

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

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

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

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

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

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.