Table S1 List of 153 genes associated with epilepsy in the MyGenostics panel

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| ADSL  | CHD2  | DHFR  | GLB1  | MAGI2  | PNPO  | SLC9A6  |
| ALDH7A1  | CHRNA2  | DIAPH3  | GLRA1  | MAPK10  | POLG  | SPTAN1  |
| ALG13  | CHRNA4  | DNAJC6  | GPR56  | MBD5  | PPT1  | SRPX2  |
| ARG1  | CHRNA7  | DNM1  | GPR98  | MDGA2  | PROC  | ST3GAL2  |
| ARHGEF15  | CHRNB2  | DOCK7  | GRIN1  | ME2  | PRRT2  | ST3GAL5  |
| ARHGEF9  | CLCN2  | EEF1A2  | GRIN2A  | MECP2  | RBFOX1  | STRADA  |
| ARX  | CLCN4  | EFHC1  | GRIN2B  | MEF2C  | RBFOX2  | STXBP1  |
| ASAH1  | CLN3  | ELP4  | HAX1  | MFSD8  | RBFOX3  | SYNGAP1  |
| ATP13A4  | CLN5  | EPHB2  | HDAC4  | MTHFR  | RELN  | SYNJ1  |
| ATP1A2  | CLN6  | ErbB4  | HEXA  | MTOR  | RYR3  | SZT2  |
| ATP1A3  | CLN8  | FASN  | HEXB  | NDE1  | SCN1A  | TBC1D24  |
| ATP6AP2  | CNTN5  | FLNA  | HNRNPH1  | NEDD4L  | SCN1B  | TCF4  |
| ATP7A  | CNTNAP2  | FOLR1  | HNRNPU  | NID2  | SCN2A  | TNK2  |
| BRAF  | COX6B1  | FOXG1  | IQSEC2  | NRXN1  | SCN8A  | TPP1  |
| BSN  | CSTB  | FOXP2  | KCNB1  | PAFAH1B1  | SHANK3  | TSC1  |
| CACNA1A  | CTNNA3  | GABBR2  | KCNH5  | PCDH19  | SLC13A5  | TSC2  |
| CACNA1H  | CTSD  | GABRA1  | KCNMA1  | PDHA1  | SLC19A3  | TUBA1A  |
| CACNB4  | CYB5R3  | GABRA6  | KCNQ2  | PIGA  | SLC1A3  | UBE3A  |
| CASK  | DBH  | GABRB2  | KCNQ3  | PIGV  | SLC25A22  | VRK2  |
| CDH13  | DCX  | GABRB3  | KCNT1  | PLCB1  | SLC2A1  | WDR45  |
| CDH9  | DEPDC5  | GABRD  | LGI1  | PNKD  | SLC35A2  | ZEB2  |
| CDKL5  | DGKD  | GABRG2  | LIAS | PNKP  | SLC46A1 |  |

Table S2 The ddPCR results in 20 *PCDH19*-related epileptic families

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Subject No. | Sample | Drop count | Mutant allelic fraction | *PCDH19* Mutation |
| Total No. | Mutant No. | Wild No. | Mean | 95%L | 95%U |
| 001 | blood | 9445727 | 11516 | 11470 | 50.10% | 49.45% | 50.75% | c.488T>G |
| 001F | blood | 8468678 | 3328 | 21036 | 13.66% | 13.23% | 14.10% | c.488T>G |
| 001F | hair | 9710907 | 141 | 3251 | 4.16% | 3.51% | 4.88% | c.488T>G |
| 001F | buccal | 9838316 | 4736 | 21175 | 18.28% | 17.81% | 18.75% | c.488T>G |
| 001F | nail | 10029597 | 10320 | 17287 | 37.38% | 36.81% | 37.96% | c.488T>G |
| 001F | saliva | 10808241 | 4202 | 30479 | 12.12% | 11.77% | 12.46% | c.488T>G |
| 001F | urine | 10106162 | 31 | 68 | 31.31% | 22.36% | 41.41% | c.488T>G |
| 001F | semen | 10347332 | 4441 | 23743 | 15.76% | 15.33% | 16.19% | c.488T>G |
| 001M | blood | 9724717 | 0 | 20173 | 0.00% | 0.00% | 0.02% | c.488T>G |
| 002 | blood | 9158314 | 23284 | 25850 | 47.39% | 46.95% | 47.83% | c.1347\_1348insAAC |
| 002F | blood | 9138940 | 2 | 18254 | 0.01% | 0.00% | 0.04% | c.1347\_1348insAAC |
| 002M | blood | 8523214 | 9 | 63371 | 0.01% | 0.01% | 0.03% | c.1347\_1348insAAC |
| 003 | blood | 9858580 | 41321 | 40570 | 50.46% | 50.12% | 50.80% | c.1019A>G |
| 003F | blood | 8885706 | 0 | 17052 | 0.00% | 0.00% | 0.02% | c.1019A>G |
| 003M | blood | 8842757 | 0 | 34161 | 0.00% | 0.00% | 0.01% | c.1019A>G |
| 004 | blood | 8641995 | 14543 | 14228 | 50.55% | 49.97% | 51.13% | c.1019A>G |
| 004M | blood | 8198298 | 11157 | 10494 | 51.53% | 50.86% | 52.20% | c.1019A>G |
| 004MGF | blood | 7747185 | 0 | 13836 | 0.00% | 0.00% | 0.03% | c.1019A>G |
| 004MGM | blood | 8792515 | 0 | 35666 | 0.00% | 0.00% | 0.01% | c.1019A>G |
| 006 | blood | 8412186 | 25686 | 25305 | 50.37% | 49.94% | 50.81% | c.1017delC |
| 006F | blood | 8863596 | 0 | 13617 | 0.00% | 0.00% | 0.03% | c.1017delC |
| 006M | blood | 8578199 | 16667 | 17153 | 49.28% | 48.75% | 49.82% | c.1017delC |
| 007 | blood | 8940039 | 12775 | 12691 | 50.16% | 49.55% | 50.78% | C.790G>C |

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| 007F | blood | 9442598 | 14888 | 47 | 99.69% | 99.58% | 99.77% | C.790G>C |
| 007M | blood | 8805498 | 1 | 29913 | 0.00% | 0.00% | 0.02% | C.790G>C |
| 008 | blood | 9261374 | 8546 | 8496 | 50.15% | 49.39% | 50.90% | c.1184G>C |
| 008F | blood | 9578248 | 1 | 18543 | 0.01% | 0.00% | 0.03% | c.1184G>C |
| 008M | blood | 8661868 | 0 | 25828 | 0.00% | 0.00% | 0.01% | c.1184G>C |
| 011 | blood | 9244373 | 5944 | 5847 | 50.41% | 49.50% | 51.32% | c.1375C>T |
| 011F | blood | 9286472 | 0 | 16650 | 0.00% | 0.00% | 0.02% | c.1375C>T |
| 011M | blood | 8979903 | 0 | 33654 | 0.00% | 0.00% | 0.01% | c.1375C>T |
| 013 | blood | 9499990 | 330 | 314 | 51.24% | 47.31% | 55.17% | c.370G>A |
| 013S | blood | 9609107 | 832 | 943 | 46.87% | 44.53% | 49.23% | c.370G>A |
| 013F | blood | 9961261 | 340 | 1649 | 17.09% | 15.46% | 18.82% | c.370G>A |
| 013F | buccal | 8705715 | 2056 | 22440 | 8.39% | 8.05% | 8.75% | c.370G>A |
| 013F | nail | 10558801 | 160 | 12407 | 1.27% | 1.08% | 1.48% | c.370G>A |
| 013F | saliva | 10420060 | 1555 | 9231 | 14.42% | 13.76% | 15.09% | c.370G>A |
| 013F | urine | 10562064 | 9 | 260 | 3.35% | 1.54% | 6.26% | c.370G>A |
| 013F | semen | 9111565 | 492 | 2080 | 19.13% | 17.63% | 20.70% | c.370G>A |
| 013M | blood | 10548038 | 1 | 2267 | 0.04% | 0.00% | 0.25% | c.370G>A |
| 014 | blood | 6801232 | 10830 | 10586 | 50.57% | 49.90% | 54.54% | c.1240G>A |
| 014F | blood | 8364969 | 1 | 12649 | 0.01% | 0.00% | 49.38% | c.1240G>A |
| 014M | blood | 8629970 | 10850 | 11104 | 49.42% | 48.76% | 48.44% | c.1240G>A |
| 014M | hair | 9748038 | 208 | 211 | 49.64% | 44.75% | 53.17% | c.1240G>A |
| 014M | saliva | 8362733 | 12550 | 13688 | 47.83% | 47.23% | 48.44% | c.1240G>A |
| 014M | urine | 9674086 | 135 | 151 | 47.20% | 41.30% | 53.17% | c.1240G>A |
| 014MGM | blood | 7816940 | 6 | 32882 | 0.02% | 0.01% | 0.04% | c.1240G>A |
| 014FGF | blood | 9566760 | 6 | 32882 | 0.01% | 0.00% | 0.03% | c.1240G>A |
| 014FGM | blood | 8802810 | 3 | 31787 | 0.01% | 0.00% | 0.02% | c.1240G>A |
| 016 | blood | 10025222 | 12628 | 13140 | 49.01% | 48.39% | 49.62% | c.1019A>G |
| 016F | blood | 9402591 | 0 | 4637 | 0.00% | 0.00% | 0.08% | c.1019A>G |
| 016M | blood | 7887228 | 0 | 20172 | 0.00% | 0.00% | 0.02% | c.1019A>G |
| 017 | blood | 9704797 | 14538 | 14478 | 50.10% | 49.53% | 50.68% | c.695A>G |
| 017F | blood | 9846025 | 0 | 6111 | 0.00% | 0.00% | 0.06% | c.695A>G |
| 017M | blood | 10715290 | 0 | 17850 | 0.00% | 0.00% | 0.02% | c.695A>G |
| 020 | blood | 9854513 | 1955 | 1974 | 49.76% | 48.18% | 51.33% | c.471C>A |
| 020F | blood | 10589917 | 0 | 1297 | 0.00% | 0.00% | 0.28% | c.471C>A |
| 020M | blood | 9810489 | 0 | 1292 | 0.00% | 0.00% | 0.29% | c.471C>A |
| 021 | blood | 9772903 | 27982 | 28081 | 49.91% | 49.50% | 50.33% | c.964G>C |
| 021F | blood | 10926262 | 0 | 16607 | 0.00% | 0.00% | 0.02% | c.964G>C |
| 021M | blood | 9881127 | 0 | 55599 | 0.00% | 0.00% | 0.01% | c.964G>C |
| 025 | blood | 9833568 | 11990 | 2780 | 81.18% | 80.54% | 81.81% | c.317T>A |
| 025 | hair | 9903094 | 3893 | 61 | 98.46% | 98.02% | 98.82% | c.317T>A |
| 025 | buccal | 10103157 | 16261 | 4182 | 79.54% | 78.98% | 80.09% | c.317T>A |
| 025 | nail | 9648471 | 354 | 171 | 67.43% | 63.23% | 71.42% | c.317T>A |
| 025 | saliva | 10328252 | 685 | 159 | 81.16% | 78.36% | 83.75% | c.317T>A |
| 025 | urine | 10205476 | 59 | 16 | 78.67% | 67.68% | 87.29% | c.317T>A |
| 025F | blood | 10559849 | 0 | 5369 | 0.00% | 0.00% | 0.07% | c.317T>A |
| 025F | hair | 10875091 | 0 | 3021 | 0.00% | 0.00% | 0.12% | c.317T>A |
| 025F | buccal | 9284060 | 1 | 25299 | 0.00% | 0.00% | 0.02% | c.317T>A |
| 025F | nail | 9911513 | 11 | 20824 | 0.05% | 0.03% | 0.09% | c.317T>A |
| 025F | saliva | 9278855 | 2 | 13583 | 0.01% | 0.00% | 0.05% | c.317T>A |
| 025F | urine | 10377045 | 0 | 429 | 0.00% | 0.00% | 0.86% | c.317T>A |
| 025F | semen | 8810601 | 0 | 30987 | 0.00% | 0.00% | 0.01% | c.317T>A |
| 025M | blood | 9444803 | 0 | 18906 | 0.00% | 0.00% | 0.02% | c.317T>A |
| 025M | hair | 8973370 | 0 | 1466 | 0.00% | 0.00% | 0.25% | c.317T>A |
| 025M | buccal | 9814500 | 0 | 41247 | 0.00% | 0.00% | 0.01% | c.317T>A |
| 025M | nail | 9970224 | 7 | 872 | 0.80% | 0.32% | 1.63% | c.317T>A |
| 025M | saliva | 10213711 | 1 | 12680 | 0.01% | 0.00% | 0.04% | c.317T>A |
| 025M | urine | 10080266 | 0 | 31412 | 0.00% | 0.00% | 0.01% | c.317T>A |
| 026 | blood | 9361738 | 8218 | 13945 | 37.08% | 36.44% | 37.72% | c158\_159insT |
| 026F | blood | 10060282 | 0 | 14739 | 0.00% | 0.00% | 0.03% | c158\_159insT |
| 026M | blood | 9400294 | 0 | 15980 | 0.00% | 0.00% | 0.02% | c158\_159insT |
| 027 | blood | 9296126 | 9327 | 9540 | 49.44% | 48.72% | 50.15% | c.262G>T |
| 027F | blood | 10523650 | 0 | 948 | 0.00% | 0.00% | 0.39% | c.262G>T |
| 027M | blood | 8907582 | 1 | 13684 | 0.01% | 0.00% | 0.04% | c.262G>T |
| 029 | blood | 10209863 | 1238 | 3396 | 26.72% | 25.45% | 28.01% | c.2341delA |
| 029F | blood | 10304373 | 0 | 5737 | 0.00% | 0.00% | 0.06% | c.2341delA |
| 029M | blood | 8634163 | 0 | 5401 | 0.00% | 0.00% | 0.07% | c.2341delA |
| 030 | blood | 10320964 | 4670 | 4694 | 49.87% | 48.85% | 50.89% | c.964G>C |
| 030F | blood | 9924059 | 0 | 3235 | 0.00% | 0.00% | 0.11% | c.964G>C |
| 030M | blood | 10050223 | 0 | 1772 | 0.00% | 0.00% | 0.21% | c.964G>C |
| 031 | blood | 10360456 | 2003 | 2006 | 49.96% | 48.40% | 51.52% | c.2113C>T |
| 031F | blood | 10058477 | 8549 | 4 | 99.95% | 99.88% | 99.99% | c.2113C>T |
| 031M | blood | 10659987 | 1 | 12452 | 0.01% | 0.00% | 0.04% | c.2113C>T |

Table S3. Males and females with a mosaic *PCDH19* variant

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Mosaic (Sex) | Analysis | *PCDH19* variantmutation | Phenotype | Mutant allelic frequency |
| A (male) | NGS, ddPCR | c.317T>A, p.M106R | *PCDH19*-related epilepsy | 81.18% (blood), 98.46% (hair), 79.54% (buccal), 67.43% (nail), 81.16% (saliva), 78.67% (urine) |
| B (male) | NGS, ddPCR | c.158dupT, p.D54GfsX35 | *PCDH19*-related epilepsy | 37.08% (blood) |
| C (male) | Sanger sequencing, ddPCR | c.370G>A, p.D124N | Asymptomatic | 17.09% (blood), 8.39% (buccal), 1.27% (nail), 14.42% (saliva), 3.35% (urine), 19.13% (sperm) |
| D (male) | Sanger sequencing, ddPCR | c.488T>G, p.V163G | Asymptomatic | 13.23% (blood), 3.51% (hair), 17.81% (buccal), 36.81% (nail), 11.77% (saliva), 22.36% (urine), 15.33% (sperm) |
| E (female) | ddPCR | c.2341delA, p.I781SfsX19 | *PCDH19*-related epilepsy | 26.72% (blood) |
| F (male) | NGS | c.1864G > C, p.Gly622Arg | *PCDH19*-related epilepsy | 60% (blood) |
| G (male) | NGS | c.840C > G, p.Tyr280X | *PCDH19*-related epilepsy | 22% (blood) |
| H (male) | NGS | c.462C > G, p.Tyr154X | *PCDH19*-related epilepsy | 65% (blood) |
| I (male) | NGS | c.1682C > G, p.Pro561Arg | *PCDH19*-related epilepsy | 78% (blood) |
| J (male) | NGS | c.799G > T, p.Glu267X | *PCDH19*-related epilepsy | 20% (blood) |
| K (male) | Microarray | del PCDH19 | *PCDH19*-related epilepsy | 100% (lymphocytes), 47% (fibroblasts) |
| L (male) | Exome sequencing | c.605C > A, p.Ser202X | *PCDH19*-related epilepsy | 50% (blood), 70% (buccal cells), 100% (urine sediment) |
| M (male) | NGS | c.918C > G, p.Tyr306X | *PCDH19*-related epilepsy | 10% (lymphocytes, saliva, hair) |
| N (male) | NGS | c.1352 C > T, p.Pro451Leu | *PCDH19*-related epilepsy | 90%(lymphocytes, urine) |
| O (male) | unknown | c.2147+2T>C | *PCDH19*-related epilepsy | unknown |
| P (female) | Restriction enzyme analysis | c.74T>C, p.L25P | Asymptomatic | unknown |
| Q (female) | Sanger sequencing | c.1019A>G, p.N340S | *PCDH19*-related epilepsy | unknown |
| L (female) | qPCR | c.1019A>G, p.N340S | Asymptomatic | 35-60% (blood, saliva), 0 (urine)？ |



**Disclosure of the ascertainment of 42 patients with *PCDH19* variants in a Chinese population**



**Workflow for the detection of Mosaic Mutations in 42 families**