**Suppl Table 4.** Rare variants found in at least 2 patients and absent in unaffected family member (10581).

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr** | **Start** | **End** | **Ref** | **Alt** | **Func.refGene** | **Gene.refGene** | **AAChange.refGene** | **gnomAD\_genome\_ALL** | **gnomAD\_genome\_NFE** | **ExAC\_ALL** | **SNP ID** | **CADD\_phred** | **10575 (ET)** | **10577 (ET)** | **10582 (ET)** | **10581** |
| **21** | **33954601** | **33954601** | **C** | **T** | **exonic** | **TCP10L** | **TCP10L:NM\_144659:exon3:c.G269A:p.R90Q** | **3.23E-05** | **6.67E-05** | **1.65E-05** | **rs151233771** | **13.03** | **0/1** | **0/1** | **0/1** | **0/0** |
| 5 | 171765672 | 171765672 | C | T | exonic | SH3PXD2B | SH3PXD2B:NM\_001017995:exon13:c.G2437A:p.G813R | . | . | 8.28E-06 | rs376893926 | 0.248 | 0/1 | 0/0 | 0/1 | 0/0 |
| 7 | 100550253 | 100550253 | G | T | exonic | MUC3A | MUC3A:NM\_005960:exon2:c.G834T:p.Q278H | 9.15E-05 | 0.0001 | . | . | 0.791 | 0/1 | 0/0 | 0/1 | 0/0 |
| 7 | 100550288 | 100550288 | G | T | exonic | MUC3A | MUC3A:NM\_005960:exon2:c.G869T:p.R290M | 5.77E-05 | 0 | . | . | 7.143 | 0/1 | 0/0 | 0/1 | 0/0 |
| 10 | 101565164 | 101565164 | A | C | exonic | ABCC2 | ABCC2:NM\_000392:exon11:c.A1490C:p.K497T | . | . | 8.24E-06 | rs779734655 | 23 | 0/1 | 0/0 | 0/1 | 0/0 |
| 14 | 23564459 | 23564459 | T | G | exonic | ACIN1 | ACIN1:NM\_001164814:exon1:c.A37C:p.T13P,ACIN1:NM\_001164815:exon1:c.A37C:p.T13P,ACIN1:NM\_014977:exon1:c.A37C:p.T13P | . | . | . | . | 20.8 | 0/1 | 0/0 | 0/1 | 0/0 |
| 19 | 12936644 | 12936644 | A | C | exonic | RTBDN | RTBDN:NM\_001080997:exon6:c.T566G:p.V189G,RTBDN:NM\_001270441:exon6:c.T596G:p.V199G,RTBDN:NM\_001270442:exon6:c.T584G:p.V195G,RTBDN:NM\_001270443:exon6:c.T566G:p.V189G,RTBDN:NM\_001270444:exon6:c.T566G:p.V189G,RTBDN:NM\_001270445:exon6:c.T548G:p.V183G,RTBDN:NM\_031429:exon7:c.T662G:p.V221G | . | . | . | . | 16.96 | 0/1 | 0/0 | 0/1 | 0/0 |
| 8 | 110402711 | 110402711 | A | G | exonic | PKHD1L1 | PKHD1L1:NM\_177531:exon9:c.A728G:p.N243S | . | . | 0.0002 | rs200280185 | 9.576 | 0/0 | 0/1 | 0/1 | 0/0 |
| 11 | 1017514 | 1017514 | A | G | exonic | MUC6 | MUC6:NM\_005961:exon31:c.T5287C:p.S1763P | . | . | 0.452 | rs79037833 | 1.418 | 0/0 | 0/1 | 0/1 | 0/0 |
| 16 | 1034730 | 1034730 | A | C | exonic | SOX8 | SOX8:NM\_014587:exon3:c.A685C:p.T229P | . | . | . | . | 24.5 | 0/0 | 0/1 | 0/1 | 0/0 |
| 22 | 37261022 | 37261022 | G | A | exonic | NCF4 | NCF4:NM\_000631:exon3:c.G179A:p.R60H,NCF4:NM\_013416:exon3:c.G179A:p.R60H | 3.24E-05 | 6.69E-05 | 3.30E-05 | rs369847561 | 34 | 0/0 | 0/1 | 0/1 | 0/0 |