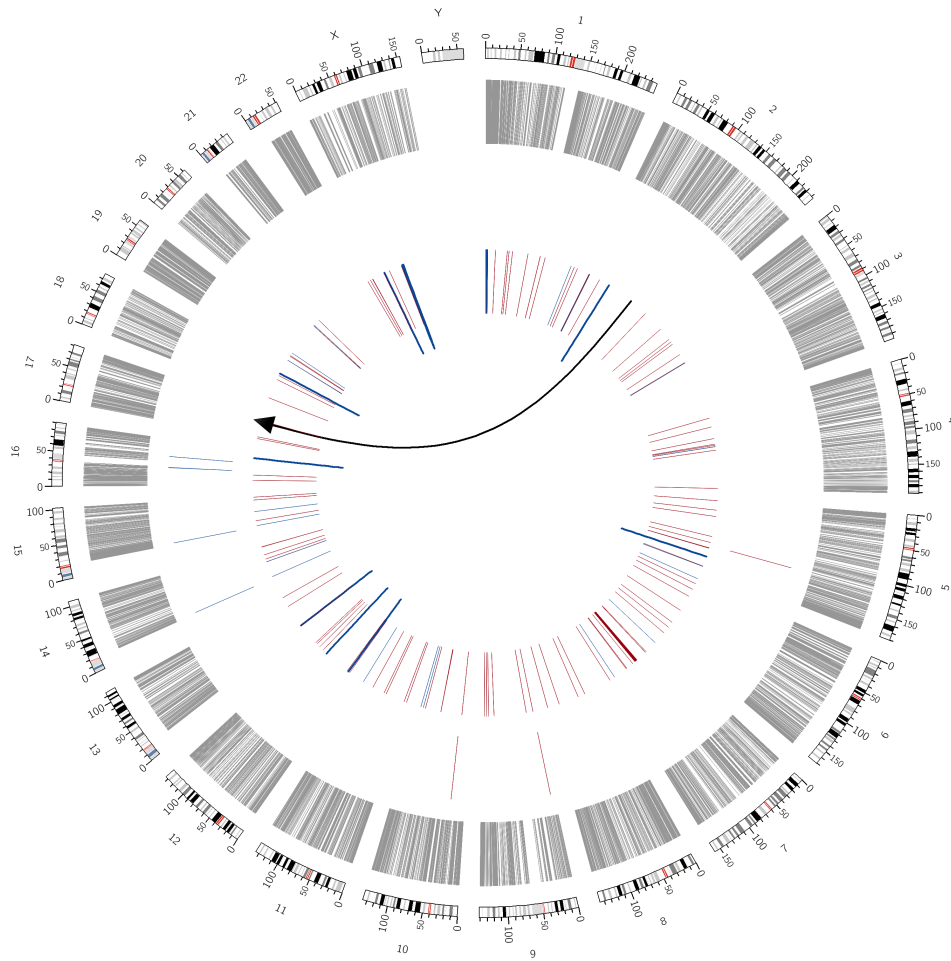


## **Supplemental Tables and Figures**

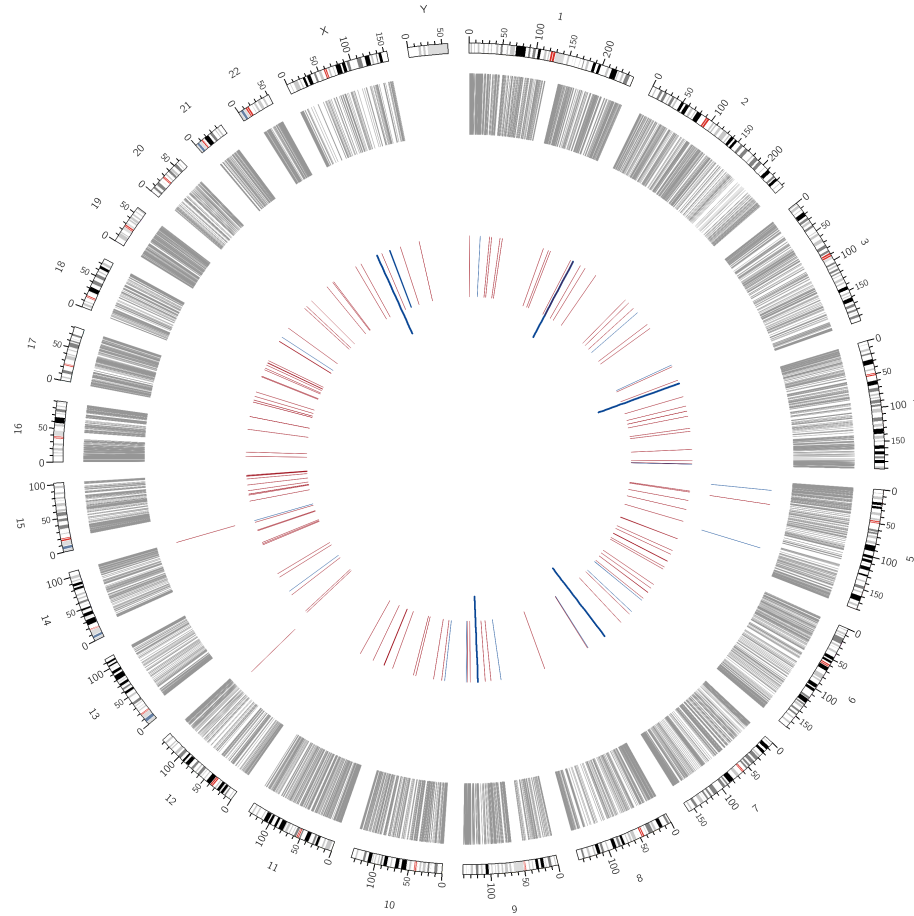
**Post-zygotic de novo changes in glutamate and dopamine pathways may explain  
discordance of monozygotic twins for schizophrenia**

Castellani, CA., Melka, MG., Gui, JL., Gallo, AJ. , O'Reilly, RL., Singh, SM

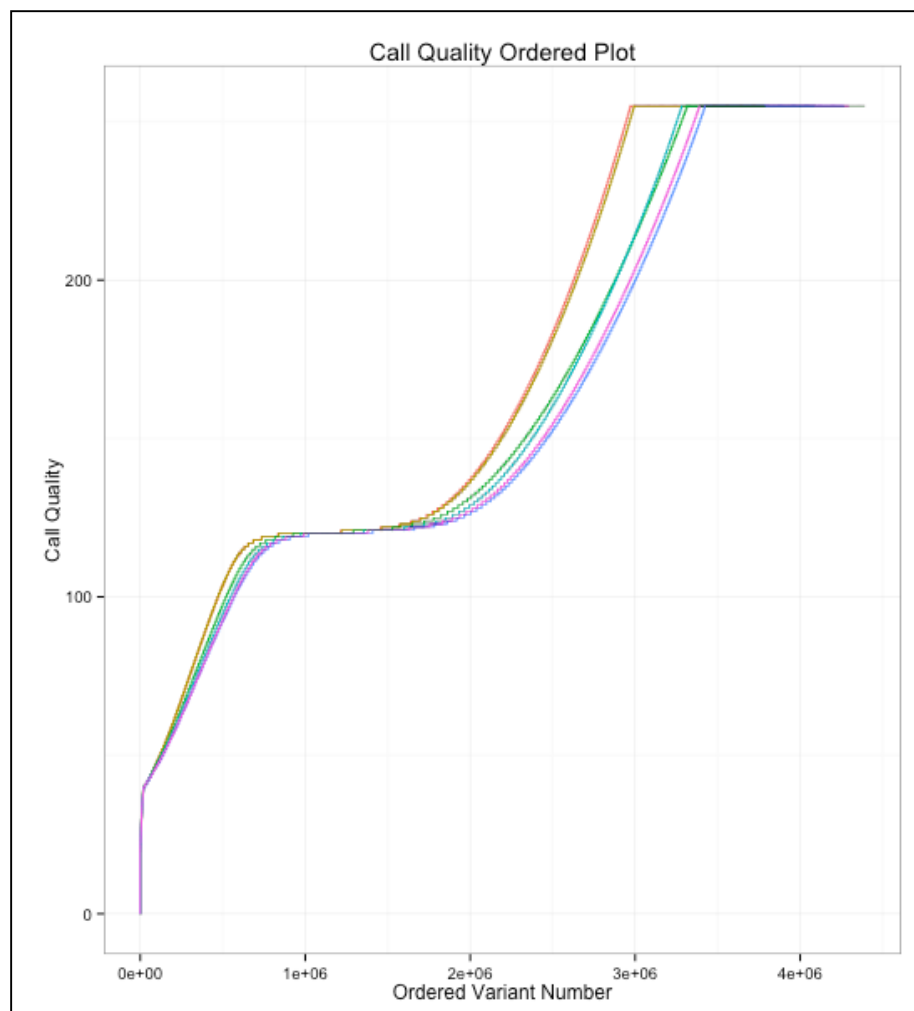
The University of Western Ontario



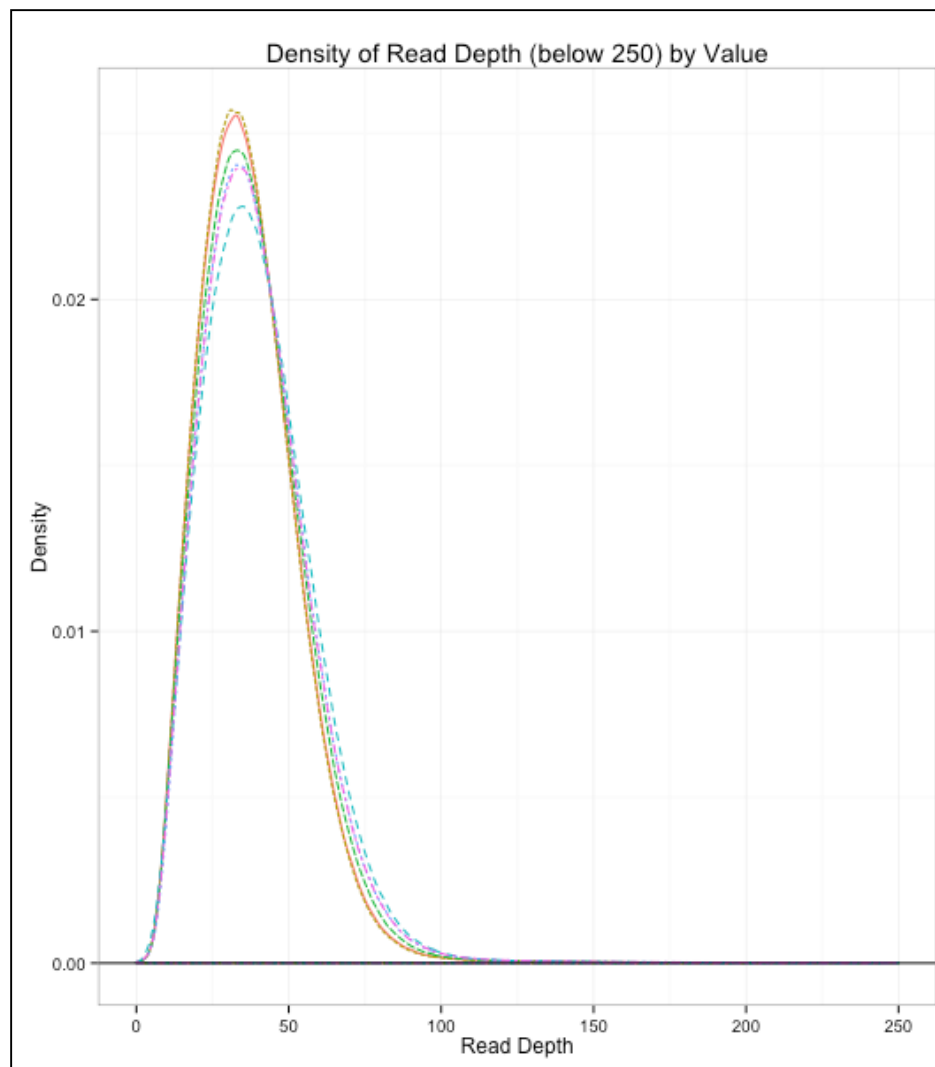
**Figure S1-A.** Circos plot of high confidence *unique* mutations in the affected member of Family 1. These variations were unshared between twins and not found in either parent. Blue represents a gain and red represents a loss. Outside track=SNVs, Indels, Subs, Middle track=CNVs and Inside track= SVs. The arrow represents an inter chromosomal event.



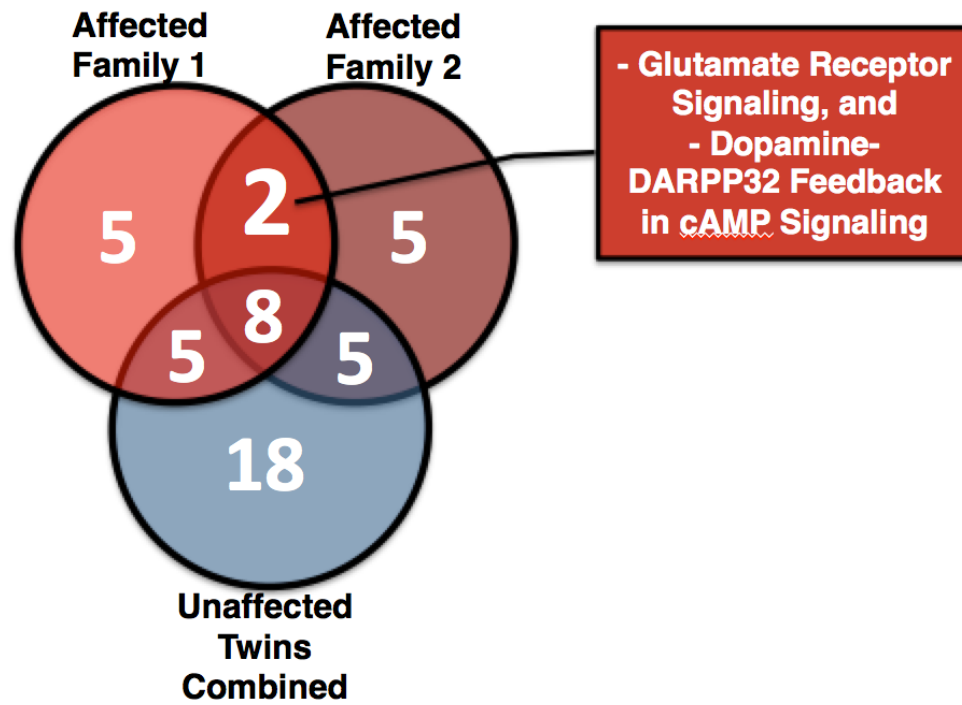
**Figure S1-B.** Circos plot of high confidence *unique* mutations in the affected member of Family 2. These variations were unshared between twins. Blue represents a gain and red represents a loss. Outside track=SNVs, Indels, Subs, Middle track=CNVs and Inside track= SVs.



**Figure S2.** Call Quality of raw variant calls.



**Figure S3.** Read Depth Density of raw variant calls. Outliers (above 250) were not included



**Figure S4.** Venn diagram of shared top 20 canonical pathways found to be overrepresented by variants in the sequencing dataset of affected and unaffected twins. The pathways from the unaffected twin in Family 1 and Family 2 have been merged to represent 36 unique pathways (4 shared). The 18 pathways unique to unaffected twins are broken down into 11 from Family 1 and 7 in Family 2.

**Table S1.** Structural Variants unique to the affected member of Family 1 (I-2-1). Shared identity is based on a 50% reciprocal overlap rule.

| Chr | Start     | End       | Size (bp) | SV Type  | Gene(s)   | Father  | Mother  | Inheritance |
|-----|-----------|-----------|-----------|----------|-----------|---------|---------|-------------|
| 1   | 5717300   | 5725895   | 8595      | Deletion | 0         | Present | Absent  | Inherited   |
| 1   | 5727877   | 5728315   | 438       | Deletion | 0         | Present | Present | Inherited   |
| 1   | 26460142  | 26464816  | 4674      | Deletion | 0         | Present | Absent  | Inherited   |
| 1   | 57242625  | 57242964  | 339       | Deletion | C1orf168  | Present | Present | Inherited   |
| 1   | 62654769  | 62657187  | 2418      | Deletion | 0         | Present | Absent  | Inherited   |
| 1   | 71237363  | 71240327  | 2964      | Deletion | 0         | Present | Absent  | Inherited   |
| 1   | 112691792 | 112704705 | 12913     | Deletion | 0         | Present | Present | Inherited   |
| 1   | 144894289 | 144895285 | 996       | Deletion | PDE4DIP   | Present | Present | Inherited   |
| 1   | 145026746 | 145027069 | 323       | Deletion | PDE4DIP   | Present | Present | Inherited   |
| 1   | 158867539 | 158869984 | 2445      | Deletion | 0         | Present | Present | Inherited   |
| 1   | 238852322 | 238853265 | 943       | Deletion | 0         | Present | Absent  | Inherited   |
| 1   | 248051492 | 248057652 | 6160      | Deletion | 0         | Present | Present | Inherited   |
| 2   | 1801743   | 1802067   | 324       | Deletion | MYT1L     | Present | Absent  | Inherited   |
| 2   | 3822800   | 3823598   | 798       | Deletion | 0         | Present | Present | Inherited   |
| 2   | 33091738  | 33095634  | 3896      | Deletion | LOC285045 | Present | Present | Inherited   |
| 2   | 182565264 | 182565592 | 328       | Deletion | 0         | Present | Absent  | Inherited   |
| 2   | 239626179 | 239627520 | 1341      | Deletion | 0         | Present | Present | Inherited   |
| 3   | 228847    | 234481    | 5634      | Deletion | 0         | Present | Present | Inherited   |
| 3   | 8718847   | 8722982   | 4135      | Deletion | 0         | Present | Absent  | Inherited   |
| 3   | 16239409  | 16241210  | 1801      | Deletion | GALNTL2   | Present | Absent  | Inherited   |
| 3   | 49494287  | 49494616  | 329       | Deletion | 0         | Present | Present | Inherited   |
| 3   | 73939035  | 73939381  | 346       | Deletion | 0         | Present | Absent  | Inherited   |
| 4   | 4568030   | 4568628   | 598       | Deletion | 0         | Present | Absent  | Inherited   |
| 4   | 28511981  | 28513202  | 1221      | Deletion | 0         | Present | Absent  | Inherited   |

|   |           |           |      |          |                   |         |         |           |
|---|-----------|-----------|------|----------|-------------------|---------|---------|-----------|
| 4 | 59428999  | 59435542  | 6543 | Deletion | 0                 | Present | Absent  | Inherited |
| 4 | 73831249  | 73831821  | 572  | Deletion | 0                 | Present | Absent  | Inherited |
| 4 | 76743108  | 76744645  | 1537 | Deletion | 0                 | Present | Present | Inherited |
| 4 | 80888062  | 80894107  | 6045 | Deletion | ANTXR2            | Present | Present | Inherited |
| 4 | 90427886  | 90428672  | 786  | Deletion | 0                 | Present | Absent  | Inherited |
| 4 | 190535505 | 190538363 | 2858 | Deletion | 0                 | Present | Absent  | Inherited |
| 5 | 668063    | 668593    | 530  | Deletion | LOC100132605;TPPP | Present | Present | Inherited |
| 5 | 25959078  | 25959871  | 793  | Deletion | 0                 | Present | Absent  | Inherited |
| 5 | 42628539  | 42631218  | 2679 | Deletion | GHR               | Present | Absent  | Inherited |
| 5 | 95343326  | 95343680  | 354  | Deletion | 0                 | Present | Present | Inherited |
| 5 | 112224253 | 112226906 | 2653 | Deletion | REEP5             | Present | Absent  | Inherited |
| 5 | 126126039 | 126127716 | 1677 | Deletion | LMNB1             | Present | Absent  | Inherited |
| 5 | 127172332 | 127173282 | 950  | Deletion | LOC728586         | Present | Present | Inherited |
| 5 | 176387587 | 176390188 | 2601 | Deletion | UIMC1             | Present | Absent  | Inherited |
| 6 | 14745281  | 14745697  | 416  | Deletion | 0                 | Present | Present | Inherited |
| 6 | 31193512  | 31194660  | 1148 | Deletion | 0                 | Present | Absent  | Inherited |
| 6 | 51739577  | 51745615  | 6038 | Deletion | PKHD1             | Present | Absent  | Inherited |
| 6 | 95193324  | 95194338  | 1014 | Deletion | 0                 | Present | Present | Inherited |
| 6 | 133371501 | 133371845 | 344  | Deletion | 0                 | Present | Absent  | Inherited |
| 6 | 149409430 | 149410432 | 1002 | Deletion | 0                 | Present | Absent  | Inherited |
| 6 | 168059226 | 168059550 | 324  | Deletion | 0                 | Present | Absent  | Inherited |
| 7 | 33898856  | 33899187  | 331  | Deletion | 0                 | Present | Present | Inherited |
| 7 | 36441222  | 36441996  | 774  | Deletion | ANLN              | Present | Absent  | Inherited |

|    |           |           |         |          |                                                                                                                                                                                                                                                                                                                                                                           |         |         |           |
|----|-----------|-----------|---------|----------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------|---------|-----------|
| 7  | 66193913  | 73382120  | 7188207 | Deletion | ABHD11;AUTS2;BAZ1B;BCL7B;C7orf42;CALN1;CLDN3;CLDN4;DNAJC30;FKBP6;FZD9;GTF2IRD2P;LOC100093631;LOC100128031;LOC100131972;LOC100288540;LOC100289307;LOC100289339;LOC442572;LOC644794;LOC729156;MLXIPL;NCF1B;NSUN5;NSUN5C;PMS2L4;POM121;RABGEF1;SBDS;SBDSP;SPDYE7P;SPDYE8P;STAG3L3;STAG3L4;STX1A;TBL2;TRIM50;TRIM74;TYW1;TYW1B;VPS37D;WBSCR17;WBSCR22;WBSCR26;WBSCR27;WBSCR28 | Present | Present | Inherited |
| 7  | 81441673  | 81442580  | 907     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Present | Inherited |
| 7  | 100613739 | 100614479 | 740     | Deletion | MUC12                                                                                                                                                                                                                                                                                                                                                                     | Present | Absent  | Inherited |
| 7  | 102800742 | 102801402 | 660     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 7  | 155199777 | 155201525 | 1748    | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 8  | 40774677  | 40779833  | 5156    | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 8  | 59078464  | 59078800  | 336     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 8  | 113085436 | 113085766 | 330     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Present | Inherited |
| 8  | 144056582 | 144057161 | 579     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 9  | 6700565   | 6710685   | 10120   | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Present | Inherited |
| 9  | 24502071  | 24519103  | 17032   | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Present | Inherited |
| 9  | 110018286 | 110020857 | 2571    | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 9  | 124411502 | 124411804 | 302     | Deletion | DAB2IP                                                                                                                                                                                                                                                                                                                                                                    | Present | Absent  | Inherited |
| 9  | 129492472 | 129492919 | 447     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 9  | 135622738 | 135627436 | 4698    | Deletion | C9orf98                                                                                                                                                                                                                                                                                                                                                                   | Present | Absent  | Inherited |
| 10 | 22081665  | 22082212  | 547     | Deletion | DNAJC1                                                                                                                                                                                                                                                                                                                                                                    | Present | Absent  | Inherited |
| 10 | 78346590  | 78351578  | 4988    | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 10 | 78800911  | 78801230  | 319     | Deletion | KCNMA1                                                                                                                                                                                                                                                                                                                                                                    | Present | Absent  | Inherited |
| 10 | 119634283 | 119634614 | 331     | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Absent  | Inherited |
| 11 | 5784571   | 5809284   | 24713   | Deletion | OR52N1;OR52N5                                                                                                                                                                                                                                                                                                                                                             | Present | Absent  | Inherited |
| 11 | 12152558  | 12152882  | 324     | Deletion | MICAL2                                                                                                                                                                                                                                                                                                                                                                    | Present | Present | Inherited |
| 11 | 63698909  | 63701654  | 2745    | Deletion | 0                                                                                                                                                                                                                                                                                                                                                                         | Present | Present | Inherited |

|    |           |           |        |          |                    |         |         |           |
|----|-----------|-----------|--------|----------|--------------------|---------|---------|-----------|
| 11 | 69400843  | 69401405  | 562    | Deletion | 0                  | Present | Present | Inherited |
| 11 | 102751130 | 102752828 | 1698   | Deletion | LOC100288111       | Present | Absent  | Inherited |
| 12 | 9017095   | 9018122   | 1027   | Deletion | A2ML1              | Present | Present | Inherited |
| 12 | 16420123  | 16421283  | 1160   | Deletion | 0                  | Present | Absent  | Inherited |
| 12 | 99793948  | 99802771  | 8823   | Deletion | ANKS1B             | Absent  | Absent  | De novo   |
| 12 | 112296544 | 112297279 | 735    | Deletion | MAPKAPK5           | Present | Absent  | Inherited |
| 12 | 122634715 | 122650584 | 15869  | Deletion | 0                  | Present | Absent  | Inherited |
| 12 | 127199963 | 127200302 | 339    | Deletion | 0                  | Absent  | Present | Inherited |
| 13 | 21951418  | 21951834  | 416    | Deletion | ZDHC20             | Present | Absent  | Inherited |
| 13 | 81747255  | 81747583  | 328    | Deletion | 0                  | Absent  | Absent  | De novo   |
| 13 | 108560717 | 108561673 | 956    | Deletion | 0                  | Present | Absent  | Inherited |
| 14 | 66157661  | 66158245  | 584    | Deletion | FUT8               | Present | Absent  | Inherited |
| 14 | 73389502  | 73390207  | 705    | Deletion | 0                  | Absent  | Absent  | De novo   |
| 14 | 85139146  | 85139626  | 480    | Deletion | 0                  | Present | Absent  | Inherited |
| 14 | 106932640 | 107174930 | 242290 | Deletion | 0                  | Absent  | Present | Inherited |
| 15 | 39267016  | 39267370  | 354    | Deletion | 0                  | Present | Present | Inherited |
| 15 | 93897281  | 93897642  | 361    | Deletion | 0                  | Absent  | Absent  | De novo   |
| 15 | 101051762 | 101052327 | 565    | Deletion | LASS3              | Present | Present | Inherited |
| 16 | 11683765  | 11685325  | 1560   | Deletion | 0                  | Present | Absent  | Inherited |
| 16 | 25340111  | 25343129  | 3018   | Deletion | 0                  | Present | Present | Inherited |
| 17 | 801626    | 802228    | 602    | Deletion | NXN                | Absent  | Present | Inherited |
| 17 | 5587702   | 5588656   | 954    | Deletion | 0                  | Absent  | Absent  | De novo   |
| 17 | 48427538  | 48427922  | 384    | Deletion | LOC100288444;XYLT2 | Present | Present | Inherited |
| 18 | 108872    | 109083    | 211    | Deletion | 0                  | Absent  | Absent  | De novo   |
| 18 | 52447413  | 52447773  | 360    | Deletion | 0                  | Present | Absent  | Inherited |
| 18 | 63766868  | 63769201  | 2333   | Deletion | 0                  | Present | Present | Inherited |
| 19 | 1182661   | 1183179   | 518    | Deletion | 0                  | Absent  | Absent  | De novo   |

|    |           |           |         |            |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |         |         |           |
|----|-----------|-----------|---------|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------|---------|-----------|
| 19 | 12753340  | 12754497  | 1157    | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Absent  | De novo   |
| 19 | 14732344  | 14734142  | 1798    | Deletion   | EMR3                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Present | Present | Inherited |
| 20 | 23671899  | 23674222  | 2323    | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Present | Absent  | Inherited |
| 20 | 26214307  | 26220376  | 6069    | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Absent  | De novo   |
| 20 | 44535505  | 44535941  | 436     | Deletion   | PLTP                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Absent  | Present | Inherited |
| 22 | 35645166  | 35646141  | 975     | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Present | Inherited |
| 22 | 43101594  | 43102348  | 754     | Deletion   | A4GALT                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Present | Absent  | Inherited |
| 22 | 49063815  | 49064594  | 779     | Deletion   | FAM19A5                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Present | Present | Inherited |
| 22 | 49764594  | 49769018  | 4424    | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Absent  | De novo   |
| X  | 899553    | 899984    | 431     | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Present | Absent  | Inherited |
| X  | 16427688  | 16428223  | 535     | Deletion   | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Present | Inherited |
| X  | 49731220  | 49733608  | 2388    | Deletion   | CLCN5                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | Absent  | Absent  | De novo   |
| 1  | 825765    | 5726936   | 4901171 | Tandem Dup | ACAP3;ACTRT2;AGRN;AJAP1;ARHGEF16;ATAD3A;ATAD3B;ATAD3C;AURKAIP1;B3GALT6;C1orf159;C1orf170;C1orf174;C1orf222;C1orf70;C1orf86;C1orf93;CALML6;CCDC27;CCNL2;CDC2L1;CDC2L2;CPSF3L;DFFB;DVL1;FAM132A;FLJ14100;FLJ39609;FLJ42875;GABRD;GLTPD1;GNB1;HES4;HES5;ISG15;KIAA0495;KIAA0562;KIAA1751;KLHL17;LOC100128003;LOC100128838;LOC100129381;LOC100129534;LOC100131742;LOC100132814;LOC100133612;LOC100287685;LOC100287750;LOC100287848;LOC100287898;LOC100288202;LOC100288271;LOC100288313;LOC100288379;LOC100288479;LOC115110;LOC148413;LOC284661;LOC388588;LOC401934;LOC441869;LOC643988;LOC728661;LOC728716;LRRC47;MEGF6;MIB2;MMEL1;MMP23A;MMP23B;MORN1;MRPL20;MXRA8;NADK;NOC2L;PANK4;PEX10;PLCH2;PLEKHN1;PRDM16;PRKCZ;PUSL1;RER1;SAMD11;SCNN1D;SDF4;SKI;SLC35E2;SSU72;TAS1R3;TMEM52;TMEM88B;TNFRSF14;TNFRSF18;TNFRSF4;TP73;TPRG1L;TTLL10;UBE2J2;VWA1;WDR8 | Absent  | Absent  | De novo   |
| 1  | 230107606 | 230107783 | 177     | Tandem Dup | 0                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Absent  | Absent  | De novo   |

|    |                         |                         |       |            |             |         |         |           |
|----|-------------------------|-------------------------|-------|------------|-------------|---------|---------|-----------|
| 2  | 1426068                 | 1522926                 | 96858 | Tandem Dup | TPO         | Absent  | Present | Inherited |
| 2  | 2649502                 | 2649583                 | 81    | Tandem Dup | 0           | Present | Absent  | Inherited |
| 3  | 75699636                | 75699709                | 73    | Tandem Dup | 0           | Present | Absent  | Inherited |
| 4  | 80273622                | 80274729                | 1107  | Tandem Dup | 0           | Absent  | Absent  | De novo   |
| 5  | 178012521               | 178012708               | 187   | Tandem Dup | COL23A1     | Absent  | Present | Inherited |
| 6  | 1053869                 | 1054275                 | 406   | Tandem Dup | LOC285768   | Absent  | Absent  | De novo   |
| 7  | 605977                  | 606269                  | 292   | Tandem Dup | PRKAR1B     | Present | Present | Inherited |
| 7  | 140181586               | 140189220               | 7634  | Tandem Dup | 0           | Present | Present | Inherited |
| 10 | 125919520               | 125919748               | 228   | Tandem Dup | 0           | Absent  | Absent  | De novo   |
| 10 | 132294621               | 132294747               | 126   | Tandem Dup | 0           | Absent  | Absent  | De novo   |
| 11 | 131550263               | 131550706               | 443   | Tandem Dup | NTM         | Absent  | Absent  | De novo   |
| 14 | 24369410                | 24369615                | 205   | Tandem Dup | 0           | Absent  | Absent  | De novo   |
| 14 | 66258294                | 66258410                | 116   | Tandem Dup | 0           | Present | Present | Inherited |
| 15 | 25468396                | 25470000                | 1604  | Tandem Dup | SNORD115-29 | Absent  | Absent  | De novo   |
| 15 | 63185677                | 63185745                | 68    | Tandem Dup | 0           | Absent  | Present | Inherited |
| 19 | 544936                  | 545072                  | 136   | Tandem Dup | GZMM        | Absent  | Absent  | De novo   |
| 19 | 23262787                | 23265543                | 2756  | Tandem Dup | 0           | Present | Absent  | Inherited |
| 20 | 24510425                | 24510694                | 269   | Tandem Dup | C20orf39    | Present | Absent  | Inherited |
| 2  | 61702932/<br>61700753   | 61703455/<br>61700765   | 523   | Distal Dup | 0           | Absent  | Present | Inherited |
| 5  | 150319009/<br>150319494 | 150319103/<br>150319759 | 94    | Distal Dup | LOC134466   | Present | Absent  | Inherited |
| 12 | 12544853/<br>12546495   | 12544955/<br>12546622   | 102   | Distal Dup | LOH12CR1    | Present | Present | Inherited |
| 12 | 87205701/<br>87204798   | 87205819/<br>87204806   | 118   | Distal Dup | MGAT4C      | Present | Absent  | Inherited |
| 13 | 21538303/<br>21545834   | 21541177/<br>21545834   | 2874  | Distal Dup | 0           | Present | Absent  | Inherited |
| 16 | 69761819/<br>69762887   | 69762131/<br>69763048   | 312   | Distal Dup | 0           | Absent  | Present | Inherited |

|    |                       |                       |          |            |                                |         |         |           |
|----|-----------------------|-----------------------|----------|------------|--------------------------------|---------|---------|-----------|
| 18 | 69711799/<br>69712885 | 69712015/<br>69712888 | 216      | Distal Dup | 0                              | Absent  | Present | Inherited |
| 23 | 2141240/<br>2143040   | 2141701/<br>2143092   | 461      | Distal Dup | DHRX;LOC100288477;LOC100288983 | Present | Absent  | Inherited |
| 23 | 52886722/55678<br>952 | 52892121/5567<br>9111 | 5399     | Distal Dup | XAGE-4;XAGE3                   | Absent  | Present | Inherited |
| 2  | 133022029             | 17                    | 45213172 | Interchrom | CDC27                          | Absent  | Absent  | De novo   |

**Table S2.** Structural Variants unique to the affected member of Family 2 (II-2-1). Shared identity is based on a 50% reciprocal overlap rule.

| Chr | Start     | End       | Size (bp) | SV Type  | Gene(s) |
|-----|-----------|-----------|-----------|----------|---------|
| 1   | 1142719   | 1143140   | 421       | Deletion | 0       |
| 1   | 56401293  | 56401614  | 321       | Deletion | 0       |
| 1   | 62390602  | 62390951  | 349       | Deletion | INADL   |
| 1   | 85748634  | 85748964  | 330       | Deletion | 0       |
| 1   | 91914111  | 91914566  | 455       | Deletion | 0       |
| 1   | 207292362 | 207293195 | 833       | Deletion | C4BPA   |
| 1   | 222034158 | 222034490 | 332       | Deletion | 0       |
| 1   | 228572175 | 228572504 | 329       | Deletion | 0       |
| 2   | 8616813   | 8617120   | 307       | Deletion | 0       |
| 2   | 16271827  | 16274059  | 2232      | Deletion | 0       |
| 2   | 39071490  | 39071832  | 342       | Deletion | DHX57   |
| 2   | 53625772  | 53628009  | 2237      | Deletion | 0       |
| 2   | 88821957  | 88822489  | 532       | Deletion | 0       |
| 2   | 194689502 | 194698766 | 9264      | Deletion | 0       |
| 2   | 206731199 | 206731522 | 323       | Deletion | 0       |
| 2   | 226039583 | 226040060 | 477       | Deletion | 0       |
| 3   | 10397475  | 10397834  | 359       | Deletion | ATP2B2  |
| 3   | 23764073  | 23764403  | 330       | Deletion | 0       |
| 3   | 144888449 | 144888910 | 461       | Deletion | 0       |
| 3   | 185017344 | 185017669 | 325       | Deletion | 0       |
| 4   | 3612027   | 3612659   | 632       | Deletion | 0       |
| 4   | 29661122  | 29661450  | 328       | Deletion | 0       |
| 4   | 43931496  | 43931837  | 341       | Deletion | 0       |
| 4   | 44509394  | 44509712  | 318       | Deletion | 0       |

|   |           |           |      |          |           |
|---|-----------|-----------|------|----------|-----------|
| 4 | 58087343  | 58087741  | 398  | Deletion | 0         |
| 4 | 92280245  | 92285214  | 4969 | Deletion | KIAA1680  |
| 4 | 100023050 | 100023385 | 335  | Deletion | 0         |
| 4 | 149990582 | 149992581 | 1999 | Deletion | 0         |
| 4 | 181549440 | 181549754 | 314  | Deletion | 0         |
| 4 | 189969695 | 189970049 | 354  | Deletion | 0         |
| 4 | 190624640 | 190624987 | 347  | Deletion | 0         |
| 5 | 42165941  | 42167782  | 1841 | Deletion | 0         |
| 5 | 45004820  | 45014528  | 9708 | Deletion | 0         |
| 5 | 68724047  | 68724498  | 451  | Deletion | MARVELD2  |
| 5 | 93916299  | 93916636  | 337  | Deletion | C5orf36   |
| 5 | 127172332 | 127173282 | 950  | Deletion | LOC728586 |
| 5 | 177821921 | 177823857 | 1936 | Deletion | COL23A1   |
| 5 | 178106931 | 178111943 | 5012 | Deletion | 0         |
| 6 | 857494    | 857907    | 413  | Deletion | 0         |
| 6 | 11556106  | 11556426  | 320  | Deletion | TMEM170B  |
| 6 | 32624895  | 32625860  | 965  | Deletion | 0         |
| 6 | 33026728  | 33028610  | 1882 | Deletion | 0         |
| 6 | 33125920  | 33126843  | 923  | Deletion | 0         |
| 6 | 50765971  | 50766282  | 311  | Deletion | 0         |
| 6 | 57297190  | 57301246  | 4056 | Deletion | PRIM2     |
| 6 | 106983636 | 106983955 | 319  | Deletion | AIM1      |
| 6 | 119417519 | 119417847 | 328  | Deletion | FAM184A   |
| 6 | 133371501 | 133371845 | 344  | Deletion | 0         |
| 6 | 134062939 | 134063433 | 494  | Deletion | 0         |
| 6 | 154446930 | 154447253 | 323  | Deletion | OPRM1     |
| 7 | 9634650   | 9635993   | 1343 | Deletion | 0         |

|    |           |           |       |          |          |
|----|-----------|-----------|-------|----------|----------|
| 7  | 42549714  | 42550036  | 322   | Deletion | 0        |
| 7  | 158029478 | 158030452 | 974   | Deletion | PTPRN2   |
| 8  | 98595409  | 98595748  | 339   | Deletion | 0        |
| 9  | 71895487  | 71896647  | 1160  | Deletion | 0        |
| 9  | 87775285  | 87776256  | 971   | Deletion | 0        |
| 9  | 129520461 | 129523267 | 2806  | Deletion | 0        |
| 9  | 136379222 | 136379564 | 342   | Deletion | 0        |
| 10 | 35593222  | 35593552  | 330   | Deletion | CCNY     |
| 10 | 57702043  | 57702383  | 340   | Deletion | 0        |
| 10 | 107253002 | 107253355 | 353   | Deletion | 0        |
| 10 | 114157801 | 114158139 | 338   | Deletion | ACSL5    |
| 11 | 7716915   | 7717231   | 316   | Deletion | OVCH2    |
| 11 | 31916727  | 31917056  | 329   | Deletion | 0        |
| 11 | 32286653  | 32286986  | 333   | Deletion | 0        |
| 11 | 33537687  | 33538010  | 323   | Deletion | 0        |
| 11 | 65642111  | 65643526  | 1415  | Deletion | 0        |
| 11 | 104755159 | 104762566 | 7407  | Deletion | CASP12   |
| 12 | 124458567 | 124496090 | 37523 | Deletion | ZNF664   |
| 12 | 130060303 | 130060634 | 331   | Deletion | TMEM132D |
| 13 | 23055219  | 23055526  | 307   | Deletion | 0        |
| 13 | 81747255  | 81747583  | 328   | Deletion | 0        |
| 13 | 93056847  | 93057422  | 575   | Deletion | GPC5     |
| 14 | 35428814  | 35432457  | 3643  | Deletion | 0        |
| 14 | 38643511  | 38643828  | 317   | Deletion | 0        |
| 14 | 42486335  | 42486650  | 315   | Deletion | 0        |
| 14 | 76813774  | 76814209  | 435   | Deletion | 0        |
| 14 | 78828245  | 78828596  | 351   | Deletion | 0        |

|    |           |           |       |          |              |
|----|-----------|-----------|-------|----------|--------------|
| 14 | 100845014 | 100845347 | 333   | Deletion | WDR25        |
| 15 | 22336853  | 22344093  | 7240  | Deletion | LOC727924    |
| 15 | 39372537  | 39373645  | 1108  | Deletion | 0            |
| 15 | 41864762  | 41865200  | 438   | Deletion | TYRO3        |
| 15 | 42017748  | 42018080  | 332   | Deletion | MGA          |
| 15 | 50946438  | 50947428  | 990   | Deletion | TRPM7        |
| 15 | 67083771  | 67084069  | 298   | Deletion | 0            |
| 15 | 83773883  | 83774212  | 329   | Deletion | 0            |
| 15 | 91173990  | 91174326  | 336   | Deletion | CRTC3        |
| 15 | 91981580  | 91989268  | 7688  | Deletion | 0            |
| 15 | 93515798  | 93516111  | 313   | Deletion | CHD2         |
| 16 | 12002336  | 12002661  | 325   | Deletion | GSPT1        |
| 16 | 23910887  | 23911193  | 306   | Deletion | PRKCB        |
| 16 | 86284321  | 86284629  | 308   | Deletion | 0            |
| 17 | 136772    | 138726    | 1954  | Deletion | RPH3AL       |
| 17 | 193726    | 197057    | 3331  | Deletion | RPH3AL       |
| 17 | 41382745  | 41399871  | 17126 | Deletion | 0            |
| 17 | 41383489  | 41466010  | 82521 | Deletion | LOC100130581 |
| 17 | 46904824  | 46907714  | 2890  | Deletion | 0            |
| 17 | 47228439  | 47228769  | 330   | Deletion | B4GALNT2     |
| 17 | 57363991  | 57365775  | 1784  | Deletion | 0            |
| 17 | 69852154  | 69852481  | 327   | Deletion | 0            |
| 18 | 1199564   | 1200310   | 746   | Deletion | TFG/GPR128   |
| 18 | 3997022   | 3997479   | 457   | Deletion | 0            |
| 18 | 22892494  | 22892818  | 324   | Deletion | ZNF521       |
| 18 | 27629690  | 27630016  | 326   | Deletion | 0            |
| 18 | 37061646  | 37061962  | 316   | Deletion | LOC647946    |

|    |           |           |        |            |                        |
|----|-----------|-----------|--------|------------|------------------------|
| 18 | 43261014  | 43261339  | 325    | Deletion   | SLC14A2                |
| 18 | 44338433  | 44338770  | 337    | Deletion   | 0                      |
| 18 | 50462567  | 50463032  | 465    | Deletion   | DCC                    |
| 19 | 1182661   | 1183179   | 518    | Deletion   | 0                      |
| 19 | 2128535   | 2129051   | 516    | Deletion   | AP3D1                  |
| 19 | 45473177  | 45474322  | 1145   | Deletion   | CLPTM1                 |
| 20 | 25583242  | 25583678  | 436    | Deletion   | 0                      |
| 20 | 33241939  | 33244364  | 2425   | Deletion   | PIGU                   |
| 20 | 53292665  | 53292978  | 313    | Deletion   | 0                      |
| 21 | 16588380  | 16591454  | 3074   | Deletion   | 0                      |
| 21 | 36072707  | 36073562  | 855    | Deletion   | CLIC6                  |
| 21 | 39590542  | 39590888  | 346    | Deletion   | 0                      |
| 22 | 33295772  | 33296090  | 318    | Deletion   | SYN3                   |
| 22 | 49622515  | 49624301  | 1786   | Deletion   | 0                      |
| X  | 31577478  | 31577794  | 316    | Deletion   | DMD                    |
| X  | 85605177  | 85609869  | 4692   | Deletion   | DACH2                  |
| X  | 138384911 | 138385237 | 326    | Deletion   | 0                      |
| 1  | 30569469  | 30965397  | 395928 | Tandem Dup | 0                      |
| 2  | 242929421 | 242929637 | 216    | Tandem Dup | 0                      |
| 3  | 139670597 | 139674454 | 3857   | Tandem Dup | CLSTN2                 |
| 4  | 190478831 | 190479023 | 192    | Tandem Dup | 0                      |
| 6  | 160877802 | 160956684 | 78882  | Tandem Dup | LOC100289195;LPA;LPAL2 |
| 7  | 606117    | 606324    | 207    | Tandem Dup | PRKAR1B                |
| 7  | 156124573 | 156124795 | 222    | Tandem Dup | 0                      |
| 9  | 44070664  | 44070787  | 123    | Tandem Dup | LOC100289454           |
| 9  | 140563766 | 140564031 | 265    | Tandem Dup | 0                      |
| 10 | 27224456  | 27229304  | 4848   | Tandem Dup | C10orf51               |

|    |                         |                     |         |            |                                                                                                                                                                                                                                                                                                 |
|----|-------------------------|---------------------|---------|------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| 13 | 34811550                | 34815591            | 4041    | Tandem Dup | 0                                                                                                                                                                                                                                                                                               |
| 14 | 105943990               | 105944114           | 124     | Tandem Dup | CRIP2                                                                                                                                                                                                                                                                                           |
| 19 | 14706528                | 14706835            | 307     | Tandem Dup | CLEC17A                                                                                                                                                                                                                                                                                         |
| X  | 52886720                | 55678950            | 2792230 | Tandem Dup | XAGE4;ALAS2;APEX2;FAM104B;FAM120C;FAM156A;FAM156B;FGD1;FOXR2;GNL3L;GPR173;HSD17B10;HUWE1;IQSEC2;ITIH5L;KDM5C;LOC100132984;LOC100288024;LOC100288052;LOC100288498;LOC100288560;LOC644893;MAGED2;MAGEH1;PAGE2;PAGE2B;PAGE3;PAGE5;PFKFB1;PHF8;RIBC1;SMC1A;SNORA11;TRO;TSPYL2;TSR2;USP51;WNK3;XAGE3 |
| 2  | 16407764/<br>16406392   | 16407988/16406410   | 224     | Distal Dup | 0                                                                                                                                                                                                                                                                                               |
| 3  | 194546260/<br>194543294 | 194546429/194543309 | 169     | Distal Dup | 0                                                                                                                                                                                                                                                                                               |
| 7  | 100550585/<br>100551244 | 100550785/100551247 | 200     | Distal Dup | MUC3A                                                                                                                                                                                                                                                                                           |
| 9  | 107816635/<br>107817341 | 107816979/107817348 | 344     | Distal Dup | 0                                                                                                                                                                                                                                                                                               |
| 23 | 17060848/<br>17063276   | 17061023/17063295   | 175     | Distal Dup | REPS2                                                                                                                                                                                                                                                                                           |