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Supplementary information

Neurodevelopmental comorbidities in juvenile systemic autoimmune and autoinflammatory diseases

In the format provided by the authors and unedited

Supplementary Table 1:
Juvenile autoimmune and autoinflammatory diseases associated with neurological features, neuroinflammation and/or neurodevelopment disorders.

Juvenile auto-immune and auto-inflammatory diseases (OMIM if relevant)	Implicated Gene(s)	Other clinical features	Neurologic features - Neuroinflammation	NDD	Ref
Auto-inflammation					
Type I interferonopathies					
AGS1 (225750), chilblain lupus (610448), SLE (152700)	*TREX1*	Chilblain lupus, SLE	AGS - most severe	Global developmental delay	[1]
AGS4 (610333)	*RNASEH2A*		AGS	Global developmental delay	[1]
AGS2, spastic paraparesis (610181)	*RNASEH2B*	Chilblain lupus, SLE	AGS - least severe, jNPSLE	Global developmental delay	[1,2]
AGS3 (610329)	*RNASEH2C*		AGS	Global developmental delay	[1]
AGS5, cerebrovascular disease (612952), chilblain lupus (614415)	*SAMHD1*	Chilblain lupus	AGS, cerebrovascular disease (Moya-Moya)	Global developmental delay	[1]
AGS6, bilateral striatal necrosis (615010), dyschromatosis symmetrica hereditaria (127400)	*ADAR1*	Dyschromatosis symmetrica hereditaria	AGS, bilateral striatal necrosis	Global developmental delay	[1]
AGS7, spastic paraparesis, NMO (615846), SMS (182250)	*MDA5*	SMS	AGS, spastic paraparesis, NMO	Global developmental delay	[1]
Ataxia telangiectasia (208900)	*ATM*	Telangiectasia, primary immunodeficiency	Ataxia, eye movement abnormalities, movement disorders	Mild to moderate developmental delay, cognitive impairment	[1,3,4]
AGS-like, bilateral striatal necrosis (610316)	*PNPT1*		AGS, bilateral striatal necrosis, some Leigh syndrome, Ataxia, seizures, neuropathy, hearing-loss, nystagmus, hypotonia	Global developmental delay	[1,5]
ATAD3A deficiency (617183)	*ATAD3A*	Systemic sclerosis (SSc), vitiligo, hypertrophic cardiomyopathy	Spastic paraparesis, dystonia, elevated CSF neopterins, peripheral neuropathy	Global development delay	[1,6]
AGS8 (619486)	*LSM11*		AGS	Global development delay	[1]
AGS9 (619487)	*U7 (RNU7-1)*		AGS	Global development delay	[1]
ARF1 deficiency (103180)	*ARF1*	Chilblain lupus	AGS-like, periventricular nodular heterotopia (PVNH)	Global development delay, delays in motor skills and speech, oral-motor coordination difficulties	[1]
STAT2 loss of negative regulation (600556)	*STAT2*	Severe fatal early-onset autoinflammation	AGS-like, spastic paraparesis / dystonic tetraparesis	Global development delay	[1,7,8]
Immunodeficiency with basal ganglia calcification (616126)	*ISG15*	MSMD	Seizures and ICC	Global development delay	[1]
USP18 deficiency (617397)	*USP18*	Autoinflammation and mycobacterial disease	AGS-like / TORCH-like syndrome, seizures, encephalopathy, polymicrogyria	Global developmental delay	[1,8,9]
Trisomy 21	Trisomy of chromosome 21	Auto-immune predisposition	Down syndrome, ICC, Regression syndrome with catatonia	Global development delay, Regression syndrome	[1]
TLR7 GOF (300365)	*TLR7*	(j)SLE	NMO, ICC, NPSLE, seizures	Mild motor developmental delay	[1,10]
Leukoencephalopathy cystic without megalencephaly (612951)	*RNASET2*	Autoinflammation	AGS-like, TORCH pseudo-CMV disease, encephalopathy	Global developmental delay	[1]
DADA2 (615688)*	*ADA2*	Autoinflammation, vasculitis, cytopenias, immunodeficiency	Cerebrovasculitis with strokes	Neurocognitive disorders, ASD, ADHD	[1,11,12]
SPENCD (607944)	*ACP5*	SLE, short stature, thrombocytopenia and AIHA	ICC, spastic paraparesis	Mild to moderate developmental delay	[1,13,14]
Complement deficiencies** (613652, 620321, 620322)	*C1QA, C1QB, C1QC*	jSLE, susceptibility to encapsulated organisms/immunodeficiency	jNPSLE, seizures, AGS-like, cerebrovascular disease	Global developmental delay	[1,15]
Complement deficiencies** (216950)	*C1r*	jSLE, susceptibility to encapsulated organisms/immunodeficiency	jNPSLE	NA	[1,16]
Complement deficiencies** (614380)	*C4A*	jSLE, susceptibility to encapsulated organisms/immunodeficiency	Cerebral vasculitis, seizures	NA	[1,17]
PRAAS1 (256040)	*PSMB8*	Contractures, panniculitis, recurrent fever	ICC, aseptic meningitis	NA	[8,18]
PRAAS6 (617591)	*PSMB9*	Skin rash, recurrent fever, anaemia, elevated LFTs, myositis (elevated CK), pulmonary hypertension	Seizures and ICC	NA	[19]

PRAAS2 / PRAID (618048)	*POMP*	Chronic atypical neutrophilic dermatitis with lipodystrophy, ILD	Seizures	NA	20
Stankiewicz-Isidor syndrome (617516)	*PSMD12*	Urticarial skin rash, autoinflammation		Global developmental delay, ASD	8,21,22
PTPN1 haploinsufficiency	*PTPN1*	Raised LFTs, recurrent fever	Subacute spastic dystonia, encephalopathy	Rare mild motor delay, regression	23
DNASE II deficiency (619858)	*DNASE2*	Recurrent fevers, neonatal HSM and pancytopenia	NA	Global developmental delay (learning difficulties)	24
Inflammasomopathies					
FMF (249100)	*MEFV*	FMF	Headaches, seizures, peripheral neuropathy, demyelinating disorders, cerebrovascular disease	Inattention, hyperactivity, ADHD, specific learning disorders	25–27
PAANI	*MEFV* (E583A)	Autoinflammation (fever, chest/abdominal/muscle pain, orchitis)	Seizures, aseptic meningitis	NA	28,29
CAPS (607115)	*NLRP3*	Urticarial rash, hearing loss, arthritis	Chronic aseptic meningitis, headaches, hearing loss, visual impairment	Severe intellectual disability	30,31
Majeed syndrome (609628)	*LPIN2*	CRMO, anaemia, failure to thrive	Irritability	Delayed motor milestones	32
MKD (260920, 610377)	*MVK*	Periodic fever, gastro-intestinal features, MKD	Headaches, cerebellar ataxia, seizures, allodynia, cerebral atrophy, paralysis, seizures, stroke, tremor, and meningitis	Mental or motor retardation, language disorder	33,34
NLRP12-related AID or FCAS2 (611762)	*NLRP12*	Nonpruritic urticaria, arthritis, fever	Headaches, sensorineural hearing loss	NA	35
DIRA (612852)	*IL1RN*	Neonatal CRMO, periostitis, pustulosis	Aseptic meningitis, sensorineural deafness	Global developmental delay	36
NLRC4 associated AID or FCAS4 (616115, 616050)	*NLRC4*	MAS, urticarial rash / FCAS	Aseptic meningitis, sensorineural deafness	Brain atrophy, mental retardation	37
H syndrome (602782)	*SLC29A3*	Hyperpigmentation, hypertrichosis, HSM, heart anomalies, hypogonadism, low height, diabetes mellitus, recurrent fever	Sensorineural deafness	Global developmental delay	38
X-linked proliferative disease 2 (300079)	*XIAP*	EBV infection, splenomegaly, lymphoproliferation, HLH, colitis, IBD, hepatitis	CNS vasculitis	Dementia	8,39,40
NF-kB-pathies					
HA20*** (616744)	*TNFAIP3*	Bipolar aphtous ulceration, Behcet's disease-like, uveitis, joint pain, jSLE-like	Neuro-Behçet's disease, CNS vasculitis, seizures	Global development delay	41,42
RAID or Rela haploinsufficiency*** (618287)	*RelA*	IBD-like, Behcet's disease-like, JIA-like and jSLE-like	NMO	NA	8,43
Blau Syndrome (186580)****	*NOD2*	Blau syndrome, early onset sarcoidosis	Granulomatous CNS involvement	NA	44
TBK1 deficiency (620880)	*TBK1*	Chronic systemic autoinflammation, polyarthritis, vasculitis, rash	Seizures	Global development delay (delayed cognitive development)	45
ROSAH syndrome (614979)	*ALPK1*	Retinal dystrophy, splenomegaly, anhidrosis, recurrent fever, arthritis, colitis, dental abnormalities, uveitis	Optic nerve oedema, headache, migraine, premature CNS mineralisation, Focal meningeal enhancement, neuro-Behçet's disease-like, multiple sclerosis-like	NA	8,46
NEMO-NDAS (301081)	*IKBKG*	Recurrent fever, skin rash, systemic autoinflammation, infection, panniculitis, uveitis, HSM, ectodermal dysplasia	CNS involvement (CNS bleed)	NA	8
SOCS1 haploinsufficiency 619375)	*SOCS1*	Early-onset autoimmunity, cytopenia, HSM, SLE, arthritis	Seizures, limbic encephalitis, MS	Delayed psychomotor development	47
Actin Cytoskeletonopathies					
CDC42 deficiency / NOCARH syndrome (616737) ***	*CDC42*	Pancytopenia, fever, rash, hepatosplenomegaly, systemic inflammation, myelofibrosis, HLH, enterocolitis, failure to thrive	CNS inflammatory disease	Neurodevelopmental delay	8,48,49
Others					
DADA2* (607575)	*CECR1*	Autoinflammation, vasculitis, cytopenias, immunodeficiency	Cerebrovasculitis with strokes	Neurocognitive disorders, ASD, ADHD	8,11
SIFD (612907)	*TRNT1*	Congenital sideroblastic anaemia	Seizures, sensorineural hearing loss, CNS malformations	Global development delay	8,50

VEXAS (NA)	*UBA1*	Fatigue, recurrent fever, neutrophilic dermatosis, chondritis, myelodysplastic syndrome	6-7,8% CNS manifestations (optic neuritis, aseptic meningitis, inflammatory pseudotumor, CNS vasculitis and rhombencephalitis, PRES)	NA	51,52
Prolidase deficiency (613230)	*PEPD*	Ulcerative skin disease, telangiectasia, HSM, recurrent fever, MAS	Seizures, neuropathy, WM abnormalities	Variable global development delay	53
NASA (607676)	*IRAK4*	Recurrent fever, splenomegaly, elevated inflammatory markers, hypochromic microcytic anaemia	Seizures, suspected meningoencephalitis, elevated CSF IL-6	NA	8
Autoimmunity					
jSLE					
Multifactorial jSLE	None	jSLE	jNPSLE	Learning disabilities	54
jSLE/Complement deficiencies**	See above				
X-linked jSLE (NA)	*SAT1*	jSLE	jNPSLE	NA	55
TLR7 GOF (300365)	*TLR7*	(j)SLE	NMO, ICC, jNPSLE, seizures	Mild motor developmental delay	1,10
NFkB-pathies***	See above				
JDM					
NXP2 JDM	None	JDM	Seizures, CNS vasculitis	NA	56
JIA					
JIA (different subtypes)	None		Exceptional: Seizures, headaches, cerebellitis, cerebral vasculitis, aseptic meningitis, NMDA encephalitis	ADHD, cognitive deficits	54
Vasculitis					
KD - MIS-C	None (*OAS1-RNASE L*)	Kawasaki disease / MIS-C	Acute aseptic meningitis, headaches, irritability, confusion, cerebellitis	Memory impairment, cognitive impairment	57,58
Multifactorial juvenile Behçet's disease	None	Bipolar ulcerations, uveitis,	Neuro-Behçet's disease	Development delay secondary to inflammation	59,60
Multifactorial vasculitis	None	Systemic vasculitis	CNS vasculitis		61
Sarcoidosis					
Multifactorial juvenile Sarcoidosis	None	Granulomatous disease, granulomatous uveitis	Neuro-sarcoidosis		62
Blau Syndrome (186580)****	See above				

AGS: Aicardi-Goutières syndrome; CAPS: Cryopyrin-associated periodic syndromes; CNS: central nervous system; DADA2: adenosine deaminase 2 deficiency; DIRA: deficiency of the interleukin 1 (IL-1) receptor antagonist; FCAS: familial cold-autoinflammatory syndrome; FMF: familial Mediterranean fever; GOF: gain of function; ICC: intracranial calcifications; HA20: haploinsufficiency in A20; JDM: juvenile dermatomyositis; JIA: juvenile idiopathic arthritis; jNPSLE: juvenile neuro-psychiatric systemic lupus erythematosus; jSLE: juvenile systemic lupus erythematosus; KD: Kawasaki disease; MIS-C: multisystem inflammatory syndrome in children; MKD: mevalonate kinase deficiency; MS: multiple sclerosis; NA: not available; NASA: systemic autoinflammation splenomegaly and anaemia; NEMO-NDAS: NEMO deleted exon 5 autoinflammatory syndrome; NMO: neuromyelitis optica; NOCARH: neonatal onset of cytopenia, autoinflammation, rash, and heamophagocytosis; PAANI: pyrin-associated autoinflammatory disease with neuroinflammation; PRAAS: Proteasome-associated autoinflammatory syndrome; ROSAH: retinal dystrophy, optic nerve oedema, splenomegaly, anhidrosis, and migraine headache; SIFD: congenital sideroblastic anaemia with immunodeficiency, fevers, and developmental delay; SMS: Singleton-Merten syndrome; VEXAS: vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic syndrome; WM: white matter.

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