|  |  |  |
| --- | --- | --- |
| **Individual** | **Additional variants** | **Comment** |
| **1** | - |  |
| **2** | BECN1; NM\_003766.3:c.1231G>A / CNTD1; NM\_173478.2:c.\*1347C>T | De novo.  pph2 probability 0.301 / benign, dbSNP rs763193803: MAF: T=0.000008/1, ExAC MAF: 8.242e-06, RVI Score Percentiles (Petrovsi et al., PLoS Genet. 2013;9(8):e1003709): BECN1: 39.5, CNTD1: 70.3 |
|  | ARMCX5-GPRASP2; NM\_001199818.1:c.-480+48868T>C / GPRASP1; NM\_001184727.1:c.608T>C NP\_001171656.1:p.Phe203Ser | X-chromosomal hemizygous.  neither gene listed as disease-associated in OMIM or in in-house curated list of X-chromosomal disease-associated genes, GPRASP1: several hemizygous carriers of loss-of-function variants in ExAC, GPRASP2: one hemizygous carrier of loss-of-function variant in ExAC |
|  | TMEM187; NM\_003492.2:c.406G>A NP\_003483.1:p.Gly136Ser | X-chromosomal hemizygous.  not listed as disease-associated gene in OMIM or in in-house curated list of X-chromosomal disease-associated genes, several hemizygous carriers of loss-of-function variants in ExAC |
|  | CASK; NM\_001126055.2:c.1720-1413C>T (NM\_003688.3:c.1790C>T NP\_003679.2:p.Thr597Ile) | X-chromosomal hemizygous.  missense variant predicted to be benign (pph2 probabiliy 0, SIFT score 0.53) |
| **3** | - |  |
| **4** | - |  |
| **5** | - |  |
| **6** | - |  |
| **7** | n.a. | there was no SNV analysis performed in WGS |
| **8** | - |  |
| **9** | JAKMIP3; chr10: 133,950,737C>T; NM\_001105521 | De novo.  intronic (Iossifov et al., 2014) |
|  | HOXC11; chr12: 54,367,011CAGGAG>C; NM\_014212 | De novo.  indel; localized in 5'UTR (Iossifov et al., 2014) |
| **10** | - |  |
| **11** | TRIM23; chr5:64,892,348C>T; NM\_033228; c. 1320G>A; p.Val440Val | De novo.  silent mutation (Iossifov et al., 2014) |
|  | IFNA4; chr9:21,187,406G>C; NM\_021068; c.125C>G; p.Ala42Gly | De novo.  missense mutation predicted to be benign (Iossifov et al., 2014) |
|  | PPP1R15A; chr19:49,376,940A>C; NM\_014330; c.450A>C; p.Thr150Thr | De novo.  silent mutation (Iossifov et al., 2014) |
|  | POU3F4; chrX:82,764,134A>G; NM\_000307; c.802A>G; p.Ser268Gly | De novo.  missense mutation predicted to be benign (Iossifov et al., 2014) |

**Supplemental Table 3**: Additional variants.