**Suppl Table 3.** CNVs found in at least two ET patients and absent in unaffected family member (10581).

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr** | 1 | 17 | 3 | 4 | 4 | 7 | 7 |
| **SV start** | 144590001 | 36284001 | 196626001 | 3573001 | 49523501 | 56557501 | 63022501 |
| **SV end** | 144612500 | 36321000 | 196661000 | 3613500 | 49549500 | 56564500 | 63041500 |
| **SV length** | 22500 | 37000 | 35000 | 40500 | 26000 | 7000 | 19000 |
| **SV type** | DUP | DUP | DUP | DUP | DUP | DUP | DUP |
| **natorP1** | 4.76E-08 | 0 | 4.55E-12 | 0 | 6.74E-09 | 2.26E-06 | 0 |
| **natorP2** | 4.06E+07 | 2.68E-67 | 0.655941 | 3.68E-112 | 5.23E+06 | 9.42E-58 | 6957.12 |
| **natorP3** | 1.08E-06 | 0 | 4.83E-12 | 0 | 7.09E-08 | 0.00076081 | 1.87E-11 |
| **natorP4** | 5.92E+07 | 3.45E-63 | 2.33232 | 3.44E-106 | 8.49E+06 | 1.44E-68 | 27136 |
| **natorQ0** | 0.168048 | 0.361554 | 0.190946 | 0.00525089 | 0.311403 | 0.00098503 | 0.046435 |
| **natorRD** | 1.61571 | 2.26486 | 0.830952 | 2.00834 | 2.8091 | 1.91497 | 1.62943 |
| **FORMAT** | GT:CN | GT:CN | GT:CN | GT:CN | GT:CN | GT:CN | GT:CN |
| **AnnotSV type** | full | full | full | full | full | full | full |
| **Gene name** | NBPF19/NBPF20/NBPF8/PFN1P2 | TBC1D3/TBC1D3C/TBC1D3D/TBC1D3E/TBC1D3F/TBC1D3G/TBC1D3H/TBC1D3I/TBC1D3K/TBC1D3L | SENP5 | LINC00955 | . | DKFZp434L192 | . |
| **DGV\_GAIN\_IDs** | gssvG1745 | gssvG16374 | gssvG26773 | gssvG27035,gssvG27052,gssvG27040 | gssvG27515 | gssvG34498 | gssvG34620 |
| **DGV\_GAIN\_n\_samples\_with\_SV** | 4 | 3 | 3 | 12 | 7 | 2 | 3 |
| **DGV\_GAIN\_n\_samples\_tested** | 12129 | 12129 | 12365 | 15754 | 20703 | 3377 | 12365 |
| **DGV\_GAIN\_Frequency** | 0.00032979 | 0.00024734 | 0.00024262 | 0.00076171 | 0.00033812 | 0.00059224 | 0.00024262 |
| **DGV\_LOSS\_IDs** | . | . | . | . | gssvL90104,gssvL90106,gssvL90114 | gssvL115683 | gssvL115955 |
| **DGV\_LOSS\_n\_samples\_with\_SV** | 0 | 0 | 0 | 0 | 48 | 9 | 2 |
| **DGV\_LOSS\_n\_samples\_tested** | 0 | 0 | 0 | 0 | 12470 | 11287 | 12129 |
| **DGV\_LOSS\_Frequency** | -1 | -1 | -1 | -1 | 0.00384924 | 0.00079738 | 0.00016489 |
| **GD\_ID** | . | . | gnomAD-SV\_v2.1\_DUP\_3\_10707 | . | . | . | . |
| **GD\_AN** | . | . | 21694 | . | . | . | . |
| **GD\_N\_HET** | . | . | 1 | . | . | . | . |
| **GD\_N\_HOMALT** | . | . | 0 | . | . | . | . |
| **GD\_AF** | -1 | -1 | 4.60E-05 | -1 | -1 | -1 | -1 |
| **GD\_POPMAX\_AF** | -1 | -1 | 0.000105 | -1 | -1 | -1 | -1 |
| **GD\_ID\_others** | . | gnomAD-SV\_v2.1\_INV\_17\_731 | . | . | . | . | gnomAD-SV\_v2.1\_DEL\_7\_81663 |
| **DDD\_SV** | 1:142551423-144717850;1:144183014-144804913;1:144523134-144828537 | 17:34492843-36395738;17:36253047-36441674;17:36294507-36338070 | . | 4:3572961-3631831 | 4:49121961-49626026 | . | . |
| **DDD\_DUP\_n\_samples\_with\_SV** | 77 | 80 | . | 0 | 130 | . | . |
| **DDD\_DUP\_Frequency** | 0.09112426 | 0.09467456 | -1 | 0 | 0.15384615 | -1 | -1 |
| **DDD\_DEL\_n\_samples\_with\_SV** | 60 | 113 | . | 1 | 158 | . | . |
| **DDD\_DEL\_Frequency** | 0.07100592 | 0.13372781 | -1 | 0.00118343 | 0.18698225 | -1 | -1 |
| **IMH\_AF** | -1 | -1 | -1 | -1 | -1 | -1 | -1 |
| **IMH\_ID\_others** | 1:83946-209634271\_INV;1:4599854-170869838\_INV;1:8923698-236647622\_INV;1:9585556-193606524\_INV;1:17124868-234920285\_INV;1:17216755-149229918\_INV;1:17225076-149220997\_INV;1:17229844-149217063\_INV;1:47908261-216684957\_INV;1:69129972-188838391\_INV;1:76428345-179322403\_INV;1:108097959-232960701\_INV;1:109720024-154259996\_INV;1:121137793-148856171\_INV | 17:30436598-65922464\_INV;17:32367053-60556542\_INV;17:32367222-60556516\_INV;17:32367434-60556508\_INV | 3:112314645-197202040\_INV;3:195349061-197394417\_INV;3:195371561-197367616\_INV;3:195376258-197363816\_INV | 4:2823209-17706072\_INV | 4:43358175-53119726\_DEL;4:4773638-77530254\_INV;4:21758290-84442445\_INV;4:21758875-84442542\_INV;4:24549534-125995071\_INV;4:32219468-79170284\_INV;4:45105686-118544232\_INV;4:49230198-49569276\_INV | 7:23891012-65526751\_INV;7:39546632-99949025\_INV;7:55955254-100292055\_INV;7:56087154-62517970\_INV;7:56087327-62517980\_INV;7:56087505-62518011\_INV | 7:23891012-65526751\_INV;7:39546632-99949025\_INV;7:55955254-100292055\_INV;7:56638503-63892526\_INV;7:56768643-63321375\_INV;7:56801223-63306820\_INV;7:57223326-63739339\_INV |
| **promoters** | NBPF8 | . | . | LINC00955 | . | DKFZp434L192 | . |
| **GHid\_not\_elite** | GH01J120414 | . | GH03J196908;GH03J196931 | GH04J003575 | . | . | . |
| **GHtype** | Enhancer | . | Enhancer | Enhancer | . | . | . |
| **GHgene\_elite** | NBPF8 | . | PAK2;PIR47781;PIR59931;SENP5 | LINC00955 | . | . | . |
| **GHgene\_not\_elite** | LOC101929788 | . | CEP19;ENSG00000230732;GC03M196873;PIGX;RNF168;UBXN7 | GC04P003604;GC04P003605;GC04P003606;MFSD10;ZBTB49 | . | . | . |
| **GHtissue** | A549;Adrenal gland (fetal);B cells (PB);B cells (VB);CD14+ monocytes (PB);CD4+ ab T cells (VB);CD8+ ab T cells (CB);Central memory CD4+ ab T cells (VB);DND-41;EPC (VB);Endothelial cells of umbilical vein;Endothelial cells of umbilical vein (CB);GM12878;H1 mesenchymal;H1 trophoblast;HSMM (myotube);HeLa-S3;HepG2;IMR90;K562... | . | B cell;GM12878;T cell | large intestine;small intestine | . | . | . |
| **GCcontent\_left** | 0.355 | 0.57 | 0.4 | 0.475 | 0.38 | 0.41 | 0.52 |
| **GCcontent\_right** | 0.555 | 0.345 | 0.35 | 0.6 | 0.43 | 0.635 | 0.635 |
| **Repeats\_coord\_left** | . | . | 3:196625775-196625967 | 4:3572616-3573108 | 4:49522801-49523527/4:49523527-49523705 | 7:56557552-56557570/7:56557570-56557599/7:56557599-56557885 | 7:63022312-63022586 |
| **Repeats\_type\_left** | . | . | AluY | L1MC4 | L1PB/AluSp | AluSx/(TTG)n/AluSx | AluJr |
| **Repeats\_coord\_right** | . | 17:36320772-36320924/17:36321057-36321257 | . | . | 4:49548968-49549628 | . | 7:63041588-63041619 |
| **Repeats\_type\_right** | . | L1MC5/AluJb | . | . | SATR1 | . | (TCTCC)n |
| **HI\_DDDpercent** | 96.77 | 93.79 | 10.84 | 93.6 | . | . | . |
| **delZ\_ExAC** | . | . | -1.7175691 | . | . | . | . |
| **dupZ\_ExAC** | . | . | -1.0345077 | . | . | . | . |
| **cnvZ\_ExAC** | . | . | -1.2815249 | . | . | . | . |
| **synZ\_ExAC** | 1.84159365 | 2.39402605 | 0.00561158 | . | . | . | . |
| **misZ\_ExAC** | 2.31463274 | 2.82547992 | 1.18600666 | . | . | . | . |
| **pLI\_ExAC** | 0.00028244 | 0.62292873 | 0.99276066 | . | . | . | . |
| **AnnotSV ID** | 1\_144590001\_144612500\_DUP\_1 | 17\_36284001\_36321000\_DUP\_1 | 3\_196626001\_196661000\_DUP\_1 | 4\_3573001\_3613500\_DUP\_1 | 4\_49523501\_49549500\_DUP\_1 | 7\_56557501\_56564500\_DUP\_1 | 7\_63022501\_63041500\_DUP\_1 |
| **AnnotSV ranking** | 2 | 2 | 4 | 2 | 2 | 2 | 2 |
| **10575 (ET)** | . | ./1:2 | 0/1:2 | 1/1:2 | ./1:3 | 1/1:2 | 0/1:2 |
| **10577 (ET)** | 1/1:2 | . | . | . | . | . | . |
| **10582 (ET)** | 0/1:2 | ./1:3 | 0/1:2 | 1/1:2 | ./1:3 | 1/1:2 | 0/1:2 |
| **10581** | . | . | . | . | . | . | . |