ERRATUM: Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases

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In the published version of this paper, some of the columns in the last three rows of Table 3 were mistakenly transposed. The corrected table appears below. In col. 6 of the row for DNMT3A, “S3” was published in the original article. However, in the revised table for the corrigendum, it has been corrected to “S1”. In col. 6 of the row for SON, “S3” was published in the original article. However, in the revised table for the corrigendum, it has been corrected to “S2”.

Table 3 Candidate gene finding classifications

Gene	Primary reason for referral	Alteration(s)	Alteration type	Proposed inheritance	Candidate gene categories (refer to Table 1)	Candidate gene classification
ABCA5 ^a	MCA (+/-)	c.4320+1G>C	Splice	AR, homozygous	A1, C1, C2, S1	Suspected candidate
ACTG2	Single-organ system: gastrointestinal	p.R257H	Missense	AD, de novo, mosaic	B1, B3, C1, C6	Candidate
ACTG2	Single-organ system: gastrointestinal	p.R257C	Missense	AD, de novo	A2 ^g , B1, B3, C1, C6	Candidate
ACTG2	Single-organ system: gastrointestinal	p.R178H	Missense	AD, de novo	A2 ^g , B1, B3, C1, C6	Candidate
AIMP2 ^b	Single-organ system: brain	p.M241fsX25, c.575-2A>G	Frameshift, splice	AR, compound het	B1, C1, C6, S3	Suspected candidate
ARHGAP11A ^c	Oncology/cancer susceptibility	p.T790TfsX2	Frameshift	AD, cosegregates	B5, C1, C5, S3	Suspected candidate
ARV1	ND (+/-)	c.674-2A>T	Splice	AR, homozygous	A2 ^g , C1, C5, S1	Suspected candidate
BASP1	ND (+/-)	p.V225L	Missense	AD, de novo	B1, B2, C1, S1	Suspected candidate
BPTF	MCA (+/-)	p.E867Rfs*23	Frameshift	AD, de novo	B1, B3, C1, C2	Candidate
BRD4	Oncology/cancer susceptibility	p.Q517P	Missense	AD, de novo	B1, B2, C1	Candidate
CACNA1E	Neuromuscular NOS	p.F698S, p.R879W	Missense, missense	AR, compound het	B1, B2, B3, C1	Candidate
CDC42	Single-organ system: brain	p.Y23C	Missense	AD, de novo	B3, C1, C2, C4, C5	Candidate
CDC42	MCA (+/-)	p.R66G	Missense	AD, de novo	A2 ^g , B3, C1, C2, C4, C5	Candidate
CLTC	MCA (+/-)	p.D913Efs*59	Frameshift	AD, de novo	B2, C1, C4, C5	Candidate
COQ4	MCA (+/-)	p.D68H, p.Q157K	Missense, missense	AR, compound het	B1, B2, C1, C5	Candidate
DAP3	Metabolic	p.L138F, p.L152Dfs*28	Missense, frameshift	AR, compound het	C1, C2, C3, C5, S1	Suspected candidate
DGKZ ¹	MCA (+/-)	p.P560S	Missense	AR, homozygous	B1, B2, C1, C5	Candidate
DLL4	MCA (+/-)	p.P560Qfs*23	Frameshift	AD, de novo	B1, C1, C2, S3	Suspected candidate
DNMT3A	ND: Intellectual disability	p.G298W	Missense	AD, de novo	C1, C2, C5, C6, S1	Suspected candidate
DNM1	Single-organ system: brain	p.G397D	Missense	AD, de novo	B1, B2, B3, C1, C3	Candidate
DPYSL2 ^d	MCA (+/-)	p.S119R	Missense	AD, de novo	B3, B5, C1, C2	Candidate
EMILIN1	Single-organ system: connective tissue	p.A22T	Missense	AD, cosegregates	B3, C1, C2, C5, S1	Suspected candidate
ETV6 ^e	Oncology/cancer susceptibility	c.1153-5_1153-1del	Splice	AD, cosegregates	B4, C1, C4, C6	Candidate
FGR ⁵	MCA (+/-)	p.R213W	Missense	AD, cosegregates	B3, C1, C2, C5, S1	Suspected candidate
GNB1	ND (+/-)	p.I80T	Missense	AD, de novo	C1, C2, C5, C6, S1	Suspected candidate
H3F3A	ND (+/-)	p.Q126R	Missense	AD, de novo	A2 ^g , B1, C1, S1	Suspected candidate
HDAC1	ND (+/-)	p.N154A	Missense	AD, de novo	C1, C4, C5, C6	Candidate
HDAC3 ^f	ND (+/-)	p.R301Q	Missense	AD, de novo	B1, B3, C1, C2	Candidate
HNRNPK	MCA (+/-)	R287*	Nonsense	AD, de novo	B2, C1, C5, S3	Suspected candidate
HNRNPK	ND (+/-)	p.R22H	Missense	AD, de novo	A2 ^g , B2, C1, C3, C5, S3	Candidate
HNRNPR	ND (+/-)	p.Q555*	Nonsense	AD, de novo	B2, B3, C1, C5	Candidate
HTR2C	ND: Seizures	p.S407Gfs*16	Frameshift	XLR, inherited	B2, C1, C5, S3	Suspected candidate
IL21R	Single-organ system: immune	p.Y178C, p.G367S	Missense, missense	AR, compound het	C1, C2, C5, S4	Suspected candidate
ITGA11	MCA (+/-)	p.S1148L, p.T821M	Missense, Missense	AR, compound het	B3, C1, C2, C5, S1	Suspected candidate

Table 3 Continued

Gene	Primary reason for referral	Alteration(s)	Alteration type	Proposed inheritance	Candidate gene categories (refer to Table 1)	Candidate gene classification
<i>ITSN1</i> ^d	MCA (+/-)	p.V743L	Missense	AD, de novo	B3, B4, C1, C6	Candidate
<i>JAK1</i> ^e	MCA (+/-)	p.A634D	Missense	AD, cosegregates	B1, C1, C2, S1	Suspected candidate
<i>KCNA2</i>	NM: Ataxia/spasticity	p.R294H	Missense	AD, cosegregates	A2^g , B3, C1, C2, C5	Candidate
<i>LAS1L</i>	Neuromuscular NOS	p.S477N	Missense	XLD, de novo	B3, C1, C5, C6	Candidate
<i>LPHN1</i> ^f	ND (+/-)	p.R596H	Missense	AR, homozygous	B1, B3, C1, C2, S1	Suspected candidate
<i>LRFN2</i> ^b	Single-organ system: brain	p.V572X	Frameshift	AD, de novo	B3, C1, C5, S3	Suspected candidate
<i>LRFN5</i>	ND (+/-)	p.S192F, p.T290I	Missense, Missense	AR, compound het	B3, B5, C1, C4, S1	Suspected candidate
<i>MADD</i>	NM: Muscular dystrophy	p.D483G	Missense	AD, de novo	B1, C1, C2, S3	Suspected candidate
<i>MAP1B</i>	NM: Neuropathy	p.Y423H	Missense	AD, inherited	B2, C1, C5, S3	Suspected candidate
<i>MAPK1</i>	MCA (+/-)	p.135T	Missense	AD, de novo	C2, C4, C5, S4	Suspected candidate
<i>MN1</i>	MCA (+/-)	p.R1295*	Nonsense	AD, de novo	B1, C1, C2, C4, S3	Suspected candidate
<i>MSMO1</i>	ND (+/-)	p.P179L, p.Y244C	Missense, missense	AR, compound het	A2^g , C1, C5, S1	Suspected candidate
<i>MTIF2</i>	MCA (+/-)	p.V300Afs*9, p.H191Q	Frameshift, missense	AR, compound het	B1, C1, C3, S3	Suspected candidate
<i>MTOR</i>	MCA (+/-)	(p.E1799K)	Missense	AD, de novo, mosaic	B1, C1, C3, S3	Suspected candidate
<i>MYH10</i>	ND: Intellectual disability	p.E908*	Nonsense	AD, de novo	B1, B2, C1	Candidate
<i>NFASC</i>	ND (+/-)	c.1978+1G>A	Splice	AD, inherited	B1, B3, C1, C2, S1	Suspected candidate
<i>NMNAT2</i>	MCA (+/-)	p.R232Q, p.Q135Pfs*44	Missense, Frameshift	AR, compound het	B1, B2, C1	Candidate
<i>NTF4</i>	Metabolic	p.Q174R	Missense	AD, de novo	C1, C2, C5, C6	Candidate
<i>OGDHL</i>	Seizures	p.M296L, p.F734S	Missense, missense	AR, compound het	B1, C1, C6, S3	Suspected candidate
<i>OGT</i>	ND: Seizures	p.A259T	Missense	XLR, inherited	B1, B3, C1, C2, S1	Suspected candidate
<i>PCBP1</i>	MCA (+/-)	p.Q184*	Nonsense	AD, de novo	B1, B5, C1, C4	Candidate
<i>PIGQ</i>	MCA (+/-)	p.L323Pfs*119, p.Y400del	Frameshift, In-frame del	AR, compound het	B5, C1, C3, S3	Suspected candidate
<i>PLD1</i>	ND (+/-)	p.H844R	Missense	AD, cosegregates	C1, C2, C5, S4	Suspected candidate
<i>POU3F3</i>	ND: Intellectual disability	p.Q331_K335del	In-frame del	AD, de novo	C1, C2, C4, C5, S2	Suspected candidate
<i>PURA</i>	ND: Intellectual disability	p.T310GfsX2	Frameshift	AD, de novo	B1, B2, B5, C1	Candidate
<i>PURA</i>	MCA (+/-)	p.L295Afs*22	Frameshift	AD, de novo	A2^g , B1, B2, B5, C1	Candidate
<i>RAD54L</i>	Oncology/cancer susceptibility	p.G345C	Missense	AD, de novo	B1, B5, C1, C2	Candidate
<i>RAD54L</i>	Oncology/cancer Susceptibility	p.R542C	Missense	AD, cosegregates	B1, B5, C1, C2	Candidate
<i>RORB</i>	ND: ASD (autism)	p.L73P	Missense	AD, de novo	A1, C1, C6	Candidate
<i>RYR3</i>	ND: ASD (autism)	p.G1664A, p.K2857E	Missense, Missense	AR, compound het	B3, C1, C2, C5, S1	Suspected candidate
<i>SETD5</i>	Single-organ system: brain	p.T552Nfs*5	Frameshift	AD, de novo	A1, A2^g , B3, C1	Candidate
<i>SIN3A</i>	ND: ASD (autism)	p.Q827Kfs*3	Frameshift	AD, de novo	B1, C1, C4, C6, S3	Candidate
<i>SLIT2</i>	Single-organ system: ocular	p.D1407G	Missense	AD, de novo	B3, C1, C2, C6, S1	Suspected candidate
<i>SLC9A1</i>	Neuromuscular NOS	p.I451L, p.H529Y	Missense, missense	AR, compound het	A2^g , C1, C2, C5	Candidate
<i>SNAP25</i>	ND (+/-)	p.V48F	Missense	AD, de novo	B1, B2, B5, C1	Candidate
<i>SON</i>	Single-organ system: brain	p.R1850fs*3	Frameshift	AD, de novo	B1, B3, C1	Candidate
<i>SON</i>	MCA (+/-)	p.R1906Kfs*99	Frameshift	AD, de novo	B1, B3, C1, C6, S2	Suspected candidate

Table 3 Continued

Gene	Primary reason for referral	Alteration(s)	Alteration type	Proposed inheritance	Candidate gene categories (refer to Table 1)	Candidate gene classification
STAT2	Oncology/cancer susceptibility	p.R330Q	Missense	AD, cosegregates	B1, C1, C2, S3	Suspected candidate
SV2A	ND (+/-)	p.R383Q	Missense	AR, homozygous	B2, B3, B5, C1, S1	Suspected candidate
TMM50	Metabolic	p.G372S	Missense	AR, homozygous	B1, C1, C6, S3	Suspected candidate
UNC45B	ND: intellectual disability	p.A780V	Missense	AR, homozygous	B2, B5, C1, C5, S1	Suspected candidate
ZBTB18	MCA (+/-)	p.N461S	Missense	AD, de novo	A1, B3, C2, C5	Candidate
ZBTB20	MCA (+/-)	p.K604T	Missense	AD, de novo	A1, C1, C2	Candidate
ZFXH3	Single-organ system: endocrine	p.R2284*, p.S2903G	Nonsense, missense	AD, de novo	B3, C1, C4, C5	Candidate

^aProband with a candidate gene finding in both ABCA5 and DGKZ. ^bProband with a candidate gene finding in both AIMP2 and LRFV2. ^cProband with a candidate gene finding in both ARHGAP11A and ETV6. ^dProband with a candidate gene finding in both DPYSL2 and ITSN1. ^eProband with a candidate gene finding in both FGR and JAK. ^fProband with a candidate gene finding in both HDAC3 and LPHN1. ^gEvidence category A2 indicates that the gene had received a clinical validity assessment of "limited" at the time of reporting. AD, autosomal dominant; AR, autosomal recessive; ASD, autism spectrum disorder; het, heterozygous; MCA (+/-), multiple congenital anomalies (+/-); neurodevelopmental symptoms; ND (+/-), neurodevelopmental NOS (+/-); minor dysmorphic features; NM, neuromuscular; NOS, not otherwise specified; XLD, X-linked dominant; XLR, X-linked recessive; ?, unknown.