	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**3**		B
GJB2	121011	Deafness, autosomal recessive 1A (DFNB1A)	220290	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
GJB2	121011	GJB2-related hearing loss and related disorders	148210	AD	Communication deficits with or without vision loss	Adulthood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
GJB6	604418	Non-Syndromic Deafness, Recessive 1B	612645	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
GLA	300644	Fabry Disease	301500	X-linked	Syndromic manifestations	Variable	Referral to specialists for surveillance and preemptive management	Childhood	2	3	2	2	3	12	**1**		A
GLB1	611458	Beta-galactosidase-1 deficiency GLB1 deficiency	230500	AR	Skeletal dysplasia	Childhood	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	3	7	**2**		A
GM2A	613109	GM2-ganglosidosis, AB variant	272750	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		
GNMT	606628	Glycine N-Methyltransferase Deficiency	606664	AR	Hypermethioninemia	Childhood	Referral to specialists for surveillance and dietary management	Infant	0	3	0	0	0	3	**2**	Secondary	
GNPTAB	607840	Mucolipidosis II (I-cell disease)	252500	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
GNPTG	607838	Mucolipidosis III Gamma	252605	AR	Skeletal abnormalities	Childhood	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	2	6	**2**		A
GNS	607664	Mucopolysaccharidosis type IIID	252940	AR	Respiratory distress	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
GP1BA	606672	Bernard-Soulier Syndrome, type A1	231200	AR	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	2	11	**2**		
GP1BA	606672	Pseudo - von Willebrand disease, platelet-type	177820	AD	Thrombocytopenia and bleeding	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	2	11	**2**		
GP1BA	606672	Bernard-Soulier Syndome, type A2	153670	AD	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	0	3	3	0	6	**2**		
GP1BB	138720	Bernard-Soulier Syndrome, type B	231200	AR	Bleeding	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	2	11	**1**		
GP6	605546	Bleeding disorder, platelet-type, 11	614201	AR	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
GP9	173515	Bernard-Soulier Syndrome, type C	231200	AR	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	2	11	**1**		
GPD1L	611778	Brugada Syndrome 2	611777	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	2	2	1	9	**1**		B
GPSM2	609245	Chudley-McCullough Syndrome	604213	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
GRHL2	608576	Ectodermal dysplasia/short stature syndrome	616029	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	2	3	3	0	9	**2**		
GRHL2	608576	Deafness, autosomal dominant 28	608641	AD	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Variable	0	3	3	3	1	10	**2**		C
GRXCR1	613283	Deafness, autosomal recessive 25	613285	AR	Communication deficits with or without vestibular involvement	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		C
GSS	601002	Glutathione Synthetase Deficiency	266130	AR	Syndromic manifestations	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	1	10	**1**		A
GUSB	611499	Mucopolysaccharidosis VII	253220	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	1	1	9	**2**		A
GYS1	138570	Glycogen storage disease 0, muscle	611556	AR	Left ventricular hypertrophy and cardiac arrest	Childhood	Referral to specialists for surveillance and supportive care	N / A	3	0	0	0	0	3	**2**		
GYS2	138571	Glycogen Storage Disease, type 0	240600	AR	Hypoglycemia	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	3	3	1	11	**1**		A
HADH	601609	3-Hydroxyacyl-CoA Dehydrogenase Deficiency	231530	AR	Hypoglycemia - profound	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	3	2	13	**1**	Secondary	C
HADHA	600890	LCHAD deficiency	609016	AR	Hypoglycemic crises	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	3	3	3	3	15	**1**	Core	
HADHA	600890	Trifunctional protein deficiency	609015	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	0	1	2	1	7	**2**		A

HADHB	143450	Trifunctional protein deficiency	609015	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	3	2	3	2	13	**1**	Core	A
HAMP	606464	Hemochromatosis, type 2B	613313	AR	Iron overload	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	2	3	3	3	2	13	**1**		C
HARS	142810	Usher Syndrome, type 3B	614504	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	1	10	**2**		B
HARS2	600783	Perrault Syndrome 2	614926	AR	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	0	10	**2**		C
HBB	141900	Hb S/C	N/A	Multiple and/or complex pattern	Sickle cell anemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	3	3	3	13	**1**		
HBB	141900	Thalassemias, beta- (Major, AR)	613985	AR	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	2	3	3	2	3	13	**1**		A
HBB	141900	Sickle cell anemia	603903	AR	Sickle cell anemia manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	3	12	**1**		
HBG2	142250	Cyanosis, transient neonatal	613977	AD	Anemia and cyanosis	Birth	Referral to specialists for surveillance and early management	Neonatal	0	3	3	3	2	11	**1**		
HCN4	605206	Brugada Syndrome 8	613123	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		C
HEXA	606869	Tay-Sachs Disease	272800	AR	Psychomotor degeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
HEXB	268800	Sandhoff Disease	606873	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
HFE - C282Y / H63D	613609	Hereditary hemochromatosis C282Y / H63D Compound Hets	N/A	AR	Iron overload (heart, liver)	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	1	3	3	3	11	**3**		
HFE - C282Y homozvaous	613609	Hereditary hemochromatosis	235200	AR	Iron overload	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	2	3	3	3	12	**3**		C
HFE2	608374	Hemochromatosis, type 2A	602390	AR	Iron overload	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	2	3	3	3	3	14	**1**		C
HGD	607474	Alkaptonuria	203500	AR	Arthritis	Adulthood	Referral to specialists for surveillance and supportive care	Adulthood	1	3	0	0	3	7	**4**		A
HGF	142409	Deafness, autosomal recessive 39	608265	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
HGSNAT	610453	Mucopolysaccharidosis type IIIC	252930	AR	Neurodegeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
HLCS	609018	Holocarboxylase synthetase deficiency	253270	AR	Seizures	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**	Core	A
HMGCL	613898	HMG-CoA lyase deficiency	246450	AR	Hypoglycemia - severe	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	2	11	**1**		A
HMGCS2	600234	HMG-CoA synthase-2 deficiency	605911	AR	Hypoketotic hypoglycemic	Infant	Referral to specialists for surveillance and dietary management	Infant	2	3	3	3	1	12	**1**	Core	
HNF1A	142410	MODY, type III (MODY3)	600496	AD	Diabetes mellitus and hyperglycemia	Childhood-Adolescence	Referral to specialists for surveillance and preemptive management	Childhood	1	3	2	3	3	12	**1**		
HNF1B	189907	Renal cysts and diabetes syndrome (MODY5)	137920	AD	Diabetes mellitus and renal cysts	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	1	2	2	9	**2**		C
HNF4A	600281	MODY, type I (MODY1)	125850	AD	Diabetes mellitus	Childhood-Adult	Referral to specialists for surveillance and preemptive management	Childhood	1	3	2	3	2	11	**1**		
HPD	609695	Tyrosinemia, type III	276710	AR	Intellectual disability and psychomotor degeneration	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	1	0	1	2	0	4	**2**	Secondary	C
HPS1	604982	Hermansky-Pudlak syndrome 1	203300	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	1	3	3	3	3	13	**1**		A
HPS3	606118	Hermansky-Pudlak Syndrome 3	614072	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	0	3	3	3	2	11	**1**		A
HPS4	606882	Hermansky-Pudlak Syndrome 4	614073	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	1	3	3	3	3	13	**1**		A
HPS5	607521	Hermansky-Pudlak syndrome 5	614074	AR	Bleeding	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	1	10	**2**		A
HPS6	607522	Hermansky-Pudlak syndrome 6	614075	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	0	3	3	3	1	10	**2**		C
HRAS	190020	Costello syndrome (pulmonary hypertension, pneumothorax, pulmonary fibrosis, & broncho/tracheo-malacia)	218040	AD	Cardiomyopathy	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	3	12	**1**		A
HSD11B2	614232	Apparent mineralocorticoid excess	218030	AR	Hypertensive crisis	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	2	3	3	3	2	13	**1**		
HSD17B10	300256	17-Beta-Hydroxysteroid Dehydrogenase X Deficiency	300438	X-linked	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and supportive care	Neonatal	0	0	0	0	1	1	**2**	Secondary	A
HSD17B4	601860	D-bifunctional protein deficiency	261515	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		A
HSD17B4	601860	Perrault Syndrome 1	233400	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and supportive care	Neonatal	1	3	2	3	0	9	**2**		
HSD3B2	613890	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	201810	AR	Salt-wasting crises	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	3	3	3	14	**1**		
HYAL1	607071	Mucopolysaccharidosis type IX	601492	AR	Swollen joints and periarticular masses	Variable	Referral to specialists for surveillance and supportive care	N / A	0	3	0	0	1	4	**2**		
HYDIN	610812	Ciliary dyskinesia, primary, 5	608647	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		C
IDS	300823	Mucopolysaccharidosis Type II	309900	X-linked	Syndromic manifestations	Variable	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		A
IDUA	252800	Mucopolysaccharidosis Ih	607014	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Infant	3	3	2	1	2	11	**1**	Core	A
IKBKG	300248	Immunodeficiency, isolated	300584	X-linked	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	0	1	2	1	6	**2**		A
IL21R	605383	Immunodeficiency, primary, autosomal recessive, IL21R-related	615207	AR	Immunodeficiency	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	2	3	3	2	0	10	**2**		
IL2RA	147730	Interleukin-2 receptor, alpha chain, deficiency of	606367	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Childhood	2	3	3	2	1	11	**1**		
IL2RG	308380	Severe Combined Immunodeficiency, X-linked	300400	X-linked	Immunodeficiency - combined	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	3	15	**1**		A
IL7R	146661	Severe Combined Immunodeficiency, T-cell Negative, B-cell / Natural Killer Cell-Positive Type	608971	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	2	13	**1**		
ILDR1	609739	Deafness, autosomal recessive 42	609646	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
INS	176730	Diabetes mellitus, permanent neonatal	606176	Multiple and/or complex pattern	Hyperglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
ITGA2	192974	Glycoprotein Ia deficiency / Bleeding disorder, platelet-type, 9	614200	AD	Bleeding	Birth	Referral to specialists for surveillance and supportive care	N / A	0	3	0	0	1	4	**2**		
ITGA2B	607759	Glanzmann thrombasthenia	273800	AR	Bleeding	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
ITGA2B	607759	Bleeding disorder, platelet-type, 16	187800	AD	Bleeding	Birth	Referral to specialists for surveillance and supportive care	N / A	0	2	0	0	1	3	**2**		
ITGB3	173470	Glanzmann thrombasthenia	273800	AR	Bleeding	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
ITGB3	173470	Bleeding disorder, platelet-type, 16	187800	AD	Bleeding	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	2	3	3	1	10	**1**		
ITK	186973	Lymphoproliferative syndrome 1	613011	AR	Immunodeficiency - Epstein-Barr virus associated	Childhood	Referral to specialists for surveillance and early management	Childhood	3	3	1	1	1	9	**1**		
IVD	607036	Isovaleric acidemia	243500	AR	Metabolic decompensation	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	2	3	2	3	13	**1**	Core	A
IYD	612025	Thyroid dyshormonogenesis 4	274800	AR	Hypothyroidism	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		C

JAG1	601920	Alagille syndrome / Tetralogy of Fallot / ?Deafness, congenital heart defects, and posterior embryotoxon	118450	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	2	11	**1**		A
JAK3	600173	SCID, AR, T-negative/B-positive type	600802	AR	Immunodeficiency - combined	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	3	15	**1**		A
JPH2	605267	Cardiomyopathy, Familial Hypertrophic, 17	613873	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	1	8	**2**		C
JUP	173325	Naxos disease (recessive)	601214	AR	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Childhood	3	2	2	2	3	12	**1**		A
JUP	173325	Arrhythmogenic right ventricular dysplasia 12	611528	AD	Arrhythmia and severe cardiac events	Adolescent	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		B
KARS	601421	Deafness, autosomal recessive 89	613916	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
KCNA5	176267	Atrial fibrillation, familial, 7	612240	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**1**		B
KCND3	605411	Brugada Syndrome 9	616399	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		C
KCNE1	176261	Jervell and Lange-Nielsen syndrome 2 (Recessive)	612347	AR	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	3	3	2	3	14	**1**		A
KCNE1	176261	Long QT Syndrome - 5 (Dominant)	613695	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	2	2	2	10	**1**		B
KCNE2	603796	Long QT syndrome 6	613693	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	1	2	2	1	8	**2**		B
KCNE3	604433	Brugada Syndrome 6	613119	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	0	2	2	1	8	**2**		C
KCNH2	152427	Romano-Ward Long QT syndrome 2	613688	AD	Arrhythmia	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	3	12	**1**		B
KCNJ10	602208	SESAME syndrome	612780	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	1	3	2	3	2	11	**1**		
KCNJ11	600937	Hyperinsulinemic hypoglycemia, familial 2	601820	AR	Hyperinsulinemic hypoglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	1	12	**1**		A
KCNJ11	600937	Diabetes mellitus, transient and permanent neonatal with neurologic features / Maturity-onset diabetes of the young, type 13	606176	AD	Hyperglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	2	2	12	**1**		
KCNJ2	600681	Andersen-Tawil syndrome; LQT 7	170390	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	2	2	2	2	10	**1**		A
KCNJ5	600734	Hyperaldosteronism, familial, type III	613677	AD	Hypertensive crisis	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	2	3	3	2	1	11	**1**		
KCNJ5	600734	Long QT syndrome 13	613485	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	0	2	2	0	6	**2**		C
KCNK3	603220	Pulmonary hypertension, primary, 4	615344	AD	Pulmonary hypertension	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	1	2	1	9	**1**		
KCNQ1	607542	Jervell and Lange-Nielsen syndrome	220400	AR	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	3	2	2	3	13	**1**		A
KCNQ1	607542	Romano-Ward Long QT syndrome 1	192500	AD	Arrhythmia	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	3	12	**1**		B
KCNQ4	603537	Deafness, autosomal dominant 2A	600101	AD	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	2	12	**1**		A
KLF11	603301	MODY, type VII (MODY7)	610508	AD	Diabetes mellitus	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	3	0	0	0	4	**4**		
KRIT1	604214	Cerebral Cavernous Malformations - 1	116860	AD	Cerebral cavernous malformation	Adolescent-Adulthood	Referral to specialists for surveillance and early management	Infant	1	3	1	3	3	11	**3**		
LAMA4	600133	Cardiomyopathy, dilated, 1JJ	615235	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		C
LAMP2	309060	Glycogen Storage Disease IIb/Danon disease	300257	X-linked	All cardiovascular manifestations	Childhood	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**1**		A
LARS2	604544	Perrault Syndrome 4	615300	AR	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	0	10	**1**		C
LCK	153390	Immunodeficiency 22	615758	AR	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Childhood	3	0	3	2	1	9	**2**		
LDB3	605906	Cardiomyopathy, Familial Hypertrophic, 24 / Dilated IC w/ or w/o LVNC / Left ventricular noncompaction 3	601493	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		C
LDLR	606945	Familial hypercholesterolemia (homozygous)	143890	AR	Hypercholesterolemia and cardiovascular disease	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	1	3	11	**1**		
LDLR	606945	Familial hypercholesterolemia (heterozygous)	143890	AD	Hypercholesterolemia and cardiovascular disease	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	3	14	**1**		A
LDLRAP1	605747	Hypercholesterolemia, familial, autosomal recessive	603813	AR	Hypercholesterolemia	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	2	3	2	3	2	12	**1**		
LHFPL5	609427	Deafness, autosomal recessive 67	610265	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**2**		A
LHX3	600577	Pituitary hormone deficiency, combined, 3	221750	AR	Pituitary hormone deficiency manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
LIG4	601837	LIG4 Syndrome	606593	AR	Immunodeficiency - combined	Variable	Referral to specialists for surveillance and preemptive management	Infant	1	3	1	3	1	9	**2**		A
LIPA	613497	Cholesteryl Ester Storage Disease (Lysosomal Acid Lipase) *OR* Wolman Disease	278000	AR	Liver disease - late onset, slowly progressive	Variable	Referral to specialists for surveillance and supportive care	Variable	3	3	0	0	1	7	**2**		A
LMBRD1	612625	Methylmalonic aciduria and homocystinuria, cblF type	277380	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	2	11	**1**		A
LMNA	150330	Cardiomyopathy, dilated IA	115200	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		B
LOXHD1	613072	Deafness, autosomal recessive 77	613079	AR	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**1**		A
LRRC6	614930	Ciliary dyskinesia, primary, 19	614935	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		A
LRTOMT	612414	Deafness, autosomal recessive 63	611451	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
LTBP4	604710	Cutis laxa, AR, type IC	613177	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and supportive care	Neonatal	3	3	0	0	2	8	**2**		A
LYST	606897	Chediak-Higashi syndrome	214500	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	2	1	2	10	**2**		A
MAGT1	300715	X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection, and neoplasia (XMEN)	300853	X-linked	Recurrent infection	Variable	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	0	2	**2**		
MAN2B1	609458	Mannosidosis, alpha-, types I and II	248500	AR	Intellectual disability and psychomotor impairment	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	2	1	2	9	**2**		A
MANBA	609489	Beta Manosidosis	248510	AR	Intellectual disability and seizures	Childhood	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	2	6	**2**		
MARVELD2	610572	Deafness, autosomal recessive 49	610153	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
MAT1A	610550	Methionine Adenosyltransferase Deficiency, AR	250850	AR, AD	Hypermethioninemia - benign	Birth	Referral to specialists for surveillance and supportive care	N / A	0	3	0	0	2	5	**2**	Secondary	C
MAX	154950	Pheochromocytoma susceptibility	171300	AD	Malignant pheochromocytoma / paraganglioma	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	2	2	3	2	2	11	**1**		
MCCC1	609010	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	210200	AR	Mild weakness	Variable	Referral to specialists for surveillance and dietary management	Variable	0	3	1	3	1	8	**2**	Core	B
MCCC2	609014	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	210210	AR	Encephalopathy	Infant	Referral to specialists for surveillance and supportive care	Neonatal	2	1	0	0	1	4	**2**	Core	B
MCIDAS	614086	Ciliary dyskinesia, primary	N/A	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
MCOLN1	605248	Mucolipidosis IV	252650	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A

MEFV (heterozygous/homozygous	608107	Familial Mediterranean fever, AD/AR, Classic mutations associated with renal failure	134610 / 249100	Multiple and/or complex pattern	Amyloidosis and renal failure	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	2	3	3	3	12	**1**		A
MEN1	613733	Multiple endocrine neoplasia 1	131100	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	1	3	3	3	3	13	**1**		A
MFSD8	611124	Ceroid Lipofuscinosis, neuronal, 7 (CLN7)	610951	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
MIR96	611606	Deafness, autosomal dominant 50	613074	AD	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	1	11	**2**		C
MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8	614456	AD	Melanoma	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	2	1	2	3	1	9	**2**		
MITF	156845	Waardenburg Syndrome, type 2A	193510	AD	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
MLH1	120436	Mismatch repair cancer syndrome	276300	AR	Hereditary cancer predisposition	Childhood	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	1	2	1	9	**2**		
MLH1	120436	Lynch syndrome	609310	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	3	13	**3**		C
MLYCD	606761	Malonyl-CoA Decarboxylase Deficiency	248360	AR	Cardiomyopathy	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	2	2	3	2	11	**2**	Secondary	A
MMAA	607481	Methylmalonic aciduria, vitamin B12-responsive	251100	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	2	3	3	3	13	**1**	Core	A
MMAB	607568	Methylmalonic aciduria, vitamin B12-responsive, due to defect insynthesis of adenosylcobalamin, cblB complementation type	251110	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	3	3	13	**1**	Core	A
MMACHC	609831	Methylmalonic aciduria and homocystinuria, cblC type	277400	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	3	3	13	**1**	Secondary	A
MMADHC	611935	Methylmalonic aciduria and homocystinuria, cblD type	277410	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	1	10	**1**	Secondary	A
MPI	154550	CDG1b	602579	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Neonatal	1	3	3	3	3	13	**1**		A
MSH2	609309	Mismatch repair cancer syndrome	276300	AR	Hereditary cancer predisposition	Childhood	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	1	2	1	9	**2**		
MSH2	609309	Lynch syndrome	120435	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	3	13	**3**		C
MSH6	600678	Mismatch repair cancer syndrome	276300	AR	Hereditary cancer predisposition	Childhood	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	1	2	1	9	**2**		
MSH6	600678	Lynch syndrome	120435	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	3	13	**3**		C
MSRB3	613719	Deafness, autosomal recessive 74	613718	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
MTHFR	607093	{Thromboembolism, susceptibility to}	188050	Multiple and/or complex pattern	Thrombosis	Variable	Referral to specialists for surveillance and preemptive management	Variable	2	0	1	3	2	8	**2**		
MTHFR	607093	Homocystinuria due to MTHFR deficiency	236250	AR	Thrombosis	Variable	Referral to specialists for surveillance and preemptive management	Variable	2	3	3	3	2	13	**1**	Core	B
MTR	156570	Homocystinuria-megaloblastic anemia, cblG complementation type	250940	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	2	12	**1**	Core	A
MTRR	602568	Homocystinuria-megaloblastic anemia, cbl E type	236270	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	2	12	**1**	Core	A
MUC5B	600770	{Pulmonary fibrosis, idiopathic, susceptibility to} / Pulmonary fibrosis	178500	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	1	3	**4**		C
MUT	609058	Methylmalonic aciduria, mut(0) type	251000	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and dietary management	Neonatal	3	3	2	2	3	13	**1**	Core	A
MUTYH	604933	Attenuated FAP / MUTYH-associated polyposis	608456	AR	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	3	13	**3**		A
MYBPC3	600958	Cardiomyopathy (Hypertrophic 4, Dilated IMM)	115197	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**		B
MYH11	160745	Familial Thoracic Aortic Aneurysms 4	132900	AD	Aortic dissection	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	3	2	2	3	1	11	**1**		B
MYH14	608568	Non-Syndromic Deafness, Dominant 4A	600652	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Infant	0	3	3	3	1	10	**2**		A
MYH6	160710	Cardiomyopathy (Hypertrophic 14, dilated, 1EE)	613251	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		C
MYH7	160760	Cardiomyopathy (Hypertrophic 1, Dilated, 1S / Left Ventricular Noncompaction 5) and related myopathies	192600	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	2	3	3	12	**1**		A,B
MYH9	160775	Deafness, autosomal dominant 17	603622	AD	Communication deficits	Childhood - Adolescence	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	1	11	**2**		
MYH9	160775	MYH9-related disorders (MYH9RD)	153650	AD	Macrothrombocytopenia	Childhood	Referral to specialists for surveillance and early management	Childhood	0	3	3	3	3	12	**1**		A
MYL2	160781	Hypertrophic cardiomyopathy 10	608758	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	2	2	3	2	11	**1**		B
MYL3	160790	Hypertrophic cardiomyopathy 8	608751	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		B
MYLK	600922	Familial Thoracic Aortic Aneurysms 7	613780	AD	Aortic dissection	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	3	3	1	2	1	10	**4**		B
MYLK2	606566	Hypertrophic cardiomyopathy 1, digenic	192600	AD	Arrhythmia and severe cardiac events	Adolecent	Referral to specialists for surveillance and early management	Neonatal	0	0	2	3	0	5	**2**		C
MYO15A	602666	Deafness, autosomal recessive 3	600316	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
MYO3A	606808	Deafness, autosomal recessive 30	607101	AR	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	0	3	3	3	0	9	**2**		A
MYO6	600970	Non-Syndromic Deafness, Recessive 37	607821	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
MYO6	600970	Non-Syndromic Deafness, Dominant 22	606346	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	2	12	**1**		
MYO7A	276903	Deafness, autosomal recessive, 2	600060	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		
MYO7A	276903	Usher Syndrome, type 1B	276900	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	3	13	**1**		A
MYO7A	276903	Deafness, autosomal dominant 11	601317	AD	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**2**		
MYOZ2	605602	Cardiomyopathy, Familial Hypertrophic, 16	613838	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		C
MYPN	608517	Cardiomyopathy, Familial Hypertrophic, 22 / Dilated 1KK / Familial restrictive 4	615248	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	1	8	**2**		C
NAA10	300013	Ogden syndrome	300855	X-linked	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		C
NADK2	615787	?2,4-dienoyl-CoA reductase deficiency	616034	AR	Encephalopathy	Neonatal	Referral to specialists for surveillance and supportive care	N/A	3	3	0	0	1	7	**2**	Secondary	
NAGA	104170	Kanzaki disease (Schindler's Disease type II)	609242	AR	Syndromic manifestations	Adolescent/Adult	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	1	5	**4**		A
NAGLU	609701	Mucopolysaccharidosis type IIIB (Sanfilippo B)	252920	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		A
NAGS	608300	N-acetylglutamate synthase deficiency	237310	AR	Hyperammonemic crisis	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	3	3	2	3	14	**1**		A
NARS2	612803	Combined oxidative phosphorylation deficiency 24	616239	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		
NBEAL2	614169	Gray Platelet syndrome	139090	AR	Bleeding	Infant	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	2	12	**1**		
NEU1	608272	Sialidosis/ MUCOLIPIDOSIS I	256550	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		A

NEUROD1	601724	MODY, type VI (MODY6)	606394	AD	Diabetes mellitus	Adulthood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	0	0	1	5	**2**		
NEXN	613121	Cardiomyopathy (Familial Hypertrophic 20, Dilated 1CC)	613876	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		C
NF1	613113	Neurofibromatosis, type 1	162200	AD	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Infant	1	2	2	2	3	10	**1**		A
NF2	607379	Neurofibromatosis type 2	101000	AD	Acoustic neuroma and meningiomas	Adulthood	Referral to specialists for surveillance and early management	Adolescent	1	3	1	3	3	11	**3**		A
NFKB2	164012	Immunodeficiency, common variable, 10	615577	AD	Immunodeficiency	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	1	10	**1**		
NHEJ1	611290	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation	611291	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	0	2	3	1	9	**2**		A
NKX2-1	600635	Choreoathetosis, hypothyroidism, and neonatal respiratory distress	610978	AD	Hypothyroidism	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
NKX2-5	600584	Hypothyroidism, congenital nongoitrous, 5	225250	Multiple and/or complex pattern	Thyroid dysgenesis	Variable	Referral to specialists for surveillance and preemptive management	Variable	2	0	3	3	0	8	**2**		B
NKX2-6	611770	Persistent truncus arteriosus / Conotruncal heart defects	217095	AR	Cyanotic heart disease	Neonatal	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	1	10	**1**		
NLRP3	606416	Muckle-Wells syndrome / CINCA syndrome / Familial cold-induced inflammatory syndrome 1	191900	AD	Syndromic manifestations	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	0	3	3	2	10	**1**		
NME8	607421	Ciliary dyskinesia, primary, 6	610852	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	0	8	**2**		C
NODAL	601265	Heterotaxy, Visceral	270100	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	1	2	2	3	1	9	**2**		
NOTCH2	600275	Alagille syndrome 2	610205	AD	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	2	11	**1**		
NPC1	607623	Niemann-Pick Disease, types C1 and D	257220	AR	Neurodegeneration	Childhood	Referral to specialists for surveillance and preemptive management	Infant	2	3	1	3	3	12	**1**		A
NPC2	601015	Niemann-Pick Disease, type C2	607625	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	1	3	2	11	**1**		A
OAT	613349	Gyrate Atrophy of the Choroid and Retina	258870	AR	Progressive chorioretinal degeneration	Childhood	Referral to specialists for surveillance and dietary management	Neonatal	1	3	2	2	3	11	**1**		
OFD1	300170	Orofaciodigital syndrome I / Joubert syndrome 10 / Simpson Golabi-Behmel syndrome, type 2 / ?Retinitis pigmentosa 23	311200	X-linked	Syndromic manifestations	Birth	Referral to specialists for surveillance and supportive care	N / A	1	3	0	0	2	6	**2**		A
OPA1	605290	Optic atrophy plus syndrome / Optic atrophy 1	125250	AD	Communication deficits and visual impairment	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	1	3	2	10	**2**		
OPA3	606580	3-Methylglutaconic Aciduria, type III	258501	AR	Neurologic involvement	Variable	Referral to specialists for surveillance and supportive care	Neonatal	1	3	0	0	2	6	**2**	Secondary	A
OPLAH	614243	5-oxoprolinase deficiency	260005	Multiple and/or complex pattern	Syndromic manifestations	Variable	Referral to specialists for surveillance and supportive care	N / A	0	3	0	0	1	4	**2**		
OSBPL2	606731	Deafness, autosomal dominant 67	616340	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	1	10	**2**		
OTC	300461	Ornithine transcarbamylase deficiency (Females)	311250	X-linked	Hyperammonemic crisis	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	2	3	3	3	13	**1**		
OTC	300461	Ornithine transcarbamylase deficiency (Males)	311250	X-linked	Hyperammonemic crisis	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	3	3	3	2	3	14	**1**		A
OTOA	607038	Deafness, autosomal recessive 22	607039	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
OTOF	603681	Deafness, autosomal recessive 9	601071	AR	Communication deficits and/or auditory neuropathy	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
OTOG	604487	Deafness, autosomal recessive 18B	614945	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
OTOGL	614925	Deafness, autosomal recessive 84B	614944	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
P2RX2	600844	Deafness, autosomal dominant 41	608224	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	3	3	1	11	**2**		
P2RY12	600515	Bleeding disorder, platelet-type, 8	609821	AR	Bleeding	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
PAH	612349	Phenylketonuria	261600	AR	Intellectual disability	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	2	3	13	**1**	Core	A
PALB2	610355	Fanconi anemia, complementation group N	610832	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	1	10	**1**		
PALB2	610355	{Breast cancer, susceptibility to}	114480	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	2	2	3	2	11	**3**		C
PAX3	606597	Waardenburg Syndrome, type 3	148820	Multiple and/or complex pattern	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Infant	1	2	3	3	2	11	**1**		
PAX3	606597	Waardenburg Syndrome, type 1	193500	AD	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
PAX4	167413	MODY, type IX (MODY9)	612225	AD	Diabetes mellitus	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	3	0	0	0	4	**4**		
PAX8	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia	218700	Multiple and/or complex pattern	Thyroid dysgenesis	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	3	3	2	12	**1**		A
PCBD1	126090	Hyperphenylalaninemia, BH4-deficient, D	264070	AR	Hyperphenylalaninemia without cognitive impairment	Infant	Referral to specialists for surveillance and supportive care	N / A	0	3	0	0	1	4	**2**	Secondary	
PCCA	232000	Propionicacidemia	606054	AR	Organic aciduria manifestations	Neonatal	Referral to specialists for surveillance and early management	Neonatal	3	3	1	3	3	13	**1**	Core	A
PCCB	232050	Propionicacidemia	606054	AR	Organic acidemia manifestations	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	1	3	3	13	**1**	Core	A
PCDH15	605514	Deafness, autosomal recessive 23	609533	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		
PCDH15	605514	Usher Syndrome, type 1F	602083	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	3	13	**1**		A
PCSK9	607786	Familial hypercholesterolemia 3	603776	AD	Hypercholesterolemia and cardiovascular disease	Variable	Referral to specialists for surveillance and preemptive management	Adolescent	2	3	3	3	2	13	**1**		B
PDX1	600733	Pancreatic agenesis 1	260370	AR	Neonatal diabetes mellitus	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
PDX1	600733	MODY, type IV (MODY4)	606392	AD	Diabetes mellitus	Adolescence	Referral to specialists for surveillance and preemptive management	Childhood	1	3	0	0	1	5	**2**		
PDZD7 / ADGRV1	612971	Usher Syndrome, type 2C, GPR98/PDZD7 Digenic	605472	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	0	2	3	0	6	**2**		
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger) / Peroxisome biogenesis disorder 1B (NALD/IRD)	234580	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		
PEX6	601498	Peroxisome biogenesis disorder 4A (Zellweger) / Peroxisome biogenesis disorder 4B / Heimler syndrome 2	616617	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
PFKM	610681	Glycogen storage disease VII	232800	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Neonatal	1	2	2	3	1	9	**2**		A
PGAM2	612931	Glycogen storage disease X	261670	AR	Syndromic manifestations	Adolescent	Referral to specialists for surveillance and early management	Infant	1	2	2	3	1	9	**2**		
PHKA2	300798	Glycogen Storage Disease, type IXa1/Xa2	306000	X-linked	Severe hypoglycemic episode	Childhood	Referral to specialists for surveillance and dietary management	Infant	2	2	3	3	2	12	**1**		A
PHKG2	172471	Liver Phosphorylase Kinase Deficiency / Glycogen Storage Disease IXC (GSD9C)	613027	AR	Hypoglycemia and hepatomegaly	Infant	Referral to specialists for surveillance and dietary management	Infant	1	3	3	3	1	11	**1**		A
PHOX2B	603851	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease	209880	AD	Hypoventilation	Birth	Referral to specialists for surveillance and early management	Neonatal	3	3	3	2	3	14	**1**		B
PIK3CD	602839	Immunodeficiency 14	615513	AD	Immunodeficiency	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	2	3	2	3	2	12	**1**		
PKP2	602861	Arrhythmogenic right ventricular cardiomyopathy 9	609040	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**1**		B

PLAU	191840	Quebec Platelet Disorder	601709	AD	Bleeding	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		
PLCG2	600220	Autoinflammation and PLCG2-associated antibody deficiency and immune dysregulation (APLAID)	614878	AD	Blistering skin lesions	Infant	Referral to specialists for surveillance and early management	Infant	1	0	1	2	0	4	**2**		
PLN	172405	Cardiomyopathy (Familial Hypertrophic 18, dilated, 1P)	613874	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		C
PMS2	600259	Mismatch repair cancer syndrome	276300	AR	Hereditary cancer predisposition	Childhood	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	1	2	1	9	**2**		
PMS2	600259	Lynch syndrome	120435	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	2	3	2	3	12	**3**		C
PNP	164050	Immunodeficiency due to purine nucleoside phosphorylase deficiency	613179	AR	Immunodeficiency	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	3	3	1	2	2	11	**2**		
PNPO	603287	Pyridoxamine 5'-phosphate oxidase deficiency	610090	AR	Microcephaly and neurologic involvement	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	3	14	**1**		A
POLD1	174761	{Colorectal cancer, susceptibility to, 10}	612591	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	1	11	**3**		
POLE	174762	{Colorectal cancer, susceptibility to, 12}	615083	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Adulthood	2	3	3	2	1	11	**3**		
POLR1C	610060	Leukodystrophy, hypomyelinating, 11	616494	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	1	6	**2**		
POLR1C	610060	Treacher Collins syndrome 3	248390	AR	Communication deficits	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	0	9	**2**		
POLR1D	613715	Treacher Collins syndrome 2	613717	Multiple and/or complex pattern	Communication deficits	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	1	10	**1**		
POU1F1	173110	Pituitary hormone deficiency, combined, 1	613038	Multiple and/or complex pattern	Pituitary hormone deficiency manifestations	Infant	Referral to specialists for surveillance and preemptive management	Childhood	2	3	2	3	2	12	**1**		A
POU3F4	300039	Deafness, X-linked 2	304400	X-linked	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Birth	1	3	3	3	2	12	**1**		A
POU4F3	602460	Deafness, autosomal dominant 15	602459	AD	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	3	3	2	12	**1**		A
PPT1	600722	Ceroid Lipofuscinosis, neuronal, 1 (CLN1)	256730	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
PRDM16	605557	Cardiomyopathy, dilated 1LL / Left Ventricular Noncompaction 8	615373	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	0	0	2	3	0	5	**2**		C
PRKACG	176893	?Bleeding disorder, platelet-type, 19	616176	AR	Bleeding	Childhood	Referral to specialists for surveillance and preemptive management	Infant	1	3	1	3	0	8	**2**		
PRKAG2	602743	Cardiomyopathy (Familial hypertrophic, 6 / Wolff-Parkinson-White syndrome)	600858	AD	Arrhythmia and syndromic manifestions	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	2	11	**1**		A,B
PRKAG2	602743	Glycogen storage disease of heart, lethal congenital	261740	AD	Arrhythmia	Birth	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		C
PRKDC	600899	SCID	615966	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and supportive care	N / A	3	0	0	0	0	3	**2**		
PROC	612283	Thrombophilia due to protein C deficiency, autosomal recessive	612304	AR	Thrombosis	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	1	2	2	11	**1**		A
PROC	612283	Thrombophilia due to protein C deficiency, autosomal dominant	176860	AD	Thrombosis	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	2	3	2	11	**1**		
PROP1	601538	Pituitary hormone deficiency, combined, 2	262600	AR	Growth failure / failure to thrive	Infant	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	3	13	**1**		A
PROS1	176880	Thrombophilia due to protein S deficiency, autosomal recessive	614514	AR	Thrombosis	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	1	2	1	10	**1**		A
PROS1	176880	Thrombophilia due to protein S deficiency, autosomal dominant	612336	AD	Thrombosis	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	2	3	2	11	**1**		
PRPS1	311850	Deafness, X-linked 1	304500	X-linked	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	1	11	**1**		
PSEN1	104311	Cardiomyopathy, dilated, 1U	613694	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		
PSEN2	600759	Cardiomyopathy, dilated, 1V	613697	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	0	2	3	0	7	**2**		
PTCH1	601309	Basal cell nevus syndrome	109400	AD	Medulloblastoma	Infant	Referral to specialists for surveillance and early management	Infant	2	1	1	3	1	8	**2**		A
PTEN	601728	PTEN hamartoma tumor syndrome	158350	AD	Hereditary cancer predisposition	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	2	3	2	3	2	12	**1**		
PTPN11	176876	LEOPARD syndrome 1 / Noonan syndrome 1 / Leukemia, Juvenile myelomonocytic / Metachondromatosis	151100	AD	Cardiac manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	3	12	**1**		A
PTPRC	151460	Severe Combined Immunodeficiency, T cell-negative, B-cell / Natural Killer-Cell Positive	608971	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	1	12	**1**		
PTPRQ	603317	Deafness, autosomal recessive 84A	613391	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
PTS	612719	Hyperphenylalaninemia, BH4-deficient, A	261640	AR	Hyperphenylalaninemia with cognitive impairment	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**	Secondary	A
PYGL	613741	Glycogen Storage Disease VI	232700	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	0	3	3	3	1	10	**1**		A
PYGM	608455	McArdle Disease	232600	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	2	3	3	12	**1**		
QDPR	612676	Hyperphenylalaninemia, BH4-deficient, C	261630	AR	Hyperphenylalaninemia with cognitive impairment	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**	Secondary	A
RAB23	606144	Carpenter Syndrome	201000	AR	Craniosynostosis	Birth	Referral to specialists for surveillance and early management	Infant	1	3	3	2	2	11	**1**		A
RAD51	179617	?Fanconi anemia, complementation group R	617244	AD	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**		
RAD51C	602774	Fanconi anemia, complementation group O	613390	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**		
RAG1	179615	Severe combined immunodeficiency, B cell-negative	601457	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	3	14	**1**		A
RAG2	179616	Severe combined immunodeficiency, B cell-negative	601457	AR	Immunodeficiency - combined	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	2	2	13	**1**		A
RASA1	139150	Capillary malformation-arteriovenous malformation	608354	AD	Capillary and arteriovenous malformations	Variable	Referral to specialists for surveillance and early management	Adolescent	2	2	2	3	2	11	**1**		A
RASGRP2	605577	?Bleeding disorder, platelet-type, 18	615888	AR	Bleeding	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	1	3	0	8	**2**		
RB1	614041	Retinoblastoma	180200	Multiple and/or complex pattern	Retinoblastoma	Neonatal	Referral to specialists for surveillance and/or preemptive management	Neonatal	2	3	3	2	3	13	**1**		A
RBM20	613171	Cardiomyopathy, dilated, 1DD	613172	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**		B
RDX	179410	Deafness, autosomal recessive 24	611022	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		C
RET	164761	Multiple endocrine neoplasia IIA / Multiple endocrine neoplasia IIB	171400, 162300	AD	Medullary thyroid cancer	Variable	Referral to specialists for surveillance and preemptive management	Childhood	2	3	3	2	3	13	**1**		A
RIT1	609591	Noonan syndrome 8	615355	AD	Hypertrophic cardiomyopathy	Infant	Referral to specialists for surveillance and early management	Infant	2	3	2	2	2	11	**1**		
RPL11	604175	Diamond-Blackfan anemia 7	612562	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		A
RPL15	604174	?Diamond-Blackfan anemia 12	615550	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		
RPL26	603704	?Diamond-Blackfan anemia 11	614900	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		
RPL27	607526	?Diamond-Blackfan anemia 16	617408	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		

RPL31	N/A	Diamond-Blackfan anemia	N/A	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		
RPL35A	180468	Diamond-Blackfan anemia 5	612528	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		C
RPL5	603634	Diamond-Blackfan anemia 6	612561	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		
RPS10	603632	Diamond-Blackfan anemia 9	613308	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		IC
RPS17	180472	Diamond-Blackfan anemia 4	612527	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		A
RPS19	603474	Diamond-Blackfan anemia 1	105650	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	3	12	**1**		A
RPS24	602412	Diamond-blackfan anemia 3	610629	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		A
RPS26	603701	Diamond-Blackfan anemia 10	613309	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	2	11	**1**		A
RPS27	603702	?Diamond-Blackfan anemia 17	617409	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		
RPS28	603685	Diamond Blackfan anemia 15 with mandibulofacial dysostosis	606164	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		
RPS29	603633	Diamond-Blackfan anemia 13	615909	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	1	10	**1**		
RPS7	603658	Diamond-Blackfan anemia 8	612563	AD	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**		C
RSPH1	609314	Ciliary dyskinesia, primary, 24	615481	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		
RSPH3	615876	Ciliary dyskinesia, primary, 32	616481	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		
RSPH4A	612647	Ciliary dyskinesia, primary, 11	612649	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		A
RSPH9	612648	Ciliary dyskinesia, primary, 12	612650	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**		A
RUNX1	151385	Platelet disorder, familial, with associated myeloid malignancy	601399	AD	Myeloid malignancy	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	1	3	2	10	**2**		
RYR1	180901	Malignant hyperthermia susceptibility	145600	AD	Anesthesia-induced malignant hyperthermia	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	2	1	3	3	3	12	**1**		B
RYR2	180902	Arrhythmia (Catecholaminergic polymorphic ventricular tachycardia / Arrhythmogenic Right Ventricular Dysplasia 2)	604772	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Variable	3	2	2	2	2	11	**1**		A
S1PR2	605111	Deafness, autosomal recessive 68	610419	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		
SCN1B	600235	Brugada Syndrome 5 / Cardiac conduction defect, nonspecific	612838	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	2	11	**1**		C
SCN3B	608214	Brugada Syndrome 7 / Atrial fibrillation, familial, 16	613120	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	1	7	**2**		C
SCN4A	603967	Hyperkalemic periodic paralysis, type 2	170500	AD	Episodic weakness and progressive myopathy	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		C
SCN4B	608256	Long QT syndrome-10	611819	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	2	1	10	**1**		C
SCN5A	600163	Arrhythmia, Cardiomyopathy (Romano-Ward Long QT syndrome 3 / Brugada Syndrome / Dilated, IE)	603830	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and preemptive management	Neonatal	3	2	2	2	3	12	**1**		B
SCNN1A	600228	Pseudohypoaldosteronism, type I	264350	AR	Hyponatremia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	3	15	**1**		A
SCNN1A	600228	Bronchiectasis with or without elevated sweat chloride 2	613021	AD	Bronchiectasis and chronic bronchitis	Variable	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		
SCNN1B	600760	Pseudohypoaldosteronism, type I	264350	AR	Hyponatremia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	2	14	**1**		A
SCNN1B	600760	Liddle Syndrome	177200	AD	Syndromic manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
SCNN1G	600761	Pseudohypoaldosteronism, type I	264350	AR	Hyponatremia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	2	14	**1**		C
SCNN1G	600761	Liddle Syndrome	177200	AD	Syndromic manifestations	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
SDHA	600857	Paragangliomas 5	614165	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	1	3	2	2	0	8	**2**		
SDHAF2	613019	Hereditary Paraganglioma-Pheochromocytoma Syndrome 2	601650	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and early management	Childhood	1	3	2	2	0	8	**2**		B
SDHB	185470	Hereditary Paraganglioma-Pheochromocytoma Syndrome 4	115310	AD	Malignant pheochromocytoma / paraganglioma	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	2	3	3	2	3	13	**1**		B
SDHC	602413	Hereditary Paraganglioma-Pheochromocytoma Syndrome 3	605373	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and early management	Childhood	1	3	2	2	1	9	**1**		B
SDHD	602690	Hereditary Paraganglioma-Pheochromocytoma Syndrome 1	168000	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and early management	Childhood	1	3	2	2	2	10	**1**		A
SERPINA1	107400	Emphysema-cirrhosis, due to AAT deficiency	613490	AR	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and early management	Adulthood	2	3	1	3	3	12	**3**		C
SERPINB6	173321	?Deafness, Autosomal Recessive 91	613453	AR	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	0	3	3	3	0	9	**3**		C
SERPINC1	107300	Thrombophilia due to antithrombin III deficiency	613118	Multiple and/or complex pattern	Thrombosis	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	2	3	2	11	**1**		C
SFTPA1	178630	{Pulmonary fibrosis, idiopathic, susceptibility to} / Pulmonary fibrosis	178500	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	0	2	**4**		
SFTPA2	178642	Pulmonary Fibrosis	178500	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	1	3	**4**		C
SFTPB	178640	Surfactant metabolism dysfunction, pulmonary, 1	265120	AR	Respiratory distress / failure	Birth	Referral to specialists for surveillance and early management	Neonatal	3	3	1	1	2	10	**2**		A
SFTPC	178620	Surfactant metabolism dysfunction, pulmonary, 2	610913	AD	Respiratory distress / failure	Variable	Referral to specialists for surveillance and early management	Variable	1	3	2	2	2	10	**2**		C
SFTPD	178635	Pulmonary Fibrosis, Surfactant Dysfunction	178635	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	0	2	**4**		
SGCD	601411	Cardiomyopathy, dilated, 1L	606685	AD	Arrhythmia and severe cardiac events	Adolecent	Referral to specialists for surveillance and early management	Neonatal	0	0	2	3	0	5	**2**		C
SGSH	605270	Mucopolysaccharidisis type IIIA (Sanfilippo A)	252900	AR	Neurodegeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	2	7	**2**		A
SIX1	601205	Branchio-oto-renal related disorders	608389	AD	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
SLC17A5	604322	Sialic acid storage disorder, infantile	269920	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	2	8	**2**		A
SLC17A8	607557	Deafness, autosomal dominant 25	605583	AD	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	0	3	3	3	1	10	**2**		
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	249270	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Childhood	1	3	3	3	2	12	**1**		A
SLC19A3	606152	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2)	607483	AR	Encephalopathy	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
SLC22A4	604190	Deafness, autosomal recessive	N/A	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**2**		
SLC22A5	603377	Carnitine deficiency, systemic primary	212140	AR	Metabolic decompensation	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	1	3	3	2	10	**1**	Core	A
SLC25A13	603859	Citrullinemia, type II, neonatal-onset	605814	AR	Failure to thrive and cirrhosis	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	1	2	2	3	2	10	**1**	Secondary	A

SLC25A13	603859	Citrullinemia, type II, adult-onset	603471	AR	Liver failure	Adulthood	Referral to specialists for surveillance and supportive care	N / A	2	0	0	0	1	3	**4**		
SLC25A15	603861	Hyperornithinemia-Hyperammonemia-Homocitrullinemia Syndrome	238970	AR	Neurocognitive deficits	Variable	Referral to specialists for surveillance and dietary management	Variable	1	3	2	2	2	10	**1**		A
SLC25A20	212138	Carnitine-Acylcarnitine Translocase Deficiency	212138	AR	Hypoglycemia	Neonatal	Referral to specialists for surveillance and dietary management	Neonatal	2	3	3	3	2	13	**1**	Secondary	A
SLC26A4	605646	Pendred syndrome / Deafness, autosomal recessive 4, with enlarged vestibular aqueduct;	600791	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
SLC2A1	138140	Dystonia 9, GLUT1 deficiency syndromes 1 and 2, autosomal recessive	606777	AR	Dystonia and syndromic manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	2	11	**1**		
SLC2A1	138140	Dystonia 9, GLUT1 deficiency syndromes 1 and 2, autosomal dominant	612126	AD	Dystonia and syndromic manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	2	11	**1**		A
SLC2A9	606142	Hypouricemia, renal, 2	612076	AR	Hypouricemia and exercise-induced acute renal failure	Adolescent	Referral to specialists for surveillance and early management	Adolescent	1	2	2	3	2	10	**1**		
SLC37A4	602671	Glycogen Storage Disease Ib/Ic	232220	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Infant	2	3	3	2	3	13	**1**		A
SLC39A4	607059	Acrodermatitis enteropathica	201100	AR	Syndromic manifestations	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
SLC3A1	104614	Cystinuria	220100	Multiple and/or complex pattern	Renal calculi	Childhood	Referral to specialists for surveillance, early management	Infant	1	3	1	3	2	10	**2**		A
SLC40A1	604653	Hemochromatosis, type 4	606069	AD	Iron overload	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	1	2	2	3	1	9	**3**		
SLC46A1	611672	Folate malabsorption, hereditary	229050	AR	Anemia and hypogammaglobulinemia	Infant	Referral to specialists for surveillance and dietary management	Neonatal	2	3	2	2	2	11	**1**		A
SLC5A5	601843	Thyroid dyshormonogenesis 1	274400	AR	Hypothyroidism	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		A
SLC6A8	300036	Cerebral creatine deficiency syndrome 1, X-linked	300352	X-linked	Intellectual disability and seizures	Infant	Referral to specialists for surveillance and supportive care	Neonatal	1	3	0	0	2	6	**2**		A
SLC7A7	603593	Lysinuric protein intolerance	222700	AR	Gastrointestinal intolerance	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	2	3	3	12	**1**		A
SLC7A9	604144	Cystinuria	220100	Multiple and/or complex pattern	Renal calculi	Childhood	Referral to specialists for surveillance, early management	Infant	1	3	1	3	2	10	**2**		A
SLITRK6	609681	Deafness and myopia	221200	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	2	12	**1**		
SLX4	613278	Fanconi anemia, complementation group P	613951	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**		
SMAD3	603109	Loeys-Dietz Syndrome 3	613795	AD	Aortic dissection	Adulthood	Referral to specialists for surveillance and early management	Childhood	2	3	2	3	2	12	**1**		A
SMAD4	600993	Juvenile polyposis / hereditary hemorrhagic telangiectasia syndrome	175050	AD	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Infant	2	3	2	2	3	12	**1**		A
SMAD9	603295	Pulmonary hypertension, primary, 2	615342	AD	Pulmonary hypertension	Variable	Referral to specialists for surveillance and early management	Childhood	2	0	1	2	0	5	**2**		C
SMPD1	607608	Niemann-Pick Disease, Type B	607616	AR	Hepatomegaly and respiratory infection	Childhood	Referral to specialists for surveillance and supportive care	N / A	2	3	0	0	3	8	**2**		A
SMPD1	607608	Niemann-Pick Disease, Type A	257200	AR	Neurodegeneration	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**		A
SMPX	300226	Deafness, X-Linked 4 (Affected hemizygous males and homozygous females)	300066	X-linked	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Infant	1	3	3	3	1	11	**1**		A
SMPX	300226	Deafness, X-Linked 4 (Carrier Females)	300066	X-linked	Communication deficits	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	0	3	3	1	8	**2**		
SNAI2	602150	Waardenburg Syndrome, type 2D	608890	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	0	10	**1**		
SNTA1	601017	Long QT syndrome 12	612955	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	0	2	2	0	6	**2**		B
SOX10	602229	Waardenburg syndrome, type 4C / type 2E	613266	AD	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		A
SOX10	602229	PCWH Syndrome	609136	AD	Neurologic involvement	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	1	3	1	10	**2**		
SPAG1	603395	Ciliary dyskinesia, primary, 28	615505	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**		
SPR	182125	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency	612716	AR	Dystonia	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
SRY	480000	46XY sex reversal 1	400044	Y-linked	Gonadoblastoma	Childhood	Referral to specialists for surveillance and early management	Infant	2	2	3	2	3	12	**1**		
STAR	600617	Lipoid adrenal hyperplasia	201710	AR	Salt-wasting crises	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	3	3	3	3	2	14	**1**		
STAT3	102582	Atopic & eosinophilic disease, Hyper-IgE recurrent infection syndrome	147060	AD	Sinopulmonary infections and pneumatocele formation	Infant	Referral to specialists for surveillance and preemptive management	Infant	2	3	2	2	3	12	**1**		A
STK11	602216	Peutz-Jeghers Syndrome	175200	AD	Gastrointestinal Cancer	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	2	3	2	2	3	12	**1**		A
STRC	606440	Deafness, autosomal recessive 16	603720	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
SUCLA2	603921	Mitochondrial DNA Depletion Syndrome 5 (Encephalomyopathic with or without Methylmalonic Aciduria)	612073	AR	Encephalomyopathy	Infant	Referral to specialists for surveillance and supportive care	Neonatal	2	3	0	0	2	7	**2**		A
SUCLG1	611224	Mitochondrial DNA Depletion Syndrome 9 (Encephalomyopathic type with Methylmalonic Aciduria)	245400	AR	Encephalomyopathy	Infant	Referral to specialists for surveillance and supportive care	Neonatal	3	3	0	0	1	7	**2**		A
SUMF1	607939	Multiple Sulfatase Deficiency	272200	AR	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	1	7	**2**		
SYNE4	615535	Deafness, autosomal recessive 76	615540	AR	Communication deficits	Birth - Childhood	Referral to specialists for surveillance and preemptive management	Variable	1	3	3	3	1	11	**2**		C
TAT	613018	Tyrosinemia, type II	276600	AR	Tyrosinemia manifestations	Infant	Referral to specialists for surveillance and dietary management	Neonatal	1	3	3	2	2	11	**1**	Secondary	A
TAZ	300394	Barth Syndrome	302060	X-linked	Arrhythmia and severe cardiac events	Birth	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**	Secondary	A
TBC1D24	613577	Deafness, autosomal recessive 86	614617	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	1	11	**1**		A
TBC1D24	613577	Deafness, autosomal dominant 65	616044	AD	Communication deficits	Adulthood	Referral to specialists for surveillance and preemptive management	Adulthood	0	3	3	3	1	10	**4**		
TBX19	604614	Adrenocorticotropic hormone deficiency	201400	AR	Hypoglycemia	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**		
TCAP	604488	Cardiomyopathy, Familial Hypertrophic, 25	607487	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	1	11	**1**		A
TCN2	613441	Transcobalamin II deficiency	275350	AR	Failure to thrive	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	2	3	3	2	11	**1**		
TCOF1	606847	Treacher Collins syndrome 1	154500	AD	Communication deficits	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	2	11	**1**		A
TECTA	602574	Deafness, autosomal recessive 21 (DFNB21)	603629	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**		A
TECTA	602574	Deafness, autosomal dominant 8/12 (DFNA12)	601543	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	3	13	**1**		
TERC	602322	Dyskeratosis congenita, autosomal dominant 1 / Pulmonary fibrosis	127550	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	2	1	1	1	7	**2**		B
TERT	187270	{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1} / Pulmonary Fibrosis	614742	AD	Pulmonary fibrosis and pneumonia	Adulthood	Referral to specialists for surveillance and supportive care	Adulthood	1	2	0	0	1	4	**4**		B
TFR2	604720	Hemochromatosis, type 3	604250	AR	Iron overload	Adulthood	Referral to specialists for surveillance and preemptive management	Adolescent	1	3	3	3	2	12	**1**		C
TG	188450	Thyroid dyshormonogenesis 3	274700	AR	Hypothyroidism	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	3	14	**1**		A

TGFB2	190220	Loeys-Dietz Syndrome 4	614816	AD	Aortic dissection	Adulthood	Referral to specialists for surveillance and early management	Childhood	2	3	2	3	1	11	**1**	
TGFB3	190230	Arrhythmogenic right ventricular cardiomyopathy 1	107970	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	0	2	2	0	6	**2**	C
TGFB3	190230	Loeys-Dietz Syndrome 5	107970	AD	Arrhythmia and severe cardiac events	Adolescent	Referral to specialists for surveillance and early management	Childhood	2	2	3	3	2	12	**1**	
TGFBR1	190181	Loeys-Dietz Syndrome 1	609192	AD	Aortic dissection	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	1	2	3	11	**1**	
TGFBR2	190182	Loeys-Dietz Syndrome 2	610168	AD	Aortic dissection	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	1	2	3	11	**1**	A
TH	191290	Segawa syndrome, recessive	605407	AR	Intellectual disability and psychomotor degeneration	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**	A
THRA	190120	Hypothyroidism, congenital, nongoitrous, 6	614450	AD	Congenital hypothyroidism	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	0	9	**1**	A
TIMM8A	300356	Mohr-Tranebjaerg syndrome / Dystonia-deafness syndrome	304700	X-linked	Syndromic manifestations	Infant	Referral to specialists for surveillance and supportive care	Neonatal	2	3	0	0	2	7	**2**	A
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3 / Revesz syndrome	613990, 268130	AD	Bone marrow failure	Variable	Referral to specialists for surveillance and early management	Variable	2	3	1	2	2	10	**2**	B
TMC1	606706	Deafness, Autosomal Recessive 7	600974	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**	A
TMC1	606706	Deafness, Autosomal Dominant 36	606705	AD	Communication deficits	Childhood	Referral to specialists for surveillance and preemptive management	Childhood	1	3	3	3	1	11	**2**	
TMEM127	613403	{Pheochromocytoma, susceptibility to}	171300	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	1	3	2	2	0	8	**2**	
TMEM43	612048	Arrhythmogenic right ventricular cardiomyopathy 5	604400	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**1**	A
TMIE	607237	Deafness, autosomal recessive 6	600971	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**	A
TMPO	188380	?Cardiomyopathy, dilated, 1T	115200	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	0	0	2	3	0	5	**2**	C
TMPRSS3	605511	Deafness, autosomal recessive 8/10	601072	AR	Communication deficits	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**	A
TNNC1	191040	Cardiomyopathy (Hypertrophic 13, dilated, 1Z)	613243	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**	B
TNNI3	191044	Cardiomyopathy (Hypertrophic 7, dilated 1FF, ?dilated 2A)	613690	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**	B
TNNT2	191045	Cardiomyopathy (Familial hypertrophic 2, Dilated 1D)	115195	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	3	13	**1**	B
TP53	191170	Li-Fraumeni syndrome	151623	AD	Syndromic manifestations	Adolescent	Referral to specialists for surveillance and early management	Childhood	2	3	2	2	2	11	**1**	A
TPM1	191010	Cardiomyopathy (Hypertrophic 3, Dilated IY)	115196	AD	Arrhythmia and severe cardiac events	Variable	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	2	12	**1**	B
TPO	606765	Thyroid dyshormonogenesis 2A	274500	AR	Hypothyroidism	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	3	14	**1**	A
TPP1	607998	Ceroid lipofuscinosis, neuronal, 2 (CLN2)	204500	AR	Neurodegeneration and retinal degeneration	Childhood	Referral to specialists for surveillance and supportive care	N / A	3	3	0	0	3	9	**2**	A
TPRN	613354	Deafness, autosomal recessive 79	613307	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**	C
TRDN	603283	Ventricular tachycardia, catecholaminergic polymorphic, 2	615441	AR	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	2	2	2	1	10	**1**	C
TRHR	188545	Thyrotropin-releasing hormone resistance, generalized	188545	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	0	3	3	3	0	9	**2**	C
TRIOBP	609761	Deafness, autosomal recessive 28	609823	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	2	12	**1**	A
TSC1	605284	Tuberous sclerosis-1	191100	AD	Syndromic manifestations	Infant	Referral to specialists for surveillance and early management	Neonatal	2	3	1	3	3	12	**1**	A
TSC2	191092	Tuberous sclerosis-2	613254	AD	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	2	3	1	3	3	12	**1**	A
TSHB	188540	Hypothyroidism, congenital, nongoitrous 4	275100	AR	Intellectual disability and growth delay	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	3	13	**1**	A
TSHR	603372	Hypothyroidism, congenital, nongoitrous, 1	275200	AR	Intellectual disability and growth delay	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	2	3	3	12	**1**	A
TSPEAR	612920	Deafness, autosomal recessive 98	614861	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	0	10	**2**	C
TSR2	300945	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis	300946	X-linked	Anemia	Infant	Referral to specialists for surveillance and early management	Infant	1	3	2	3	0	9	**2**	
TTC25	617095	Ciliary dyskinesia, primary, 35	617092	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	1	9	**1**	
TTN	188840	Cardiomyopathy (Familial Hypertrophic 9, dilated, 1G) and other myopathies	613765	Multiple and/or complex pattern	Arrhythmia and severe cardiac events	Infant	Referral to specialists for surveillance and early management	Neonatal	2	2	2	3	2	11	**1**	A,B
TTPA	600415	Ataxia with isolated vitamin E deficiency	277460	AR	Ataxia - progressive	Childhood	Referral to specialists for surveillance and dietary management	Infant	1	3	3	3	3	13	**1**	A
TWNK / C10orf2	606075	Perrault syndrome 5	616138	AR	Syndromic manifestations	Childhood	Referral to specialists for surveillance and early management	Childhood	1	3	2	3	0	9	**2**	
UBE2T	610538	Fanconi anemia, complementation group T	616435	AR	Bone marrow dysfunction and multi-system disease	Childhood	Referral to specialists for surveillance and early management	Infant	2	3	2	2	0	9	**2**	
UNC13D	608897	Hemophagocytic lymphohistiocytosis, familial, 3	608898	AR	Severe inflammation and immune dysregulation	Infant	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	2	3	12	**1**	A
USH1C	605242	Deafness, autosomal recessive 18A	602092	AR	Communication deficits	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	3	3	0	10	**1**	
USH1C	605242	Usher Syndrome, type 1C	276904	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	3	13	**1**	A
USH1G	607696	Usher Syndrome, type 1G	606943	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	2	3	2	12	**1**	A
USH2A	608400	Usher Syndrome, type 2A	276901	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	3	12	**1**	A
VCL	193065	Cardiomyopathy (Hypertrophic, 15, dilated, 1W)	613255	AD	Arrhythmia and severe cardiac events	Adulthood	Referral to specialists for surveillance and early management	Neonatal	2	3	2	3	1	11	**1**	B
VHL	608537	von Hippel-Lindau syndrome	193300	AD	Syndromic manifestations	Adulthood	Referral to specialists for surveillance and/or preemptive management	Childhood	2	3	1	3	3	12	**1**	A
VWF	613160	von Willebrand disease, types 2A, 2B, 2M, and 2N	613554	Multiple and/or complex pattern	Bleeding - mild to moderate mucocutaneous	Variable	Referral to specialists for surveillance and preemptive management	Variable	1	2	3	3	3	12	**1**	
VWF	613160	von Willibrand disease, type 3	277480	AR	Bleeding - severe mucocutaneous and musculoskeletal	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	2	3	3	3	2	13	**1**	
VWF	613160	von Willebrand disease, type 1	193400	AD	Bleeding - mild mucocutaneous	Variable	Referral to specialists for surveillance and preemptive management	Variable	0	2	3	3	3	11	**1**	B
WFS1	606201	Wolfram Syndrome	222300	AR	Syndromic manifestations	Birth / Childhood	Referral to specialists for surveillance and early management	Neonatal	1	3	1	3	3	11	**2**	A
WFS1	606201	Deafness, autosomal dominant 6/14/38	600965	AD	Communication deficits	Adolescent	Referral to specialists for surveillance and preemptive management	Childhood	0	3	3	3	2	11	**2**	
WHRN	607928	Usher Syndrome, type 2D	611383	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and preemptive management	Neonatal	1	3	2	3	1	10	**1**	
WT1	607102	WT1-related Wilms	194070	Multiple and/or complex pattern	Wilms tumor	Neonatal	Referral to specialists for surveillance and preemptive management	Neonatal	2	2	3	3	2	12	**1**	B
ZAP70	176947	Selective T-cell defect	269840	AR	Recurrent infection	Infant	Referral to specialists for surveillance and preemptive management	Infant	3	3	3	3	2	14	**1**	A
ZMYND10	607070	Ciliary Dyskinesia, Primary, 22	615444	AR	Syndromic manifestations	Birth	Referral to specialists for surveillance and early management	Neonatal	1	3	2	2	2	10	**1**	