

Inborn errors of immunity present with neuropsychiatric symptoms overlapping with autistic behavioral symptoms

Harumi Jyonouchi^{1,2}

¹Pediatrics, Saint Peter's University Hospital, New Brunswick, NJ 08901, USA.

²Pediatrics, Rutgers University-Robert Wood Johnson, School of Medicine, New Brunswick, NJ 08901, USA.

Correspondence to: Dr. Harumi Jyonouchi, Pediatrics, Saint Peter's University Hospital, 254 10 Easton Ave., New Brunswick, NJ 08901, USA. E-mail:

hjyonouchi@saintpetersuh.com

Abbreviations used:

APDS, activated PI3K delta syndrome

ASD, autism spectrum disorder

BBB, blood-brain barrier

CAPS, cryopyrin-associated periodic fever syndrome

CHARGE, coloboma, heart defect, atresia choanae, retardation of growth and development, genital hypoplasia, and ear anomalies/deafness

CHD7, chromodomain helicase DNA binding domain protein 7

COVID-19, coronavirus disease-2019

DAMPs, danger-associated molecular patterns

DGS, DiGeorge syndrome

EUA, emergency use authorization

FMF, familial Mediterranean fever

GOF, gain of function

HD, high dose

HLH, hemophagocytic lymphohistiocytosis

HSCT, hematopoietic stem cell transplant

IEI, Inborn errors of immunity

IFNs, interferons

IL-1 β , interleukin-1 β

IL-1Ra, IL-1 receptor antagonist

IVIg, intravenous immunoglobulin

JAK, Janus kinase
KS, kabuki syndrome
LCR, low copy repeats
lncRNAs, long non-coding RNAs
LOF, loss of function
MAS, macrophage activation syndrome
MIA, maternal immune activation
miRNA, microRNA
MS, multiple sclerosis
MSK, musculoskeletal
mTOR, mammalian target of rapamycin
MWS, Muckle Wells syndrome
NFAT, nuclear factor of activated T cells
NOMID, Neonatal-onset multisystem inflammatory disease
PAMPs, pathogen-associated molecular patterns.
PIP3, phosphatidylinositol 2,3,4 trisphosphate
PJP, pneumocystis jirovecii pneumonia
PHTS, PTEN hamartoma tumor syndrome
PTEN, phosphatase and tensin homolog
ROS, reactive oxygen species
SCIDS, severe combined immunodeficiency syndrome
STAT, signal transducers and activators of the transcription
TCR, T cell receptor
Th, T-helper
TMP-SMX, trimethoprim-sulfamethoxazole
TNF- α , tumor necrosis factor- α
VEXAS, vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic