


Erratum to: Haploinsufficiency of the E3 ubiquitin-protein ligase gene *TRIP12* causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features

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In original publication, the last row of Table 1 was incorrect. The correct Table 1 is given below:

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Table 1 Summary of clinical presentations, and CMA and molecular findings in nine individuals with *TRIP12* mutations

| | Subject 1 | Subject 2 | Subject 3 | Subject 4 | Subject 5 | Subject 6 | Subject 7 | Subject 8 | Subject 9 | Summary from this study | Summary from Bramswig et al. |
|-------------------------------------|--|--|--|--|--|---|---|---|----------------------|----------------------------|------------------------------|
| Gender | Male | Male | Male | Female | Female | Male | Male | Female | Male | 6 males, 3 females | 7 males, 4 females |
| Age at examination (years) | 15 | 7 | 15 | 4 | 1.8 | 4 | 10 | 12.5 | 7 | 1.8–15 | 5.7–26.3 |
| <i>TRIP12</i> variant (NM_004238) | Deletion of 9 exons (30–38) | Deletion of 17 exons (25–41) | Deletion of 4 exons (2–5) | Deletion of <i>TRIP12</i> and adjacent 7 genes | Deletion of TRIP12 and adjacent and 3 adjacent genes | Frameshift c.3446_3447delCA (p.S1149*X) | Frameshift c.2979dupA (p.G994Rfs*5) | Missense c.2282C>T (p.A761V) | Splicing c.3743+1G>A | 5 CNVs (microdeletions) | 1 translocation |
| Genomic coordinates (min/max, hg19) | chr2:230,634,390/230,636,077–230,654,539/230,655,748 | chr2: 230,631,958–7230,632,049/230,660,565/230,661,210 | chr2:230,230,679,862/698,794–230,781,114/230,801,002 | chr2:230,489,478/230,513,445–231,457,431/231,508,839 | chr2:229,076,749/229,152,599–230,801,061/230,811,273 | Chr2: 230,661,450 | Chr2: 230,666,969 | Chr2: 230,672,494 | Chr2: 230,659,894 | | |
| Ethnicity | Mexican | NA | NA | Caucasian | NA | Saudi Arabia | United Arab Emirates (second cousins parents) | North European/Caucasian | Caucasian | | |
| Inheritance | De novo | Unknown | Unknown | De novo | De novo | De novo | De novo | De novo | De novo | 7 de novo, 2 parents NA | 9 de novo, 2 parents NA |
| Maternal age at birth (years) | 45 | NA | NA | NA | NA | 25 | 25 | 26 | 29 | 25–45 | 22–34 |
| Paternal age at birth (years) | 47 | NA | NA | NA | NA | 35 | 27 | 34 | 48 | 27–48 | 25–41 |
| DD/ID | + | + | + | + | + | + | + | + | + | 9/9 | 11/11 |
| Speech delay | + | + | ND | + | + | + | + | + | + | 8/8 | 10/11 |
| Autistic behaviors | + Poor social interaction | – | ND | + Stereotypic behaviors | + | + | + | + | – | 6/8 | 8/11 |
| Other behavioral anomalies | Hyper anxiety, Untypical behaviors | ND | ND | Repetitive, aggressive behaviors/sensory issues | Repetitive, aggressive behaviors/sensory issues | Hyperactive and destructive behavior | ND | Extremely impulsive and aggressive behaviors. | – | 6/7 | 8/11 |
| First words (months) | 36 | 60 | ND | 24 | ND | ND | ND | 24 | 42 | 1/8 fluent speech | 6/11 fluent speech |
| Verbal ability | Fluent speech | <100 words at 7 years | ND | 30 words | 5–6 words | No words | Single words | Phrase speech | Few phrase speech | 6/8 phrase or single words | 5/11 phrase or single words |
| Motor delay | + | – | ND | + | + | + | + | + | + | 1/8 no words | 7/11 |
| Walking independently (months) | 15 | ND | ND | 20 | ND | ND | 22 | 23 | 29 | 15–29 | 13–23 |

Table 1 continued

| | Subject 1 | Subject 2 | Subject 3 | Subject 4 | Subject 5 | Subject 6 | Subject 7 | Subject 8 | Subject 9 | Summary from this study | Summary from Bramswig et al. |
|------------------------------------|-------------------------------|-------------------|-----------|------------|-----------|-----------------------|------------------------|--------------------|-----------|-------------------------|------------------------------|
| Seizures | - | - | ND | - | + | - | - | - | - | 1/8 | 3/11 |
| Microcephaly/OFC (cm) (percentile) | + 52.2 (2–10%) | ND | ND | + 49 (25%) | - | ND | - | - | - | 2/6 | 0/11 1 ND |
| Obesity/weight (kg, percentile) | + 86 kg (morbid obesity) | 23.3 kg (BMI 91%) | ND | + | 65% | ND | + 97% | + 90% | - 50–75% | 4/7 | 2/11 |
| Height (cm) | 174 (75–90%) | 113.5 (6%) | ND | 103 (50%) | 89 (46%) | ND | ND | ND | 123 (75%) | Variable | Variable |
| Dysmorphic features | | | | | | | | | | | |
| Head shape | Brachycephaly, dolichocephaly | - | ND | - | - | Sloping forehead | - | - | - | 2/8 | ND |
| Narrow palpebral fissures | + | - | ND | - | + | - | + | + | ND | 4/7 | ND |
| Downturned mouth corners | + | - | ND | - | + | - | + | + | - | 4/8 | ND |
| Wide mouth | + | - | ND | - | - | - | + | + | - | 3/8 | 3/8 |
| Hypertelorism | + | - | ND | - | - | - | - | - | - | 1/8 | 1/8 |
| Epicanthic folds | - | - | ND | + | + | - | + | - | - | 3/8 | 1/8 |
| Depressed nasal bridge | - | - | ND | - | - | - (high nasal bridge) | - | - | - | 0/8 | 1/8 |
| Short nose | - (long nose) | - | ND | - | - | - | - | - | - | 0/8 | 2/8 |
| Anteverted nares | - | - | ND | + | - | - | - | - | - | 1/8 | 1/8 |
| Low hanging columella | + | - | ND | - | - | - | - | - | - | 1/8 | 3/8 |
| Long philtrum | - | - | ND | - | - | - | - | - (short philtrum) | - | 0/8 | 2/8 |
| Smooth philtrum | - | - | ND | - | - | - | - | - | - | 0/8 | 2/8 |
| Thin upper lip vermillion | - (full lips) | - | ND | - | - | - | + | - | - | 1/8 | 2/8 |
| Exaggerated Cupid's bow | - | - | ND | - | - | - | - | - | - | 0/8 | 4/8 |
| Pointed chin | - | - | ND | - | - | + | - | + | - | 2/8 | ND |
| Large ear lobe | + | - | ND | - | - | + | - | - | + | 3/8 | 3/8 |
| Low set ears | - | - | ND | - | + | - | - | - | - | 1/8 | 2/8 |
| Hand anomalies | - | - | - | - | - | - | Spindle shaped fingers | - | - | - | - |
| Hearing loss (HL) | - | - | - | + | - | - | - | - | - | 1/9 | 0/11 |
| Brain anomalies | ND | - | ND | - | - | ND | ND | - | - | - | - |

NA not available, ND not determined because of non-availability or non-applicability