**Supplementary Tables**

**Supplementary Table e-1:** Detailed clinical details of the two siblings identified with PUS7 mutations

|  |
| --- |
| **Demographic features** |
| **Family 01** | **Patient II-1** | **Patient II-2** |
| Ethnicity | Afghani | Afghani |
| parents | first degree cousins | first degree cousins |
| Gender | female | male |
| Age of last examination | 13 years | 15 years |
| **Growth** |
| Birth parameters (birth weight) | Normal (2200 kg) | Normal (2250 kg) |
| Pregnancy | uneventful | uneventful |
| Delivery | uneventful | uneventful |
| Height (cm) | 145  | 156  |
| Weight (kg) | 36 | 45 |
| HC (cm) | 50 (-2.8 SD) | 55 |
| **Neurological** |
| Intellectual disability (level) | + (moderate) | + (moderate) |
| Motor delay | - | + |
| Speech delay | + | + |
| Behavioral problems | aggressive | aggressive |
| Seizures | - | - |
| **Craniofacial** |
| Smooth philtrum | - | - |
| Full lips | + | + |
| Everted lower lip | + | + |
| Hypodontia | - | + |
| Conical shaped teeth | + | + |
| strabismus | + | - |
| **CNS** |
| CNS abnormality found by MRI | - | - |
| Other | Autism (e.g.; stereotyped or repetitive motor movements, speech)scoliosis | Autism (e.g.; stereotyped or repetitive motor movements) |

**Supplementary Table e-2:** Tracks of homozygosity identified to be present in both affected siblings.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **CHR** | **SNP1** | **SNP2** | **Position 1 (bp)** | **Position 2 (bp)** | **Size (Kb)** |
| **Ch 1** | rs1875635 | rs12139552 | 50,765,982 | 51,992,336 | 1,226.36 |
| **Ch 2** | rs2407072 | rs4850224 | 130,658,715 | 132,410,986 | 1,752.27 |
| **Ch 4** | rs4130313 | rs4859319 | 32,773,352 | 34,370,370 | 1,597.02 |
| **Ch 7** | rs2044831 | rs8185482 | 37,988,589 | 58,019,061 | 20,030.47 |
| **Ch 7** | exm2262335 | rs993932 | 61,056,053 | 71,888,561 | 10,832.508 |
| **Ch 7** | rs6951030 | rs992769 | 73,133,241 | 91,210,770 | 18,077.529 |
| **Ch7** | rs2282984 | rs17161888 | 92,279,693 | 99,357,369 | 7,077.676 |
| **Ch 7** | **exm644275** | **rs11764665** | **100,679,024** | **122,157,895** | **21,478.872** |
| **Ch 21** | rs9980427 | rs2831845 | 21,788,062 | 29,850,549 | 8,062.487 |
| **Ch 21** | rs2832373 | rs8131457 | 30,877,111 | 34,453,897 | 3,576.79 |

Chromosomal locus where the **PUS7** gene is located is represented in bold.