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CORRECTION

# Correction to: Expanding the genetic heterogeneity of intellectual disability

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Variant nomenclature discrepancy was identified in the article "Expanding the genetic heterogeneity of intellectual disability", Human Genetics, November 2017, Volume 136, Issue 11–12, pp 1419–1429 after its publication. Specifically, the nomenclature of *GTF3C3* was originally listed as NM_012086.4:c.1382+3A>G when it should be NM_012086.4:c.1390+3A>G, the nomenclature of *MADD* was originally listed as NM_001135943.1:c.2930T>G:p.(Val977Gly) when it should be NM_001135943.1:c.2930T>G:p.(Leu977Arg), and the variant NM_001164416.1:c.124C>T:p.(Arg42*) was listed under the gene's name *VWA3B* when it should be *H2BFM*. The following changes were made in the attached corrections:

1. Nomenclature of *GTF3C3* is changed to NM_012086.4:c.1390+3A>G in 15DG0315 (from NM_012086.4:c.1382+3A>G) in the main text, Table S1, Table S2 and Figure S1.
2. The variant NM_001164416.1:c.124C>T:p.(Arg42*) in 17DG0782 is now correctly listed under the name *H2BFM* in the abstract, Table 1, Table S1 and Table S2.
3. Nomenclature of *MADD* is changed to NM_001135943.1:c.2930T>G:p.(Leu977Arg) in 17DG0771 (From NM_001135943.1:c.2930T>G:p.(Val977Gly) in results, Table 1, Table S1, and Table S2.

The authors sincerely apologize for these errors and appreciate the opportunity to mend the records.

The original article can be found online at https://doi.org/10.1007/s00439-017-1843-2.

**Electronic supplementary material** The online version of this article (https://doi.org/10.1007/s00439-017-1859-7) contains supplementary material, which is available to authorized users.

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**Table 1** Novel candidate genes identified in this study

| Family | Clinical synopsis (HPO terms) | Novel candidate gene and variant | Zygosity | Supporting evidences |
|---|---|---|---|---|
| 17DG0763 | Failure to thrive; speech delay and language development; microcephaly; short stature; anteriorly placed anus; abnormal facial shape; intrauterine growth retardation | *ANKHD1* (NM_017747.2:c.7365dup:p.(His2456Serfs*13)) | De novo | Involved in cell survival, cell-cycle regulation, ion channel, cell survival, cell signaling, protein–protein interactions and apoptosis (PMID:16098192) and segregated within family, pLI score of 1.00 |
| 15DG0307 | Abnormal facial shape; hypospadias; chordee; global developmental delay; depressed nasal bridge; frontal bossing; abnormality of the frontal hairline; microtia; anteverted nares; café au lait spot | *ASTN2* (NM_014010.4:c.892G>C:p.(Asp298His)) | Homo | Associated with neurodegenerative disease and hippocampal volume (PMID:25410587 and 28098162) and segregated within family |
| 13DG1545 | Intellectual disability; attention deficit hyperactivity disorder; recurrent respiratory infections; downslanted palpebral fissures; prominent nose; hyperplasia of the maxilla; abnormality of the fingernails | *ATP13A1* (NM_020410.2:c.1045G>A:p.(Glu349Lys)) | Homo | Protects from iron-induced neuro cytotoxicity (PMID: 25912790) and segregated within family |
| 13DG1202 | Abnormal CNS myelination; peripheral axonal neuropathy; abnormality of the foot; scoliosis; drooling; ulnar claw; microcephaly; areflexia; intellectual disability | *FMO4* (NM_002022.1:c.83C>A:p.(Pro28His)) | Homo | Identified by positional mapping and segregated within family |
| 17DG0782 | Autism, speech delay and language development | *H2BFM* (NM_001164416.1:c.124C>T:p.(Arg42*)) | Hemi | H2BFM contains Histone 2A domain (UniProt), which is a critical regulator of chromatin modifications and cell cycle |
| 17DG0770 | Expressive language delay; autism; poor eye contact | *MADD* (NM_001135943.1:c.593G>A:p.(Arg198His) and NM_001135943.1:c.979C>T:p.(Arg327*)) | Compound Het | Critical regulator of neurotransmitter release in synapse and of neuronal viability (PMID: 11359932 and 15007167) |
| 17DG0771 | Global developmental delay; failure to thrive; poor eye contact | *MADD* (NM_001135943.1:c.2930T>G:p.(Leu977Arg)) | Homo | Critical regulator of neurotransmitter release in synapse and neuronal viability (PMID: 11359932 and 15007167) |
| 15DG2492 | Intellectual disability; short stature; abnormal facial shape; abnormal heart morphology; abnormality of the genitourinary system | *MFSD11* (NM_001242532.1:c.143G>C:p.(Gly48Ala)) | Homo | Expressed in mouse brain particularly in excitatory and inhibitory neurons (PMID: 27272503) and segregated within family |
| 12DG1370 | Tip-toe gait; intellectual disability; speech delay and language development; attention deficit hyperactivity disorder; hypertelorism; macrocephaly; tall stature | *NCKAP1* (NM_205842.2:c.3298G>T:p.(Glu1100*)) | Hetero | Expressed in the hippocampus and cerebral cortex in mouse brain and associated with Alzheimer's disease (AD) pathology (PMID: 11418237). Our expression studies of NCKAP1 on human brain clearly show abundant expression in human brain and segregated with in family members. pLI score 1.00 |

**Table 1** (continued)

| Family | Clinical synopsis (HPO terms) | Novel candidate gene and variant | Zygosity | Supporting evidences |
|---|---|---|---|---|
| 14DG0056 | Global developmental delay; congenital laryngeal stridor; hypertonia; neonatal respiratory distress; abnormal facial shape; small anterior fontanelle; recurrent respiratory infections; hypertelorism; hypotonia; wide nasal bridge; micrognathia; glossoptosis; hyperextensibility of the finger joints; 11 pairs of ribs | *NFASC* (NM_001160331.1:c.1109G>C:p.(Arg370Pro)) | Homo | Critical for Ranvier node maintenance and myelinated axon Function (PMID: 28217083) and segregated within family |
| 14DG1188 | Intellectual disability; abnormal facial shape; bicuspid aortic valve; cleft upper lip; inguinal hernia; recurrent otitis media; microcephaly; brachycephaly; anteverted ears; bulbous nose; long philtrum; thin upper lip vermilion; downturned corners of mouth; synophrys; pes planus; arachnoid cyst | *PCDHGA10* (NM_032090.1:c.823G>A:p.(Glu275Lys)) | Homo | Cadherin family genes regulate neuronal network in the brain (PubMed: 10380929) and segregated within family |
| DD_91704 | Global developmental delay, congenital microcephaly, infantile spasms, anteverted ears, thin upper lip vermilion, hyperreflexia | *PCDHGA10* (NM_032090.1:c.2477dupA:p.(Ile827Aspfs*6)) | Homo | Cadherin family genes regulates neuronal network in the brain (PubMed: 10380929) and segregated within family |
| 17DG0773 | Severe global developmental delay; neonatal respiratory distress; dilatation of lateral ventricles; hypotonia; generalized muscle weakness; recurrent respiratory infections; abnormal facial shape; wide nasal bridge; upslanted palpebral fissures; coarse facial features; generalized hirsutism; low-set, posteriorly rotated ears; thick lower lip vermilion; high, narrow palate; hepatomegaly; myopia; rotatory nystagmus; areflexia; abnormal CNS myelination; cavum septum pellucidum; enlarged cisterna magna | *PPP1R21* (NM_001135629.2:c.2089C>T:p.(Arg697*)) | Homo | Segregated within family |
| 17DG0776 | Global developmental delay; failure to thrive; central hypotonia; microcephaly; neonatal respiratory distress; recurrent aspiration pneumonia; osteopenia | *SLC12A2* (NM_001046.2:c.2617-2A>G) | Homo | Involved in hippocampal neuronal development (PMID: 23921125) and segregated within family |
| 17DG0777 | Global developmental delay; failure to thrive; recurrent respiratory infections; hydrocephalus; cutis laxa; joint hyperextensibility; hepatomegaly; hypotonia; generalized muscle weakness; strabismus; oculomotor apraxia; ventriculomegaly | *SLK* (NM_014720.1:c.1414G>T:p.(Glu472*)) | Homo | Insertional mutagenesis of *SLK* gene in mouse shows developmental defects along with neuronal and skeletal defects (Mouse Genome Informatics (MGI)) |
| 10DG0720 | Global developmental delay; seizures; microcephaly; hypertonia; hypocalcemic seizures; hypomagnesemia; strabismus; pes planus; hypocalcemia; brachycephaly | *STK32C* (NM_173575.2:c.451G>C:p.(Val-151Leu)) | Homo | Associated with depression PMID: 24929637 and segregated within family |

**Table 1** (continued)

| Family | Clinical synopsis (HPO terms) | Novel candidate gene and variant | Zygosity | Supporting evidences |
|---|---|---|---|---|
| 15DG2661 | Global developmental delay; hypotonia; abnormal facial shape; hypotonia; hip dysplasia; failure to thrive; intrauterine growth retardation; enlarged cisterna magna; polycythemia; coarse facial features; hypertrichosis; thick eyebrow; upslanted palpebral fissure; high forehead; strabismus; prominent nose; thin vermilion border; prominent nasal tip; high, narrow palate; arachnodactyly; pectus excavatum; generalized amyotrophy; overlapping toe; osteopenia; 11 pairs of ribs | *ZFAT* (NM_020863.3 :c.1199G>A:p.(Arg400Gln) ) | Homo | Peripheral T cell homeostasis and immune development (PMID: 22828507) and segregated within family |

*Homo* homozygous change, *Het* heterozygous, *Hemi* hemizygous

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