**Title:** Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families

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Supplementary Table 1: List of Primers used for Segregation analysis

|  |  |  |
| --- | --- | --- |
| Primer Name | Forward Sequence | Reverse Sequence |
| *VPS53* | TGTTTCTGGCTTTTCACCTGG | CTTGACAGCACTCATGTTTCAA |
| *GLB1* | CGCTTTCATACATGTCTAGGGT | CTCATCCCCACCCTCACTG |
| *MLC1* | ACTCTGCTCACACCTCCTTC | CCCCACAGGCTTCTCACC |

Supplementary Table 2: Exome sequencing Family MR-4 two Patients revealed *VPS53* Mutation.

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Patient | cDNA | Exon | Protein | Mutation type | Genome AD\_All | 1000genome\_easa | ExAC\_All | CADD\_phred | GERP..\_RS | Mutation Taster |
| IV:2 | c.C605T | Exon 9 | p.P203L | missense | not found | not found | not found |  10 |  3.83 | Disease causing |
| IV:5 | c.C605T | Exon 9 | p.P203L | missense | not found | not found | not found |  10 | 3.83 | Disease causing |
|  |  |  |  |  |  |  |  |  |  |  |

Supplementary Table 3: Exome sequencing Family MR-7 two Patients revealed *GLB1* Mutation.

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Patient | cDNA | Exon | Protein | Mutation type | Genome AD\_All | 1000genome\_easa | ExAC\_All | CADD\_phred | GERP..\_RS | Mutation Taster |
| IV:4 | c.C1318T | Exon 13 | p.H440Y | missense | not found | not found | not found |  26 |  5.35 | Disease causing |
| IV:5 | c.C1318T | Exon 13 | p.H440Y | missense | not found | not found | not found |  26 | 5.35 | Disease causing |
|  |  |  |  |  |  |  |  |  |  |  |

Supplementary Table 4: Exome sequencing Family MR-8 one Patients revealed *MLC1* gene Mutation.

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Patient | cDNA | Exon | Protein | Mutation type | Genome AD\_SAS | 1000genome\_easa |  ExAC\_All | CADD\_phred | GERP..\_RS | Mutation Taster |
| IV:1 | c.C959A | Exon 11 | p.T230K | missense | NA | NA | NA |  29.9 | 4 | Disease causing |
|  |  |  |  |  |  |  |  |  |  |  |