Table 2. Probably and possibly genetic diganosis for pedigrees with neurodevelopmental disorder.

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| --- | --- | --- | --- | --- | --- | --- | --- |
| Pedigree | Gene | Location | Variant | Mutation pattern | Inheritance pattern | ACMG guideline | Syndrome (Disease ID) |
| **Probable diagnosis** | | | | | | | |
| p1 | *KDM5C* | X:53226952 | NM\_004187.5:c.2517\_2622del | - | XR | P | Mental retardation, X-linked, syndromic, Claes-Jensen type - X-linked recessive (OMIM:300534) |
| p44 | *CHD2* | 15:93487644 | NM\_001271.4:c.1053\_1153del | - | AD | P | Myoclonic-astastic epilepsy (ORPHA:1942) |
| p2 | *CC2D2A* | 4:15534887  4:15575799 | NM\_001080522.2: c.1538G>A  NM\_001080522.2: c.3626delC | CH | AR | P/P | Meckel syndrome (ORPHA:564) |
| p9 | *TSEN2* | 3:12546725  3:12574176 | NM\_025265.4: c.904G>A  NM\_025265.4: c.1354C>T | CH | AR | LP/P | Pontocerebellar hypoplasia type 2B (OMIM:612389) |
| p30 | *WWOX* | 16:79245511  16:78466441 | NM\_016373.3: c.1063G>C  NM\_016373.3: c.854delA | CH | AR | VUS/P | Epileptic encephalopathy,  early infantile, 28 (OMIM:616211) |
| p49 | *TPP1* | 11:6636466  11:6636487 | NM\_000391.4: c.1361C>A  NM\_000391.4: c.1340G>A | CH | AR | LP/P | Ceroid lipofuscinosis, neuronal, 2 (  OMIM:204500) |
| p10 | *MECP2* | X:153296516 | NM\_004992.3: c.763C>T | *De novo* | XD | P | Rett syndrome (OMIM:312750) |
| p11 | *BRWD3* | X:79945476 | NM\_153252.5: c.3718C>T | *De novo* | XR | P | Mental retardation, X-linked 93 (OMIM:300659) |
| p14 | *ARID1B* | 6:157222580 | NM\_017519.2: c.1809delG | *De novo* | AD | P | Coffin-Siris syndrome 1 (OMIM:135900) |
| p21 | *NFIX* | 19:13186465 | NM\_002501.4: c.935G>A | - | AD | P | Sotos syndrome 2 (OMIM:614753) |
| p23 | *KAT6A* | 8:41791815 | NM\_006766.5:c.3921\_3922delGA | *De novo* | AD | LP | Mental retardation,  autosomal dominant 32 (OMIM:616268) |
| p35 | *BRAF* | 7:140477811 | NM\_004333.6: c.1497A>C | *De novo* | AD | P | Cardiofaciocutaneous syndrome (OMIM:115150) |
| p19 | *NFIX* | 19:13135823 | NM\_001365985.2:c.13\_14insAGCC | *De novo* | AD | LP | Sotos syndrome 2 (OMIM:614753) |
| **Possible diagnosis** | | | | | | | |
| p3 | *HSPG2* | 1:22162077 | NM\_005529.7: c.10409G>A | - | AD | VUS | Schwartz-Jampel syndrome (ORPHA:800) |
| p27 | *TCF4* | 18:52942972 | NM\_001083962.2: c.667A>G | - | AD | VUS | Pitt-Hopkins syndrome (ORPHA:2896) |
| p33 | *KLHL15* | X:24024785 | NM\_030624.3: c.26G>A | - | XR | VUS | Mental retardation, X-linked 103 (OMIM:300982) |
| p8 | *CLIC2* | X:154509259 | NM\_001289.6: c.392C>A | - | XR | VUS | Mental retardation, X-linked, syndromic 32 (OMIM:300886) |
| p13 | *OGT* | X:70775808 | NM\_181672.3: c.929C>T | - | XR | VUS | Mental retardation, X-linked 106 (OMIM:300997) |
| p22 | *HCFC1* | X:153220494 | NM\_005334.3:c.3356C>T | - | XR | VUS | Mental retardation, X-linked 3 ([OMIM:309541](http://www.omim.org/entry/309541)) |
| p34 | *TREX2* | X:152710282 | NM\_080701.3: c.607C>T | - | XR | VUS | Mowat-Wilson syndrome (OMIM:235730) |
| p25 | *FOXO4* | X:70320910 | NM\_005938.4: c.830A>C | - | XR | VUS | Rett syndrome, congenital variant (OMIM:613454) |
| p15 | *DSCAML1* | 11:117387250 | NM\_020693.4: c.1714dup | *De novo* | AD | LP | Smith-Magenis syndrome (ORPHA:819) |

Mutation pattern, CH, Compound heterozygous; -, SNV/Indel. Inheritance mode, XR, X-lined recessive; AD, Autosomal dominant; AR, Autosomal recessive; XD, X-linked dominant. ACMG guideline, P, Pathogenic; LP, Likely pathogenic; VUS, Variant with uncertain significance. TREX2, interactome to ZEB2; FOXO4, interactome to FOXG1; DSCAML1, interactome to IQSEC2; FOXO3, interactome to FOXG1.