

Supplementary Table S3. Diagnostic CNVs identified in the cohort.

| Sample | Phenotype | Status | Copy number | Region (hg19) | Size (kb) | Cytoband |
|--------|-----------|--------|-------------|------------------------|-----------|-----------------|
| S0016 | ID | DUP | 3 | 5:176814762-178413984 | 1599.222 | 5q35.3 |
| S0020 | MA | DEL | 1 | 8:8559447-11617511 | 3058.064 | 8p23.1 |
| S0021 | ID | DUP | 3 | 6:391738-7586950 | 7195.212 | 6p25.3-p24.3 |
| S0027 | MD | DEL | 1 | 19:17986760-17994855 | 8.095 | 19p13.11 |
| S0048 | ID | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0051 | EP, MA | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0058 | ID | DUP | 2 | X:53220502-54071682 | 851.18 | Xp11.22 |
| S0061 | BT | DUP | 3 | 8:140630248-145743210 | 5112.962 | 8q24.3 |
| S0183 | ID | DEL | 1 | 9:139981378-139982635 | 1.257 | 9q34.3 |
| S0201 | EP | DUP | 3 | 7:73442118-74191693 | 749.575 | 7q11.23 |
| S0212 | EP | DEL | 0 | 19:1401295-1401396 | 0.101 | 19p13.3 |
| S0228 | MA | DEL | 1 | 16:772083-2115636 | 1343.553 | 16p13.3 |
| S0229 | MA | DEL | 1 | 7:146471362-151574210 | 5102.848 | 7q35-q36.1 |
| S0229 | MA | DEL | 1 | 7:142985307-144532711 | 1547.404 | 7q34-q35 |
| S0249 | EP, MA | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0280 | MA | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0283 | ID | DEL | 1 | 18:71920530-77514510 | 5593.98 | 18q22.3-q23 |
| S0318 | EP, MA | DEL | 1 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S0329 | EP | DEL | 1 | 1:955502-2344010 | 1388.508 | 1p36.33-p36.32 |
| S0355 | MA | DEL | 1 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S0359 | EP | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0366 | MA | DUP | 3 | 10:118888031-124274424 | 5386.393 | 10q25.3-q26.13 |
| S0366 | MA | DUP | 3 | 10:124768494-127505201 | 2736.707 | 10q26.13-q26.2 |
| S0372 | MA | DEL | 1 | 22:22123328-24237719 | 2114.391 | 22q11.21-q11.23 |

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| S0387 | MA | DUP | 3 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0391 | EP, MA | DEL | 1 | 17:2541392-3564836 | 1023.444 | 17p13.3-p13.2 |
| S0391 | EP, MA | DEL | 1 | 17:4800498-6441402 | 1640.904 | 17p13.2 |
| S0392 | ID | DUP | 3 | 5:10971951-16617167 | 5645.216 | 5p15.2-p15.1 |
| S0392 | ID | DEL | 1 | 5:1201709-10266524 | 9064.815 | 5p15.33-p15.2 |
| S0434 | MA | DEL | 1 | 7:73442118-74203657 | 761.539 | 7q11.23 |
| S0462 | EP | DEL | 1 | 20:61448918-62680994 | 1232.076 | 20q13.33 |
| S0466 | MA | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0472 | MD, MA | DEL | 1 | 5:176047084-177423243 | 1376.159 | 5q35.2-q35.3 |
| S0474 | ID | DUP | 3 | 1:196946666-247612410 | 50665.744 | 1q31.3-q44 |
| S0482 | MD | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0485 | BT, MA | DEL | 1 | 19:12757326-14041692 | 1284.366 | 19p13.2-p13.12 |
| S0492 | MA | DEL | 1 | 14:102430864-105260865 | 2830.001 | 14q32.31-q32.33 |
| S0514 | MD, MA | DEL | 1 | 11:124735281-134135749 | 9400.468 | 11q24.2-q25 |
| S0532 | EP | DEL | 1 | 4:68605045-72437804 | 3832.759 | 4q13.2-q13.3 |
| S0543 | MA | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0553 | EP, MA | DEL | 1 | 6:116440085-118881893 | 2441.808 | 6q22.1-q22.31 |
| S0562 | MD | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0571 | MD | DEL | 1 | 9:213107-6645650 | 6432.543 | 9p24.3-p24.1 |
| S0573 | EP | DEL | 1 | 10:81697495-87966405 | 6268.91 | 10q22.3-q23.2 |
| S0593 | ID | DEL | 1 | 16:74760122-74808729 | 48.607 | 16q23.1 |
| S0599 | MA | DUP | 3 | 12:389294-15750333 | 15361.039 | 12p13.33-p12.3 |
| S0599 | MA | DUP | 3 | 12:21007961-28123282 | 7115.321 | 12p12.2-p11.22 |
| S0607 | ID | DEL | 0 | X:30709239-30709316 | 0.077 | Xp21.2 |
| S0626 | MD, MA | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0655 | MA | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |

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| S0664 | MA | DEL | 1 | 6:129419317-129419560 | 0.243 | 6q22.33 |
| S0666 | ID | DEL | 0 | X:2852698-5950791 | 3098.093 | Xp22.33-p22.32 |
| S0674 | ID | DEL | 1 | 4:619372-3502552 | 2883.18 | 4p16.3 |
| S0683 | EP | DEL | 1 | 16:29802039-30200397 | 398.358 | 16p11.2 |
| S0684 | ID | DEL | 1 | 13:111077080-114440708 | 3363.628 | 13q34 |
| S0688 | ID | DEL | 1 | 1:145414692-147381393 | 1966.701 | 1q21.1-q21.2 |
| S0696 | ID | DUP | 3 | 1:201008641-216595882 | 15587.241 | 1q32.1-q41 |
| S0700 | MA | DUP | 3 | 22:41865128-51171726 | 9306.598 | 22q13.2-q13.33 |
| S0702 | ID | DEL | 1 | 8:1719097-10480730 | 8761.633 | 8p23.3-p23.1 |
| S0703 | MA | DUP | 3 | X:95939661-111003873 | 15064.212 | Xq21.33-q23 |
| S0734 | ID | DEL | 1 | 1:18957499-22456344 | 3498.845 | 1p36.13-p36.12 |
| S0737 | ID | DEL | 1 | 19:12917393-13710781 | 793.388 | 19p13.2 |
| S0742 | BT | DUP | 3 | 8:23559963-38962520 | 15402.557 | 8p21.2-p11.22 |
| S0746 | ID | DEL | 1 | 17:17115525-18083116 | 967.591 | 17p11.2 |
| S0756 | ID | DEL | 1 | 17:1303340-1680868 | 377.528 | 17p13.3 |
| S0758 | ID | DEL | 1 | 6:83074339-88299721 | 5225.382 | 6q14.1-q15 |
| S0759 | MA | DUP | 3 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0763 | ID | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0774 | BT | DEL | 0 | X:32429869-32430030 | 0.161 | Xp21.1 |
| S0780 | BT | DUP | 3 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0783 | MA | DEL | 1 | 4:619372-3245676 | 2626.304 | 4p16.3 |
| S0785 | ID | DEL | 1 | 10:90694830-94415150 | 3720.32 | 10q23.31-q23.33 |
| S0786 | ID | DEL | 1 | 16:55690351-58055522 | 2365.171 | 16q12.2-q21 |
| S0787 | ID | DEL | 1 | 2:219135114-223920355 | 4785.241 | 2q35-q36.1 |
| S0793 | ID | DUP | 3 | 6:391738-7586950 | 7195.212 | 6p25.3-p24.3 |
| S0797 | ID | DEL | 1 | 7:73442118-74203657 | 761.539 | 7q11.23 |

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| S0798 | BT, MA | DEL | 1 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S0800 | BT, MA | DEL | 1 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S0805 | BT, MA | DEL | 1 | 7:73442118-74203657 | 761.539 | 7q11.23 |
| S0806 | MA | DUP | 3 | 3:3191694-24270492 | 21078.798 | 3p26.2-p24.2 |
| S0808 | ID | DEL | 1 | 13:77564794-84456528 | 6891.734 | 13q22.3-q31.1 |
| S0810 | MA | DEL | 1 | 17:16836096-19580911 | 2744.815 | 17p11.2 |
| S0811 | MA | DEL | 1 | 4:619372-8873543 | 8254.171 | 4p16.3-p16.1 |
| S0812 | MA | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0813 | MA | DEL | 1 | 5:1201709-16617167 | 15415.458 | 5p15.33-p15.1 |
| S0816 | MA | DEL | 1 | 7:148504475-157210133 | 8705.658 | 7q36.1-q36.3 |
| S0818 | MA | DEL | 1 | 7:73442118-74148323 | 706.205 | 7q11.23 |
| S0821 | MD, MA | DEL | 1 | 22:18560688-20792146 | 2231.458 | 22q11.21 |
| S0824 | ID | DEL | 1 | 2:148695714-149868197 | 1172.483 | 2q22.3-q23.1 |
| S0825 | ID | DEL | 1 | 5:1201709-16617167 | 15415.458 | 5p15.33-p15.1 |
| S0827 | MA | DEL | 1 | 7:73442118-74148323 | 706.205 | 7q11.23 |
| S0831 | ID | DEL | 1 | 18:55221491-77514510 | 22293.02 | 18q21.31-q23 |
| S0852 | MA | DEL | 1 | 17:16836096-19580911 | 2744.815 | 17p11.2 |
| S0861 | MA | DUP | 3 | 9:20341662-33167354 | 12825.692 | 9p21.3-p21.1 |
| S0861 | MA | DUP | 3 | 9:34483445-37785061 | 3301.616 | 9p13.3-p13.2 |
| S0863 | MA | DEL | 1 | 6:139693392-146758734 | 7065.342 | 6q24.1-q24.3 |
| S0864 | MD, MA | DEL | 1 | 4:619372-10027630 | 9408.258 | 4p16.3-p16.1 |
| S0875 | ID | DUP | 3 | 2:9628275-21266945 | 11638.67 | 2p25.1-p24.1 |
| S0875 | ID | DUP | 3 | 2:25383721-32382706 | 6998.985 | 2p23.3-p22.3 |
| S0875 | ID | DUP | 3 | 2:1418179-3750261 | 2332.082 | 2p25.3 |
| S0894 | ID | DEL | 1 | 16:29802039-30200397 | 398.358 | 16p11.2 |
| S0895 | MA | DUP | 3 | 5:10250032-16617167 | 6367.135 | 5p15.2-p15.1 |

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| S0895 | MA | DEL | 1 | 5:1201709-7901237 | 6699.528 | 5p15.33-p15.31 |
| S0897 | MA | DEL | 1 | 7:73442118-74133260 | 691.142 | 7q11.23 |
| S0911 | MA | DEL | 1 | 2:238232654-242708226 | 4475.572 | 2q37.3 |
| S0911 | MA | DUP | 3 | 4:619372-6304992 | 5685.62 | 4p16.3-p16.1 |
| S0916 | ID | DEL | 1 | 5:1201709-14871887 | 13670.178 | 5p15.33-p15.2 |
| S0928 | MA | DEL | 0 | X:148558520-148568844 | 10.324 | Xq28 |
| S0930 | ID | DEL | 1 | 3:119013219-120401418 | 1388.199 | 3q13.33 |
| S0934 | ID | DUP | 3 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0937 | ID | DEL | 1 | 15:31196714-32455536 | 1258.822 | 15q13.2-q13.3 |
| S0938 | EP, MA | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0942 | ID | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0944 | BT | DUP | 3 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S0955 | ID | DEL | 1 | 7:73442118-74173185 | 731.067 | 7q11.23 |
| S0958 | ID | DEL | 1 | 5:89825264-95768984 | 5943.72 | 5q14.3-q15 |
| S0971 | MA | DEL | 1 | 3:3191694-10193904 | 7002.21 | 3p26.2-p25.3 |
| S0974 | ID | DUP | 2 | X:153194694-153588955 | 394.261 | Xq28 |
| S0979 | MD | DUP | 3 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S0990 | ID | DUP | 4 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S1000 | MA | DEL | 1 | 4:619372-4865663 | 4246.291 | 4p16.3-p16.2 |
| S1003 | BT | DEL | 1 | X:28807436-38546928 | 9739.492 | Xp21.3-p11.4 |
| S1005 | ID | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S1011 | ID | DEL | 1 | 1:240070732-244220778 | 4150.046 | 1q43-q44 |
| S1013 | MA | DEL | 1 | 15:23888690-28327041 | 4438.351 | 15q11.2-q13.1 |
| S1022 | ID | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S1023 | ID | DUP | 3 | 22:20778873-36745300 | 15966.427 | 22q11.21-q12.3 |
| S1031 | ID | DEL | 1 | 2:234235766-242708226 | 8472.46 | 2q37.1-q37.3 |

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| S1032 | MD, MA | DEL | 1 | 16:8829555-8906963 | 77.408 | 16p13.2 |
| S1033 | BT, MA | DUP | 3 | 7:73442118-74203657 | 761.539 | 7q11.23 |
| S1039 | MA | DEL | 1 | 22:46731321-51171726 | 4440.405 | 22q13.31-q13.33 |
| S1040 | MA | DUP | 4 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S1053 | MD, MA | DUP | 3 | 16:29802039-30081741 | 279.702 | 16p11.2 |
| S1054 | MA | DUP | 3 | 16:74750244-90005169 | 15254.925 | 16q23.1-q24.3 |
| S1054 | MA | DUP | 3 | 16:70514471-72058954 | 1544.483 | 16q22.1-q22.2 |
| S1055 | ID | DUP | 2 | X:152801579-153363212 | 561.633 | Xq28 |
| S1056 | ID | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S1057 | ID | DUP | 3 | 15:23043276-28327041 | 5283.765 | 15q11.2-q13.1 |
| S1058 | BT | DUP | 3 | 16:29802039-30081741 | 279.702 | 16p11.2 |
| S1059 | ID | DUP | 3 | 1:228345440-247612410 | 19266.97 | 1q42.13-q44 |
| S1060 | ID | DEL | 1 | 17:16836096-19580911 | 2744.815 | 17p11.2 |
| S1061 | MA | DEL | 1 | 6:108225832-112575917 | 4350.085 | 6q21 |
| S1063 | EP, MA | DUP | 3 | 17:76987798-80790512 | 3802.714 | 17q25.3 |
| S1063 | EP, MA | DEL | 1 | 20:61448413-62680994 | 1232.581 | 20q13.33 |
| S1078 | ID | DEL | 1 | 8:90945563-97247862 | 6302.299 | 8q21.3-q22.1 |
| S1079 | ID | DEL | 1 | 4:186422850-190873442 | 4450.592 | 4q35.1-q35.2 |
| S1080 | MA | DEL | 1 | 18:67671028-77514510 | 9843.482 | 18q22.2-q23 |
| S1081 | ID | DUP | 3 | X:48542167-50659607 | 2117.44 | Xp11.23-p11.22 |
| S1082 | EP, MA | DEL | 1 | 1:240070732-247612410 | 7541.678 | 1q43-q44 |
| S1082 | EP, MA | DEL | 1 | 1:236881157-237997288 | 1116.131 | 1q43 |
| S1084 | MD, EP | DEL | 1 | 22:18900293-21245506 | 2345.213 | 22q11.21 |
| S1090 | ID | DEL | 1 | 16:15758583-15932126 | 173.543 | 16p13.11 |

ID: Isolated DD; **MA**: DD with malformations; **EP**: DD with epilepsy; **BT**: DD with behavioral troubles; **MD**: DD with metabolic disorder.