Table S1: Technical sequencing results on all 106 samples

Batch#	Sample ID	# input reads	Read mapped (%)	Median coverage	>1X (%)	>10X (%)	>40X (%)	>80X (%)	Multi-mapped reads (%)	Duplicate reads (%)	Reads left after filtering (%)	Reads in target regions (%) before filtering	Reads in target regions (%) after filtering	Nb SNV	Nb indels	Nb known SNV	Nb new SNV	%New SNVS
1	APN-01	23 927 282	99.6%	158	99.8%	99.5%	97.5%	88.7%	0.5%	82.5%	17.0%	70.4%	64.5%	602	139	578	24	4%
	APN-02	30 366 258	99.6%	227	99.9%	99.6%	98.5%	95.1%	0.5%	80.4%	19.1%	70.0%	65.0%	562	146	543	19	3%
	APN-03	26 100 034	99.6%	209	99.8%	99.5%	98.1%	93.5%	0.6%	78.3%	21.1%	70.5%	63.4%	617	146	605	12	2%
	APN-04	35 432 234	99.6%	270	99.9%	99.6%	99.0%	97.3%	0.6%	78.3%	21.1%	67.5%	60.2%	623	152	605	18	3%
	APN-07	28 382 460	99.7%	181	99.8%	99.4%	97.6%	90.6%	0.4%	83.2%	16.4%	71.9%	65.1%	619	147	608	11	2%
	APN-08	32 866 324	99.7%	265	99.9%	99.6%	99.0%	97.0%	0.6%	78.3%	21.1%	70.9%	63.5%	562	148	549	13	2%
	APN-09	28 392 354	99.5%	207	99.8%	99.5%	98.1%	93.8%	0.6%	79.3%	20.1%	67.1%	60.4%	596	152	584	12	2%
	APN-10	25 786 264	99.6%	194	99.9%	99.6%	98.4%	94.0%	0.5%	79.2%	20.3%	69.9%	61.5%	603	150	587	16	3%
	APN-11	19 842 144	99.7%	169	99.8%	99.5%	98.1%	91.7%	0.7%	75.8%	23.6%	68.5%	59.8%	588	141	572	16	3%
	APN-12	31 237 990	98.6%	198	99.8%	99.4%	98.1%	93.8%	0.9%	71.4%	27.7%	47.0%	34.7%	615	149	595	20	3%
	APN-13	18 916 942	99.2%	155	99.8%	99.4%	97.3%	88.1%	0.6%	78.0%	21.3%	68.4%	63.9%	583	150	572	11	2%
2 [a]	APN-15	55 437 506	97.9%	91	99.8%	99.1%	92.6%	60.5%	1.8%	57.2%	41.0%	43.3%	68.4%	607	142	600	7	1%
	APN-16	69 337 832	98.7%	145	97.5%	91.6%	84.4%	75.1%	0.7%	82.7%	16.5%	55.8%	20.5%	548	142	533	15	3%
	APN-19	13 620 090	97.5%	79	99.1%	93.3%	70.5%	49.7%	2.1%	48.0%	49.9%	41.7%	20.6%	543	125	535	8	1%
3	APN-24	52 250 914	97.2%	184	99.9%	99.8%	99.4%	97.0%	0.5%	83.3%	16.2%	60.8%	27.8%	620	185	604	16	3%
	APN-25	35 420 424	97.7%	232	99.9%	99.8%	99.4%	98.4%	0.6%	77.5%	21.9%	62.4%	37.6%	634	169	623	11	2%
	APN-28	46 303 338	97.2%	402	99.9%	99.8%	99.6%	99.1%	0.7%	72.6%	26.7%	61.1%	45.2%	584	163	571	13	2%
	APN-29	44 522 830	97.5%	333	99.9%	99.7%	99.5%	98.8%	0.6%	77.0%	22.5%	63.2%	45.8%	605	167	588	17	3%
	APN-30	37 719 174	97.5%	295	99.9%	99.7%	99.2%	98.0%	0.6%	74.7%	24.7%	60.6%	44.3%	627	170	614	13	2%
	APN-31	35 946 982	97.6%	279	99.9%	99.7%	99.3%	98.1%	0.6%	74.9%	24.5%	63.0%	44.3%	638	180	620	18	3%
	APN-32	48 462 058	97.4%	454	99.9%	99.8%	99.6%	99.2%	0.6%	72.6%	26.7%	63.3%	48.9%	574	179	559	15	3%
	APN-33	41 053 574	97.4%	371	99.9%	99.8%	99.6%	99.2%	0.6%	72.8%	26.5%	62.8%	46.1%	674	179	653	21	3%
	APN-34	36 702 446	97.4%	232	99.9%	99.7%	99.2%	97.8%	0.5%	80.2%	19.3%	63.5%	44.1%	596	168	581	15	3%
	APN-35	51 145 142	96.5%	230	99.9%	99.8%	99.5%	98.3%	0.7%	77.1%	22.2%	55.6%	26.7%	596	180	582	14	2%
	APN-36	48 399 084	97.1%	241	99.9%	99.8%	99.5%	98.6%	0.5%	82.0%	17.5%	62.9%	37.0%	584	180	575	9	2%
4	APN-37	44 564 444	97.1%	356	99.9%	99.8%	99.6%	99.2%	0.7%	75.0%	24.3%	60.5%	44.4%	619	170	608	11	2%
	APN-39	40 672 474	97.6%	480	99.9%	99.8%	99.6%	99.4%	0.8%	68.1%	31.1%	63.4%	51.8%	600	154	580	20	3%
	APN-40	44 111 500	97.2%	417	99.9%	99.8%	99.6%	99.2%	0.7%	72.8%	26.5%	63.7%	49.8%	565	167	554	11	2%
	APN-41	52 875 660	97.3%	425	99.9%	99.8%	99.6%	99.4%	0.6%	75.6%	23.8%	63.3%	44.3%	624	176	614	10	2%
	APN-42	41 040 590	97.4%	428	99.9%	99.8%	99.6%	99.2%	0.7%	69.8%	29.4%	62.2%	49.1%	573	173	560	13	2%
	APN-43	49 583 266	97.4%	145	99.9%	99.8%	99.3%	88.9%	0.4%	88.3%	11.4%	64.7%	32.1%	656	190	642	14	2%
	APN-46	47 255 380	97.2%	180	99.9%	99.7%	99.1%	95.4%	0.5%	83.4%	16.1%	60.2%	32.9%	596	182	584	12	2%
	APN-47	36 227 960	97.2%	178	99.9%	99.7%	99.2%	96.0%	0.5%	82.0%	17.5%	61.5%	37.7%	614	167	602	12	2%
	APN-48	43 920 858	96.9%	183	99.9%	99.8%	99.3%	96.5%	0.5%	82.9%	16.7%	59.9%	33.6%	630	169	617	13	2%
	APN-49	50 099 296	97.0%	223	99.9%	99.8%	99.3%	97.7%	0.5%	82.6%	17.0%	62.6%	36.5%	609	169	599	10	2%
	APN-50	42 289 892	97.0%	148	99.9%	99.7%	98.6%	90.8%	0.4%	85.7%	13.9%	61.6%	35.1%	629	169	616	13	2%
5	APN-45	31 253 880	97.4%	112	99.9%	99.8%	98.6%	67.2%	0.6%	79.6%	19.8%	61.7%	22.8%	633	180	624	9	1%
	APN-51	31 867 712	97.6%	225	99.9%	99.8%	99.4%	98.3%	0.8%	73.1%	26.1%	62.3%	34.8%	596	182	585	11	2%
	APN-53	24 884 362	97.6%	161	99.9%	99.7%	99.3%	94.2%	0.7%	75.1%	24.1%	61.8%	34.4%	588	181	573	15	3%
	APN-54	28 353 188	97.5%	97	99.9%	99.7%	97.5%	57.6%	0.6%	81.6%	17.8%	62.3%	23.9%	599	182	589	10	2%
	APN-56	30 468 964	97.3%	102	99.9%	99.7%	98.0%	61.5%	0.6%	81.0%	18.4%	59.3%	23.1%	567	179	560	7	1%
	APN-57	28 507 966	97.5%	74	99.9%	99.6%	90.8%	45.4%	0.5%	84.0%	15.5%	61.5%	22.0%	572	184	560	12	2%
	APN-58 [b]	33 477 982	97.6%	35	99.9%	98.6%	41.5%	0.9%	0.3%	90.8%	8.9%	68.9%	16.3%	621	212	574	47	8%

	APN-60	27 830 732	97.4%	80	99.9%	99.6%	93.6%	50.5%	0.5%	83.3%	16.2%	61.3%	23.6%	591	169	576	15	3%
	APN-63	25 668 896	97.4%	109	99.9%	99.7%	98.3%	66.1%	0.6%	81.7%	17.7%	62.5%	30.5%	587	177	569	18	3%
	APN-65	34 420 082	97.3%	115	99.9%	99.7%	98.6%	68.8%	0.6%	82.6%	16.8%	61.8%	24.8%	590	180	583	7	1%
6	APN-14[c]	23 144 314	99.2%	196	99.9%	99.6%	99.0%	95.0%	0.4%	84.4%	15.2%	77.7%	73.6%	608	158	599	9	1%
	APN-68	19 810 224	99.2%	151	99.9%	99.6%	98.7%	90.6%	0.4%	84.9%	14.7%	77.7%	68.7%	592	164	582	10	2%
	APN-70	3 444 990	99.2%	114	99.8%	99.4%	96.4%	75.9%	1.4%	38.6%	60.0%	75.2%	73.1%	618	150	605	13	2%
	APN-73	23 056 048	99.4%	238	99.9%	99.7%	99.4%	98.2%	0.6%	77.4%	21.9%	73.4%	62.6%	613	154	610	3	0%
	APN-74	22 070 772	99.2%	130	99.9%	99.6%	98.1%	84.5%	0.3%	88.4%	11.3%	75.0%	69.1%	616	154	606	10	1%
	APN-76	21 962 484	99.2%	258	99.9%	99.7%	99.4%	98.3%	0.5%	77.2%	22.3%	76.0%	69.9%	603	164	589	14	2%
	APN-77	18 575 362	96.5%	250	99.8%	99.6%	99.2%	98.0%	1.4%	47.0%	51.6%	38.3%	32.0%	645	164	629	16	2%
	APN-80	21 129 530	99.3%	184	99.9%	99.6%	98.9%	94.4%	0.4%	83.3%	16.3%	76.6%	71.1%	595	148	584	11	1%
	APN-81	18 074 502	99.2%	174	99.9%	99.6%	98.9%	93.2%	0.4%	82.0%	17.6%	77.0%	72.3%	547	161	542	5	1%
	APN-82	20 574 088	99.3%	193	99.9%	99.7%	99.0%	95.0%	0.4%	82.5%	17.1%	77.3%	72.5%	573	151	564	9	1%
	APN-83	25 008 458	99.2%	295	99.9%	99.8%	99.4%	98.9%	0.6%	75.6%	23.8%	75.5%	65.8%	585	163	575	10	1%
	APN-84	23 851 690	99.1%	376	99.9%	99.8%	99.5%	99.1%	0.9%	67.1%	32.1%	73.9%	65.6%	597	163	585	12	2%
	APN-86	16 885 790	99.1%	241	99.9%	99.7%	99.4%	98.3%	0.8%	68.5%	30.7%	75.5%	61.1%	586	172	580	6	1%
	APN-87	21 682 206	99.1%	264	99.9%	99.8%	99.4%	98.7%	0.7%	72.6%	26.7%	74.9%	60.1%	626	170	618	8	1%
7	APN-17[c]	18 692 086	99.1%	299	99.9%	99.7%	99.3%	98.5%	0.9%	66.9%	32.2%	75.6%	66.3%	582	172	574	8	1%
	APN-18[c]	25 957 694	98.6%	251	99.9%	99.6%	99.2%	97.4%	1.1%	68.8%	30.1%	65.3%	44.1%	577	156	570	7	1%
	APN-26	25 012 706	98.9%	208	99.8%	99.6%	98.8%	95.6%	0.7%	77.9%	21.3%	71.1%	52.5%	617	163	606	11	1%
	APN-38	21 900 976	98.2%	151	99.8%	99.5%	98.2%	90.6%	1.1%	68.1%	30.8%	58.7%	30.2%	612	161	598	14	2%
	APN-69	23 433 926	97.7%	184	99.9%	99.5%	98.7%	94.2%	1.4%	59.9%	38.7%	50.6%	27.8%	615	166	605	10	1%
	APN-72	27 072 904	97.5%	248	99.9%	99.7%	98.9%	96.8%	1.3%	62.3%	36.5%	53.6%	34.5%	607	158	600	7	1%
	APN-114	17 241 784	95.5%	188	99.8%	99.3%	97.0%	89.3%	2.7%	9.7%	87.6%	18.3%	17.4%	563	140	559	4	1%
	APN-115	19 560 280	97.1%	433	99.9%	99.6%	99.0%	97.7%	2.2%	24.7%	73.0%	45.7%	41.5%	617	147	606	11	1%
	APN-116	17 207 384	98.0%	590	99.8%	99.5%	99.1%	98.2%	1.7%	23.5%	74.8%	59.1%	56.3%	599	153	586	13	2%
	APN-117	13 374 526	99.1%	609	99.8%	99.6%	99.0%	98.1%	1.5%	20.5%	78.0%	72.2%	71.3%	626	159	618	8	1%
	APN-118	20 137 422	98.2%	686	99.8%	99.6%	99.2%	98.5%	1.7%	25.8%	72.6%	60.6%	57.4%	650	152	638	12	1%
	APN-119	20 227 292	98.8%	645	99.9%	99.7%	99.2%	98.5%	1.9%	30.7%	67.3%	66.2%	63.9%	633	156	624	9	1%
	APN-120	24 984 952	98.5%	664	99.9%	99.6%	99.3%	98.7%	1.7%	36.9%	61.3%	63.0%	58.6%	609	158	601	8	1%
	APN-121	18 167 438	98.9%	563	99.9%	99.6%	99.2%	98.4%	1.8%	35.3%	62.9%	69.3%	66.4%	620	154	610	10	1%
	APN-122	20 241 724	99.0%	737	99.8%	99.6%	99.3%	98.7%	1.5%	34.9%	63.7%	72.0%	68.9%	617	165	607	10	1%
	APN-123	19 947 194	97.7%	372	99.8%	99.5%	98.6%	96.6%	1.9%	39.2%	59.0%	50.7%	42.8%	603	151	591	12	2%
	APN-124	23 381 352	97.4%	504	99.9%	99.6%	99.2%	98.3%	2.0%	24.3%	73.7%	45.0%	40.2%	585	156	570	15	2%
	APN-125	22 035 142	99.1%	874	99.8%	99.6%	99.3%	98.8%	1.4%	29.6%	69.0%	71.6%	69.4%	600	155	594	6	1%
	APN-126	21 898 210	98.7%	791	99.8%	99.6%	99.3%	98.8%	1.4%	33.5%	65.1%	70.5%	67.7%	630	156	617	13	2%
	APN-127	26 850 456	98.8%	696	99.9%	99.7%	99.4%	98.8%	1.5%	43.8%	54.8%	68.4%	63.7%	617	167	605	12	2%
8	APN-99	19 156 392	98.3%	365	99.9%	99.6%	98.8%	97.3%	1.3%	57.7%	41.0%	64.5%	62.7%	626	183	615	11	1%
	APN-100	14 919 436	98.7%	80	99.7%	98.5%	84.4%	50.0%	0.3%	89.8%	9.9%	74.1%	73.0%	596	175	581	15	2%
	APN-101	20 575 066	98.5%	417	99.9%	99.6%	98.9%	97.6%	1.1%	58.2%	40.7%	68.4%	67.1%	625	168	613	12	2%
	APN-102	24 463 598	98.7%	294	99.9%	99.5%	98.5%	95.9%	0.7%	76.3%	23.1%	71.8%	70.0%	632	173	619	13	2%
	APN-103	23 568 632	98.8%	325	99.9%	99.5%	98.4%	95.6%	0.7%	73.3%	25.9%	73.9%	72.1%	610	178	605	5	1%
	APN-104	19 208 764	97.3%	404	99.8%	99.6%	98.7%	97.1%	1.7%	39.6%	58.7%	51.1%	49.0%	629	171	620	9	1%
	APN-105	23 921 986	97.6%	402	99.9%	99.5%	98.7%	97.0%	1.3%	54.8%	43.9%	53.5%	52.6%	610	168	599	11	1%
	APN-106	19 424 216	97.1%	556	99.8%	99.6%	99.1%	98.2%	2.6%	14.9%	82.4%	49.9%	47.4%	599	176	592	7	1%
	APN-107	27 343 616	96.7%	612	99.9%	99.7%	99.2%	98.3%	2.4%	25.8%	71.8%	45.8%	42.5%	607	155	592	15	2%
	APN-108	20 657 638	96.6%	499	99.9%	99.6%	98.9%	97.6%	2.6%	6.0%	91.4%	37.0%	36.8%	603	158	596	7	1%
	APN-109	30 928 180	95.9%	554	99.8%	99.6%	99.1%	98.1%	2.7%	14.2%	83.1%	31.7%	29.8%	587	172	577	10	1%
	APN-110	23 887 290	98.6%	839	99.9%	99.7%	99.4%	98.8%	1.9%	26.2%	72.0%	67.6%	65.7%	637	168	623	14	2%
	APN-111	24 851 496	98.5%	540	99.9%	99.7%	99.3%	98.6%	1.5%	53.2%	45.3%	69.7%	64.7%	648	173	640	8	1%
	APN-112	17 718 676	98.2%	492	99.8%	99.6%	99.0%	97.9%	1.7%	35.4%	62.9%	63.1%	60.1%	617	171	604	13	2%

	APN-113	27 627 002	98.5%	775	99.9%	99.7%	99.3%	98.7%	1.7%	38.4%	59.9%	67.4%	63.2%	609	185	595	14	2%
9	APN-128	19 785 578	99.0%	607	99.9%	99.6%	99.2%	98.3%	1.8%	34.2%	64.1%	68.3%	64.8%	590	166	580	10	1%
	APN-130	19 853 062	99.2%	726	99.9%	99.6%	99.2%	98.5%	1.3%	35.0%	63.7%	71.9%	68.5%	667	159	651	16	2%
	APN-131	23 072 640	99.1%	609	99.9%	99.7%	99.3%	98.6%	1.5%	42.5%	56.0%	67.9%	63.4%	580	162	574	6	1%
	APN-132	29 360 264	98.2%	238	99.8%	99.5%	98.6%	96.0%	0.8%	77.0%	22.2%	74.0%	49.2%	614	167	607	7	1%
	APN-134	25 278 984	99.1%	751	99.9%	99.7%	99.4%	98.8%	1.6%	35.5%	62.9%	67.6%	63.6%	599	153	592	7	1%
	APN-135	25 325 698	99.0%	660	99.9%	99.7%	99.3%	98.6%	1.6%	43.7%	54.7%	68.7%	63.9%	652	156	640	12	1%
	APN-137	24 441 638	98.9%	826	99.9%	99.7%	99.3%	98.7%	1.9%	25.6%	72.5%	65.9%	62.8%	624	162	615	9	1%
	APN-138	23 128 034	99.1%	456	99.8%	99.6%	99.0%	98.0%	1.2%	54.7%	44.1%	69.1%	60.5%	622	160	609	13	2%
	APN-139	26 354 740	99.1%	632	99.9%	99.6%	99.3%	98.6%	1.3%	48.6%	50.1%	70.4%	64.3%	573	162	558	15	2%
	APN-141	26 997 926	99.1%	633	99.9%	99.6%	99.3%	98.7%	1.3%	50.0%	48.7%	71.0%	64.3%	710	161	702	8	1%
	APN-142	21 163 728	99.1%	941	99.9%	99.6%	99.2%	98.7%	1.5%	21.1%	77.4%	71.9%	69.7%	624	164	612	12	2%
	Mean	**28 458 084**	**98.2%**	**353**	**99.8%**	**99.5%**	**97.7%**	**91.7%**	**1.0%**	**61.8%**	**37.1%**	**63.7%**	**51.2%**	**607**	**164**	**595**	**12**	**2%**
	SD	**11 060 265**	**1.0%**	**218**	**0.2%**	**1.0%**	**6.6%**	**15.2%**	**0.6%**	**22.8%**	**22.2%**	**10.5%**	**16.4%**	**27**	**13**	**27**	**5**	**1%**

a : Capture reaction step was performed on a pool of two equimolar DNA samples (such as previously described in Redin et al.), what may explain the drop of coverage and of capture efficiency observed in those samples

b: APN-58 did not pass our quality criteria for sequencing data but it is however indicated in the table since a causative mutation was detected in *DYRK1A* in spite of general lower-coverage data.

c: Re-sequenced samples because of poor coverage results obtained initially with batch 2 sequencing

Table S2: List of benign or pathogenic CNVs detected in patients using a depth of coverage comparison method

Gene	Patient ID	Sex	Inheritance	Segregation	Gene MIM#	CNV type	Confirmation	Conclusion
X-Linked genes								
MECP2	APN-3	M	XL	inherited (M)	300005	inversion, hemz	Sanger	Pathogenic
FMR1	APN-26	M	XL	inherited (M)	309550	deletion, hemz	Sanger, CGH (1 deleted probe)	Pathogenic
MAGT1	APN-141	M	XL	?	300715	duplication, htz	-	Probably benign
Autosomal recessive genes								
MCPH1	APN-42	M	AR	inherited (M)	607117	deletion, htz	CGH (in report)	Benign
Others								
CYFIP1	APN-14	M	?	?	606322	duplication, htz	CGH (in report)	Benign

XL: X-linked, AR: Autosomal Recessive, ?: unclear implication in ID

Table S3: List of regions consistently poorly-covered in our assay

Gene	Exon/Intron #	Chrom	Start	End	Length of the region coding|uncoding|total (pb)			GC content	Mean depth Batch 1	Mean depth Batch 2	Mean depth Batch 3	Mean depth Batch 4	Mean depth Batch 5	Mean depth Batch 6	Mean depth Batch 7	Mean depth Batch 8	Mean depth Batch 9	Max Depth All	Mean Depth All	Mean depth [a] (Percentile: 90%)
DEAF1	E1 (5'UTR)	11	695 133	695 176	0	44	44	79%	8	3	**35**	**40**	**24**	**21**	16	13	13	58	20	39
NRXN2	E2 (5'UTR + coding)	11	64 481 117	64 481 332	55	161	216	81%	4	0	18	**23**	11	7	5	4	5	37	9	14
RAI1	E6 (coding + 3' UTR)	17	17 713 292	17 713 385	4	90	94	77%	12	4	**30**	**35**	**24**	**26**	17	15	18	66	21	29
GAMT	E1 (coding + 5'UTR)	19	1 401 471	1 401 599	5	124	129	80%	5	1	**21**	**26**	12	15	8	8	8	48	12	18
SHANK1	E22	19	51 172 026	51 172 224	199	0	199	79%	11	2	**27**	**29**	17	**22**	9	8	10	44	15	31
AFF3	I2	2	100 721 046	100 721 084	0	39	39	87%	14	1	**22**	**26**	17	**26**	**20**	17	**22**	52	20	27
PRODH	E2 (5'UTR + coding) + I2	22	18 923 494	18 923 831	273	65	338	79%	4	0	8	10	6	9	7	6	7	19	7	10
SHANK3	E1 + I1	22	51 113 030	51 113 174	63	82	145	85%	0	0	2	3	3	2	1	1	1	6	1	2
SHANK3	I10	22	51 135 785	51 135 802	0	18	18	71%	14	4	**20**	**25**	14	**30**	**32**	**29**	**26**	78	24	40
SHANK3	I10 + E11 + I11	22	51 135 837	51 136 183	152	195	347	79%	3	2	4	6	3	6	3	3	4	10	4	5
SYNGAP1	E1 (5' UTR + coding) + I1	6	33 387 996	33 388 117	67	55	122	66%	2	0	1	1	2	2	1	1	0	5	1	2
LAMC3	E1 (5' UTR + coding)	9	133 884 469	133 884 618	17	133	150	83%	3	0	**28**	**34**	13	6	4	5	4	62	11	17
RALGDS	I1 + E1	9	135 996 336	135 996 385	27	23	50	78%	10	2	**25**	**25**	16	**25**	19	17	**20**	46	19	26
RALGDS	E1 (coding + 5'UTR)	9	135 996 436	135 996 553	106	12	118	77%	7	1	**20**	**21**	13	18	11	10	15	36	14	19
NHS	E1 (5'UTR + coding)	X	17 393 847	17 394 146	266	34	300	76%	5	1	**22**	**24**	12	9	8	6	8	40	11	15
RPS6KA3	E1 (upstream + coding)	X	20 284 705	20 284 772	46	22	68	75%	13	0	20	**24**	11	**24**	**20**	14	16	62	18	29
ARX	I2+E2	X	25 031 435	25 031 939	481	24	505	81%	7	1	15	15	10	17	11	8	12	34	12	16
ATP6AP2	E1 (5' UTR + coding) + I1	X	40 440 226	40 440 443	37	181	218	77%	4	1	11	12	7	9	8	6	6	23	7	12
IQSEC2	E1 (5' UTR)	X	53 350 457	53 350 483	0	27	27	81%	5	0	**37**	**39**	15	8	9	6	9	74	14	23
FGD1	E1 (5' UTR)	X	54 521 880	54 521 891	0	12	12	91%	10	3	**39**	**37**	15	**24**	**21**	12	20	69	21	38
SRPX2	I10	X	99 924 410	99 924 449	0	40	40	64%	16	1	**29**	**40**	17	17	17	12	18	52	19	25
LAMP2	I1 + E1 (coding + 5'UTR)	X	119 602 936	119 603 057	64	58	122	69%	9	4	**24**	**27**	13	13	16	11	19	46	16	22
OCRL	E1 (5' UTR + coding) + I1	X	128 674 326	128 674 543	39	179	218	76%	3	1	7	8	4	10	5	3	5	16	6	8
SOX3	E1 (3'UTR + coding)	X	139 585 853	139 585 902	18	32	50	67%	10	1	**23**	**25**	14	**24**	**20**	13	17	43	18	25
MECP2	I1 + E1	X	153 362 925	153 363 118	58	136	194	80%	1	0	2	2	1	2	2	2	3	6	2	3
				Total	**1 977**	**1 786**	**3 763**	77%												

a: Regions where 90% of patients harbor a coverage <40X

Table S4: List of the variants annotated as pathogenic mutations according to dbSNP but unlikely associated to ID

Gene	Transcript#	Variation nomenclature	Clinical Significance in dbSNP	EVS carriers	EVS MAF	Predictions	phyloP	Grantham Dist	All Count (/106)	Responsible for ID	Reason for questioning
X-Linked genes											
FGD1 (XL)	NM_004463.2	chrX:g.54496615G>A c.935C>T p.Pro312Leu (rs28935498)	pathogenic	7 (2M hemz, 5 F htz)	0.07%	Deleterious (Sift), Neutral (Polyphen2)	1.34	98	1 hmz	No	Already discussed in Piton et al., AJHG, 2013
OTC (XL)	NM_000531.5	chrX:g.38268220A>G c.809A>G p.Gln270Arg (rs1800328)	pathogenic	**351 (93 M hemz, 6 F hmz, 246 F htz)**	3.32%	Deleterious (Sift, Polyphen2)	4.48	43	6 (4 hmz, 2 htz)	No	Frequency too high in males in control cohort
Autosomal recessive genes											
ACY1 (AR)	NM_000666.2	chr3:g.52022837C>T c.1057C>T p.Arg353Cys (rs121912698)	pathogenic	33 (33 htz)	0.25%	Deleterious (Sift, Polyphen2)	2.47	180	1 htz	Unlikely	Frequency high in control cohort
ACY1 (AR)	NM_000666.2	chr3:g.52023042G>A c.1178G>A p.Arg393His (rs121912701)	pathogenic	**59 (1 hmz, 58 htz)**	0.46%	Tolerated (Sift), Deleterious (Polyphen2)	0.13	29	1 htz	Unlikely	Frequency too high in control cohort
GAMT (AR)	NM_000156.4	chr19:g.1399056C>T c.460-31G>A p.? (rs55776826)	pathogenic	**1989** **(139 hmz, 1711 htz)**	15.29%	NA	-3.83	NA	23 (2 hmz, 21 htz)	No	Frequency too high in control cohort
PRODH (AR*)	NM_016335.4	chr22:g.18905964C>T c.1292G>A p.Arg431His (rs2904552)	pathogenic	**1061** **(48 hmz, 965 htz)**	8.16%	Deleterious (Sift, Polyphen2)	1.42	29	17 (1 hmz, 16 htz)	No	Frequency too high in control cohort
PRODH (AR*)	NM_016335.4	chr22:g.18905899G>A c.1357C>T p.Arg453Cys (rs3970559)	pathogenic	**119** **(2 hmz, 115 htz)**	0.91%	Deleterious (Sift, Polyphen2)	1.01	180	4 htz	No	Frequency too high in control cohort
PRODH (AR*)	NM_016335.4	chr22:g.18909902A>T c.865T>A p.Leu289Met (rs137852934)	pathogenic	**60 (60 htz)**	0.46%	Tolerated (Sift), Neutral (Polyphen2)	0.37	15	1 htz	Unlikely	Frequency too high in control cohort
PRODH (AR*)	NM_016335.4	chr22:g.18905934A>G c.1322T>C p.Leu441Pro (rs2904551)	pathogenic	**76 (76 htz)**	0.58%	Deleterious (Sift, Polyphen2)	3.92	98	1 htz	Unlikely	Frequency too high in control cohort
PMM2 (AR)	NM_000303.2	chr16:g.8905010G>A c.422G>A p.Arg141His (rs28936415)	pathogenic	**50 (50 htz)**	0.39%	Deleterious (Sift, Polyphen2)	5.69	29	2 htz	Unlikely	Frequency too high in control cohort

Some variants are annotated as 'pathogenic' in databases such as dbSNP but their frequency in our cohort or in a large general population is incoherent with penetrant pathogenicity

EVS: Exome Variant Server; MAF: minor allele frequency; htz: heterozygous; hemz: hemizygous; hmz: homozygous

Table S5: List of initial candidate variants with excluded/questioned pathogenicity

Gene	Patient ID	Transcript #	Variation nomenclature	Predictions	phyloP	Grantham Dist	Count in cohort (/106)	Responsible for ID	Reason for questioning
Autosomal dominant genes									
DEAF1	APN-109	NM_021008.2	chr11:g.691601G>C c.290-3C>G p.?	100% splice site disruption (MES, NNS). Alters splicing in vitro	2,55	-	1 htz	Unlikely	Inherited from unaffected mother
DOCK8	APN-105	NM_203447.3	chr9:g.197171G>T c.3496G>T p.Glu1166*	-	NA	-	1 htz	Unlikely	Proband also carries a causative splice site mutation in *PHF8*, and the implication of *DOCK8* in ID most probably substantial
ZNF599	APN-31, APN-42, APN-100	NM_001007248.2	chr19:g.35250691_35250692del c.1014_1015del p.Tyr339* (rs148227520)	-	-	-	3 htz	No	Reported in 3 patients of the cohort, among which one (APN-42) also carries a frameshift mutation in *DMD*
X-linked genes									
SHROOM4	APN-86	NM_020717.3	chrX:g.50345803G>A c.3772C>T p.Gln1258*	-	4,08	NA	1 hemz	No	Present in 2 unaffected brothers. Implication of this gene already questionned in 12
SRPX2	APN-13	NM_014467.2	chrX:g.99920309del c.602del p.Ala201Valfs*10	-	-	NA	1 hemz	Unlikely	Present in 3 maternal aunts. Absent from maternal grandmother, probably inherited from unaffected (deceased) grandfather. Implication of this gene already questionned in (12)
FLNA	APN-17	NM_001110556.1	chrX:g.153588207G>A c.3872C>T p.Pro1291Leu (rs137853319)	Deleterious (Sift), Neutral (Polyphen2)	1,74	98	1 hemz	Unlikely	Described as pathogenic but present in a control male (EVS). Patient has inconsistent phenotype compared to the initial patient with this mutation (46)
HUWE1	APN-10	NM_031407.4	chrX:g.53596663G>C c.6437C>G p.Thr2146Arg	Deleterious (Sift, Polyphen2)	3,6	71	1 hemz	Unlikely	Present in an unaffected brother
MECP2	APN-74	NM_001110792.1	chrX:g.153363068C>T c.55G>A p.Glu19Lys	Deleterious (Sift), Neutral (Polyphen2)	2.22	56	1 hemz	Unlikely	Present in an unaffected brother
FMR1	APN-132	NM_002024.5	chrX:g.147024687G>A c.1312G>A p.Asp438Asn	Deleterious (Sift, Polyphen2)	5,05	23	1 hemz	Unlikely	Present in the unaffected maternal half-brother

htz: heterozygous, hemz: hemizygous

MES: MaxEntScan, NNS: NNSplice.

Table S6: Comparison of advantages and drawbacks of trio-genome, trio-exome and targeted sequencing strategies for the diagnosis of intellectual disability

Compared features	full-genome	exome	targeted
Trio analysis	required	needed (or preferable)[a]	not needed
Cost	very expensive (for required depth)	expensive[a]	4-5x cheaper[a]
Variant analysis	highly complex, very time-consuming (several millions of variants to analyze, of which >22,000 coding)	complex, time-consuming (>20,000 variants to analyze)	more straightforward (several hundred variants to analyze)
# of variant detected	$4.4x10^6$ SNVs	~ 24,324[2]; ~21,100[3]	~700
Depth of coverage (mean)	80X [1]	64X [2]; 112X [3] ; 130X [4]	373X
% covered regions	92% >40X [1]	87%>10X [2]; 90%>20X [3] ; 95%>20X [4]	97.6%>40X
Sensitivity	very high	high	very high
	NA [1]	97.4%[2]; 97.5%[3]	100%
Diagnostic efficiency[b]	42% [1] (CI: 28-56%)	16%[2](updated to 27% in[1]) ; 31%[3] ; 33% [4]; 24.5% (updated to 30%[1]) in pooled data on 211patients (+/-6% CI)	25% (CI: 17-33%)
Gene discovery	yes	yes	no
Data re-analysis with new knowledge	yes	yes	yes/no
Incidental findings	yes	yes	no
Best suited application	**research**	**research/diagnosis**	**diagnosis**

NA: non available, SNVs: single nucleotide variants

[a] three exomes for the exome strategy vs only proband for the targeted sequencing strategy, less samples pooled per sequencing lane in cases of exome vs targeted sequencing. Prices highly vary upon the mean sequencing depth to be reached. While Yang et al. performed proband only exome seq, they had to check further by Sanger 5-8 variants/sample, and reported a 14% false positive rate of NGS calls, [b] confidence interval (CI)

[1] Gilissen et al, Nature, 2014; [2] de Ligt et al., NEJM, 2012; [3] Rauch et al., Lancet, 2012 ; [4] Yang et al., NEJM, 2013

Table S7: Genes in which certainly-causative mutations were reported in the previous trio-exome studies

Rauch et al., Lancet, 2012		De Ligt et al., NEJM, 2012	
Gene	**Mutation**	**Gene**	**Mutation**
Autosomal dominant genes			
SATB2	Missense, de novo	***CTNNB1***	Frameshift, de novo
SCN2A	Frameshift, de novo	***DYNC1H1***	Missense, de novo
SCN2A	Frameshift, de novo	***GATAD2B***	Nonsense, de novo
SNC2A	Missense, de novo	*GRIN2A*	Missense, de novo
SCN8A	Missense, de novo	*GRIN2A*	Missense, de novo
SETBP1	Nonsense, de novo	*GRIN2B*	Missense, de novo
SLC2A1	Missense, de novo	*SCN2A*	Nonsense, de novo
STXBP1	Missense, de novo	*SYNGAP1*	Splice mutation, de novo
STXBP1	Splice mutation, de novo	*TCF4*	Missense, de novo
STXBP1	Missense, de novo	***TUBA1A***	Frameshift, de novo
SYNGAP1	Frameshift, de novo		
SYNGAP1	Frameshift, de novo		
TCF4	Missense, de novo		
Autosomal recessive genes			
None	-	***LRP2***	frameshift de novo + missense inherited
X-Linked genes			
IQSEC2	Nonsense, de novo	*ARHGEF9*	Missense, inherited
MECP2	Frameshift, de novo	*ARHGEF9*	Splice mutation, inherited
NAA10	Missense, de novo	*PDHA1*	Frameshift, de novo
		PDHA1	Missense, inherited
		SLC6A8	Frameshift, de novo
Initially candidate genes for dominant ID that have now been reeplicated			
CHD2	Frameshift, de novo	***EEF1A2***	Missense, de novo
DEAF1	Missense, de novo	***KIF5C***	Missense, de novo
KCNQ3	Missense, de novo		
SETD5	Nonsense, de novo		

bold: genes not included in our panel of 217 genes

Data from the study from Yang et al., NEJM, 2013 could not be included since although it is indicated that 60 patients had been recruited for neurological disorders (developmental delay,speech delay, ASD or ID), genes in which certainly-causative mutations have been detected for such category of patients are not clearly specified.

Table S8: List of the 217 targeted genes with associated mode of inheritance, presence of truncating variants in Exome Variant Server, nature of mutations reported in patients, and side-symptoms.

Official gene symbol	Gene MIM #	Associated syndrome (Phenotype MIM #)	Associated traits	Truncating mutations in EVS	Nature of mutations reported in OMIM (Truncating, Missenses, other)	Total size of targeted regions (bp)
X-Linked genes						
ACSL4	300157	Mental retardation, X-linked 63 (MIM 300387)	NS-ID	-	1T, 2M	2 785
AFF2	300806	Mental retardation, X-linked, FRAXE type (MIM 309548)	ADHD, autistic traits or NS-ID	-	1 trinucleotide expansion, 1 large exon deletion	4 867
AP1S2	300629	Mental retardation, X-linked syndromic, Fried type (MIM 300630)	Hypotonia, walking delay, aggressivity, DF or NS-ID	-	5T	634
ARHGEF6	300267	Mental retardation, X-linked 46 (MIM 300436)	NS-ID	-	1T, 1 translocation	3 211
ARHGEF9	300429	Epileptic encephalopathy, early infantile, 8 (MIM 300607)	Hyperekplexia or NS-ID	-	1T, 1M, 1 translocation, 1 large deletion encompassing 2 other genes	2 021
ARX	300382	Epileptic encephalopathy, early infantile, 1 (MIM 308350); Hydranencephaly with abnormal genitalia/Lissencephaly, X-linked 2 (MIM 300215); Mental retardation, X-linked 29 and others (MIM 300419); Partington syndrome (MIM 309510), Proud syndrome (MIM 300004)	Epileptic encephalopathy, lissencephaly, hydranencephaly with abnormal genitalia, or NS-ID	-	10T, 7M, 1 polyAla expansion, 2 large exon deletions	1 889
ATP6AP2	300556	-	Epilepsy	1 splice hemz	1 synonymous variant, slightly affecting splicing	1 413
ATP7A	300011	Menkes disease (MIM 309400); Occipital horn syndrome (MIM 304150)	Growth retardation, cerebellar degeneration, seizures; hyperelastic skin, hyperextensible joints, skeletal anomalies	-	9T, 5M, 2 large exon deletions	5 383
ATRX	300032	Alpha-thalassemia syndrome (MIM 300448, 301040)	DF, genital abnormalities, microcephaly, hypotonia, anemia, or NS-ID	1 stop htz	8T, 18M	8 879
BCOR	300485	OFCD syndrome (MIM 300166)	Microphtalmia, cataract, cardiac anomalies, DF, dental anomalies	1 splice htz	6T, 1M, 2 large exon deletions	5 828
BRWD3	300553	Mental retardation, X-linked 93 (MIM 300659)	Macrocephaly, DF	-	2T, 1M	7 295
CACNA1F	300110	Congenital stationary night blindness (MIM 300071)	Nystagmus, autistic traits	-	5T, 2M, 1 large exon deletion	8 387

CASK	300172	FG syndrome 4 (MIM 300422) MICPCH syndrome (MIM 300749)	Hypotonia, constipation, behavioral disturbances, nystagmus, microcephaly, pontine & cerebellar hypoplasia, speech & walk delay, DF	-	4T, 6M, 1 large exon deletion, 1 splice site decreased	3 927
CCDC22	300859	-	Hypoplastic phalanges, DF, genital anomalies	-	1M	2 957
CDKL5	300203	Angelman syndrome-like (MIM 105830); Early Infantile Epileptic Encephalopathy-2 (MIM 300672)	Ataxia, hypotonia, epilepsy, abnormal behaviors, speech delay, autistic traits, DF	-	7T, 7M	3 893
CLCN4	302910	-	Infantile epileptic encephalopathy, microcephaly, regression, hypotonia	-	1M	2 804
CNKSR2	300724	-	Epilepsy, microcephaly	-	1 large deletion	4 007
CUL4B	300304	Mental retardation, X-linked, syndromic 15 (Cabezas type, MIM 300354)	Short stature, hypogonadism, abnormal gait, speech delay, tremor	-	2T, 1M	3 595
DCX	300121	Lissencephaly, X-linked (MIM 300067)	Seizures, growth retardation, hypogenitalism	-	4T, 10M	1 624
DKC1	300126	Dyskeratosis congenita (MIM 305000); Hoyeraal-Hreidarsson syndrome (MIM 300240)	Abnormal skin pigmentation, hypogenitalism, anemia, microcephaly, growth retardation, spastic paresis, ataxia, anemia, immunodeficiency, cerebellar hypoplasia	-	1T, 10M, 1 in frame deletion, 1 large exon deletion, 1 promoter mutation, 1 intronic variant	2 145
DLG3	300189	Mental retardation, X-linked 90 (MIM 300850)	NS-ID	1 stop hemz in an alternative exon	4T	3 610
DMD (DP71)	300377	Becker muscular dystrophy (MIM 300376); Duchenne muscular dystrophy (MIM 310200)	Cardiac anomalies	1 stop htz, 3 splices in 9 htz and 1 hemz)	69T, 10M, 6 large exon deletions, 1 alu insertion	12 142
EIF2S3	300161	-	Microcephaly, short stature, DF	-	1M (Borck et al., Mol. Cell., 2012)	1 932
FGD1	300546	Aarskog-Scott syndrome (MIM 305400)	ADHD, short stature, shawl scrotum, brachydactyly	-	5T, 6M, 1 large exon deletion	3 757
FLNA	300017	FG syndrome 2 (MIM 300321); Periventricular Heterotopia (MIM 300049); Melnick-Needles syndrome (MIM 309350)	Hypotonia, speech delay, macrocephaly, constipation, epilepsy, cleft palate, skeletal anomalies, deafness, urogenital defects, DF	-	15T, 14M	11 265
FMR1	309550	Fragile-X syndrome (MIM 300624)	Macroorchidism, speech delay, DF	-	3T, 1M, 1 trinucleotide expansion	2 674
FRMPD4	300838	-		-	1 partial duplication (Honda et al., J. Hum. Genet., 2010)	4 632
FTSJ1	300499	Mental retardation, X-linked 9 (MIM 309549)	Agressive behaviour or NS-ID	-	3T, 1 large exon deletion	1 689

GDI1	300104	Mental retardation, X-linked 41 (MIM 300849)	NS-ID	-	2T, 2M	2 007
GK	300474	Glycerol kinase deficiency (MIM 307030)	Gastrointestinal symptoms	-	2T, 3M, 2 large exon deletions, 1 alu insertion	2 590
GPC3	300037	Simpson-Golabi-Behmel syndrome 1 (MIM 312870)	Overgrowth, congenital cardiac anomalies, cleft palate, DF	-	5T, 4M, 2 large exon deletions	2 172
GRIA3	305915	Mental retardation, X-linked 94 (MIM 300699)	Macrocephaly, seizures, autistic traits or NS-ID	-	3M, 1 entire gene deleted, 1 translocation, 1 submicroscopic duplication, 2 partial duplication	3 440
HCCS	300056	Microphthalmia, syndromic 7 (MIM 309801)	Hyperpigmented skin	-	1T, 1M, 1 large exon deletion	1 047
HCFC1	300019	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type, MIM 309541)	Epilepsy, choreoathetosis, microcephaly, growth retardation or NS-ID	-	4M, 1 5' UTR variant	7 411
HPRT1	308000	Lesch-Nynan syndrome (MIM 300322)	Spastic cerebral palsy, choreoathetosis, uric acid urinary stones, self-destructive behaviour, megaloblastic anemia	-	16T, 16M, 1 in frame deletion, 1 in frame insertion, 6 large exon deletions, 1 large inversion, 1 large exon duplication, 1 entire gene deletion	1 017
HSD17B10	300256	17-beta-hydroxysteroid dehydrogenase X deficiency (MIM 300438)	Choreoathetosis, developmental regression, epilepsy, visual anomalies	-	5M	1 157
HUWE1	300697	Mental retardation, X-linked syndromic, Turner type (MIM 300706)	Macrocephaly, macroorchidism or NS-ID	-	3M, 6 microduplications	16 786
IDS	300823	Mucopolysaccharidosis II (MIM 309900)	Airway obstruction, skeletal anomalies, cardiomyopathy	-	7T, 9M, 1 entire gene deletion	2 079
IGBP1	300139	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia (MIM 300472)	-	-	1 5' UTR variant	1 260
IL1RAPL1	300206	Mental retardation, X-linked 21/34 (MIM 300143)	Autistic traits, synophrys, hyperextensible joints, or NS-ID	-	2T, 2 large exon deletions	2 491
IQSEC2	300522	Mental retardation, X-linked 1 (MIM 309530)	Epilepsy, speech delay, brachycephaly, strabismus, or NS-ID	-	4M	5 269
KDM5C	314690	Claes-Jensen type XLID (MIM 300534)	Spastic paraplegia, short stature, microcephaly, epilepsy, facial hypotonia, maxillary hypoplasia	-	3T, 6M	3 915
KIAA2022	300524	Mental retardation, X-linked 98 (MIM 300912)	NS-ID	-	1 pericentric inversion	4 671
KLF8	300286	-	Hypotonia	-	1 translocation (Lossi et al., J; Med. Genet., 2002)	1 320

L1CAM	308840	CRASH syndrome (MIM 303350); MASA syndrome (MIM 303350); Hydrocephalus with Hirschsprung disease (MIM 307000)	Spastic paraplegia, aphasia, shuffling gait, speech delay	-	6T, 10M, 1 large exon duplication, 1 synonymous variant leading to an in-fame deletion	5 613
LAMP2	309060	Danon disease (MIM 300257)	Cardiac anomalies, muscle weakness	-	10T, 2M	1 956
MAGT1	300715	Mental retardation, X-linked 95 (MIM 300716)	NS-ID	1 stop (1 hemz)	1M	1 504
MAOA	309850	Brunner syndrome (MIM 300615)	Impulsive aggressiveness or NS-ID	-	1T	2 184
MBTPS2	300294	IFAP/BRESHECK syndrome (MIM 308205)	Ichthyosis follicularis, atrichia, photophobia, hirschsprung disease, kidney dysplasia, cryptorchidism, cleft palate, skeletal anomalies	-	5M	2 000
MECP2	300005	Angelman syndrome (MIM 105830); Severe neonatal encephalopathy (MIM 300673); Rett syndrome (MIM 312750)	Gait anomalies, abnormal behavior, epilepsy, speech limitation, microcephaly, hypotonia, respiratory anomalies, constipation, growth retardation, developmental regression, autistic traits, DF	-	22T, 14M, 1 large in frame deletion, 1 entire gene duplication	1 683
MED12	300188	Lujan-Fryns syndrome (MIM 309520); Opitz-Kaveggia syndrome (MIM 305450); Ohdo syndrome (MIM 300895)	Marfanoid habitus, underweight, cryptorchidism, joint laxity, clinodactyly, skin pigmentations, deafness, feeding disorder, macrocephaly, hypotonia, constipation, agenesis of the CC, DF	-	5M	8 805
MID1	300552	Opitz GBBB syndrome (MIM 300000)	Hypospadias, cleft palate, laryngotracheoesophageal abnormalities, imperforate anus, cardiac defects	-	4T, 2M, 1 exon duplication, 1 in-frame deletion, 1 in-frame duplication	2 827
MIR222	300569	-	-	-	-	150
NDP	300658	Norrie disease (MIM 310600)	Early blindness, deafness, seizures, growth retardation		3T, 13M	482
NDUFA1	300078	Mitochondrial complex I deficiency (MIM 252010)	Macrocephaly, leukodystrophy, encephalopathy, cardiomyopathy, myopathy, parkinson disease, liver disease, optic neuropathy	-	3M	333
NHS	300457	Nance-Horan syndrome (MIM 302350)	Congenital cataract, dental anomalies, DF	-	6T	5 390
NLGN3	300336	Asperger syndrome susceptibility, X-linked 1 (MIM 300494); Autism susceptibility, X-linked 1 (MIM 300425)	ASD, Asperger	-	1M	2 827

NLGN4X	300427	Mental retardation, X-linked (MIM 300495); Asperger syndrome susceptibility, X-linked 2 (MIM 300497); Autism susceptibility, X-linked 2 (MIM 300495)	ASD, Asperger or NS-ID	-	2T, 1 large exon deletion	2 651
NSDHL	300275	CHILD syndrome (MIM 308050); CK syndrome (MIM 300831)	Hemidysplasia, ichythyosiform erythrodema, limb defects, seizures, microcephaly, cortical malformations, DF	-	4T, 3M, 1 in-frame deletion	1 402
NXF5	300319	-	Short stature, pectus excavatum, epilepsy, hypotonia, autistic traits, DF	2 stops in 8 hemz individuals	1 large inversion, 1 large deletion, 2 large duplications (reviewed in Piton et al., AJHG, 2013)	2 017
OCRL	300535	Lowe syndrome (MIM 309000); Dent disease (MIM 300555)	Hydrophthalmia, cataract, aminoaciduria, hypotonia, renal defects	-	3T, 6M	3 770
OFD1	311200	Joubert syndrome 1 (MIM 300804); Oral-facial-digital syndrome 1 (MIM 311200); Simpson-Golabi-Behmel syndrome, type 2 (MIM 300209)	Hypotonia, cerebellar ataxia, breathing defects, retinal dystrophy, polydactyly, dental anomalies, renal defects, macrocephaly, gastrointestinal and genitourinary anomalies, DF	-	8T, 1M, 1 large exon deletion	4 006
OPHN1	300127	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance (MIM 300486)	Hypotonia, strabismus, seizures, DF	-	4T, 2 large exon deletions, 1 translocation	3 329
OTC	300461	Ornithine Transcarbamylase Deficiency (MIM 311250)	Encephalopathy, hyperammonemia, respiratory alkalosis	-	6T, 20M, 2 intronic variants, 1 entire gene deletion	1 505
PAK3	300142	Mental retardation, X-linked 30/47 (MIM 300558)	Microcephaly, hypotonia, drooling, psychiatric features	-	2T, 3M	2 383
PCDH19	300460	Epileptic encephalopathy, early infantile, 9 (MIM 300088)	Psychiatric features	-	6T, 2M	3 687
PDHA1	300502	Leigh syndrome (MIM 308930); Pyruvate dehydrogenase E1-alpha deficiency (MIM 312170)	Breathing disorder, hypotonia, spasticity, lactic acidosis; hypotonia, seizures, spasticity, cerebellar anomalies	-	7T, 12M, 1 in-frame deletion, 3 in-frame insertions	1 788
PHF6	300414	Borjeson-Forssman-Lehmann syndrome (MIM 301900)	Epilepsy, hypogonadism, hypometabolism, obesity, DF	-	4T, 6M	1 563
PHF8	300560	Siderius mental retardation (MIM 300263)	DF, cleft lift/palate	-	2T, 1M, 1 in-frame deletion	4 332
PLP1	300401	Pelizaeus-Merzbacher disease (MIM 312080)	Nystagmus, spastic paraplegia, ataxia, spasticity, seizures	-	5T, 19M, 1 in 5' UTR, 1 entire gene deletion, 1 entire gene duplication	1 114
PORCN	300651	Focal dermal hypoplasia (MIM 305600, lethal in males)	Skin pigmentation, digit anomalies, visual defects (coloboma)	-	3T, 2M	2 243

PQBP1	300463	Renpenning syndrome (MIM 309500)	Microcephaly, short stature, hypogonadism, coloboma, DF	-	6T, 1M	1 130
PRPS1	311850	Arts syndrome (MIM 301835); Charcot-Marie-Tooth (MIM 311070); Phosphoribosylpyrophosphate Synthetase superactivity (MIM 300661)	Hypotonia, ataxia, deafness, optic atrophy, polyneuropathy, gout, hyperuricemia	-	11M	1 237
PTCHD1	300828	-	Autistic traits, NS-ID	-	6M, 1 in-frame indel	2 787
RAB39B	300774	Mental retardation, X-linked 72 (MIM 300271)	Seizures, autistic traits	-	2T	722
RBM10	300080	TARP syndrome (MIM 311900)	Cardiac anomalies, clubfoot, DF	-	2T	4 481
RPL10	312173	Autism, susceptibility to, X-linked 5 (MIM 300847)	ASD	-	2M	970
RPS6KA3	300075	Coffin-Lowry syndrome (MIM 303600)	Skeletal defects, growth retardation, hearing defects, paroxysmal movement disorders, DF or NS-ID	-	9T, 9M, 2 in-frame deletions, 1 large exon duplication	3 144
SHROOM4	300579	Stocco dos Santos syndrome (MIM 300434)	Short stature, hip luxation	1 stop htz	1M	4 842
SLC16A2	300095	Allan-Herndon-Dudley syndrome (MIM 300523)	Hypotonia, muscular atrophy, joint contractures, spastic paraplegia	-	3T, 5M, 2 large exon deletions, 1 in-frame deletion	2 082
SLC6A8	300036	Cerebral creatine deficiency syndrome 1 (MIM 300352)	Speech delay, seizures, behavioral abnormalities	-	3T, 5M, 2 in-frame deletions	2 729
SLC9A6	300231	Christianson mental retardation (MIM 300243)	Microcephaly, severe speech delay, hypotonia, seizures, impaired ocular movements, drooling	1 splice htz	3T, 2 in-frame deletions	2 746
SMC1A	300040	Cornelia de Lange syndrome 2 (MIM 300590)	Limb defects, growth retardation, epilepsy, cerebellar anomalies, DF	-	3M, 2 in-frame deletions, 1 large exon deletion	5 249
SMS	300105	Snyder-Robinson mental retardation syndrome (MIM 309583)	Hypotonia, marfanoid habitus	-	1T, 2M	1 541
SOX3	313430	Isolated growth hormone deficiency (MIM 300123)	Hypopituitarism	-	3T	
SRPX2	300642	Rolandic epilepsy, mental retardation, and speech dyspraxia (MIM 300643)	-	-	2M	1 798
SYN1	313440	Epilepsy, X-linked, with variable learning disabilities and behavior disorders (MIM 300491)	Autistic traits	-	2T, 2M	2 830
SYP	313475	Mental retardation, X-linked 96 (MIM 300802)	Epilepsy, or NS-ID	-	3T, 1M	1 182
TIMM8A	300356	Jensen syndrome (MIM 311150); Mohr-Tranebjaerg Syndrome (MIM 304700)	Progressive deafness, blindness and dystonia	-	7T, 1M, 1 entire gene deletion	520

TSPAN7	300096	Mental retardation, X-linked 58 (MIM 300210)	NS-ID	-	2T, 1M	1 030
UBE2A	312180	XLID, Nascimento-type (MIM 300860)	DF, synophrys, hypogenitalism	-	1T, 2M	699
UPF3B	300298	Mental retardation, X-linked, syndromic 14 (MIM 300676)	Autistic traits, slender build or NS-ID	1 splice htz	3T, 1M	1 892
ZDHHC15	300576	Mental retardation, X-linked 91 (MIM 300577)	Hypotonia, seizures, limb anomalies, obesity, DF	-	1 translocation	1 534
ZDHHC9	300646	Mental retardation, X-linked syndromic, Raymond type (MIM 300799)	Marfanoid habitus, arachnodactyly, strabismus		2T, 2M	1 455
ZMYM3	300061	-	NS-ID	-	1 translocation	5 296
ZNF41	314995	Mental retardation, X-linked 89 (MIM 300848)	Epilepsy, speech delay or NS-ID	1 splice hemz, 1 stop htz	1T, 1M, 1 translocation	2 500
ZNF674	300573	Mental retardation, X-linked 92 (MIM 300851)	NS-ID	2 stops, in 19 hemz, 5 htz individuals	1T	1 906
ZNF711	314990	Mental retardation, X-linked 97 (MIM 300803)	NS-ID	-	2T	2 566
ZNF81	314998	Mental retardation, X-linked 45 (MIM 300498)	NS-ID	1 stop hemz	1M	2 146
Autosomal Recessive genes						
ACY1	104620	Aminoacylase 1 deficiency (MIM 609924)	Encephalopathy, seizures, hypotonia	-	2T, 4M	2 162
ADRA2B	104260	-	NS-ID	1 stop htz	1M (Najmabadi et al., Nature, 2011)	1 383
ADSL	608222	Adenylosuccinase deficiency (MIM 103050)	Autistic traits, epilepsy, hypotonia	-	7M, 1 5' UTR variant	1 975
ALDH18A1	138250	Cutis laxa, autosomal recessive, type IIIA (MIM 219150)	Visual defects, skin elasticity, hypotonia, joint laxity	2 splices htz	1T, 2M	3 179
ALG6	604566	Congenital disorder of glycosylation, type Ic (MIM 603147)	Pyschomotor delay, hypotonia, ataxia, seizures	-	2T, 3M, 2 in-frame deletions	2 170
ANK3	600465	Mental retardation, autosomal recessive, 37 (MIM 615493)	Autistic traits, ADHD	1 splice htz	1T, 4M, 1 translocation	15 633
AP4B1	607245	Spastic paraplegia 47, autosomal recessive (MIM 614066)	Hypotonia progressing into hypertonia, speech and psychomotor delay	3 stops htz	1T, 1 in-frame insertion	2 610
AP4E1	607244	Spastic paraplegia 51, autosomal recessive (MIM 613744)	Hypotonia progressing into hypertonia, speech and psychomotor delay, seizures	-	2T, 1 large exon deletion	4 368
AP4M1	602296	Spastic paraplegia 50, autosomal recessive (MIM 612936)	Hypotonia progressing into hypertonia, strabismus, speech and psychomotor delay	1 stop htz, 1 splice htz	1T, 1M	2 307
AP4S1	607243	Spastic paraplegia 52, autosomal recessive (MIM 614067)	Hypotonia progressing into hypertonia, speech and psychomotor delay	2 stops, 3 splices in 6 htz individuals	1T	843
ASPM	605481	Microcephaly 5, primary, autosomal recessive (MIM 608716)	Speech delay, seizures	5 stops, 2 splices in 113 htz, 2 hmz individuals	11T	11 796

C12orf57	615140	Temtamy syndrome (MIM 218340)	Coloboma, DF, cerebellar defects, seizures	1 splice htz	2M	498
CACNA1G	604065	-	Cataract	1 splice htz	1T (Najmabadi et al., Nature, 2011)	9 272
CC2D1A	610055	Mental retardation, autosomal recessive 3 (MIM 608443)	NS-ID	2 splices in 14 carriers	1T	4 777
CDK5RAP2	608201	Microcephaly 3, primary, autosomal recessive (MIM 604804)	-	4 stops, 1 splice in 5 htz	2T	7 202
CENPJ	609279	Microcephaly 6, primary, autosomal recessive (MIM 608393); Seckel syndrome 4 (MIM 613676)	Joint stiffness, growth retardation, DF	7 stops in 9 htz	3T, 2M	4 821
CNTNAP2	604569	Cortical dysplasia-focal epilepsy syndrome (MIM 610042); Pitt-Hopkins like syndrome 1 (MIM 610042)	Speech regression, aberrant social interactions, hyperactivity	1 stop htz	4T, 1M, 2 large exon deletions	4 956
CRBN	609262	Mental retardation, autosomal recessive 2 (MIM 607417)	NS-ID	1 stop htz	1T	1 769
ELP2	-	-	NS-ID	4 stops in 5 htz, 1 splice htz	2M (Najmabadi et al., Nature, 2011)	3 661
FOLR1	136430	Neurodegeneration due to cerebral folate transport deficiency (MIM 613068)	Developmental regression, epilepsy, movement disturbances, leukodystrophy	2 splices in 28 htz	3T, 1 in-frame duplication	930
FTCD	606806	Glutamate formiminotransferase deficiency (MIM 229100)	Megaloblastic anemia	6 stops in 24 htz, 1 hmz	1T, 2M	2 283
GAMT	601240	Cerebral creatine deficiency syndrome 2 (MIM 612736)	Developmental delay/regression, speech impariment, seizures	1 stop htz	2T, 3M	1 252
GRIK2	138244	Mental retardation, autosomal recessive, 6 (MIM 611092)	NS-ID	-	1 large deletion/inversion	3 612
HAL	609457	Histidinemia (MIM 235800)	NS-ID or no ID	4 stops in 49 htz, 4 splices in 8 htz & 1 hmz	4M	3 078
HIST1H4B	602829	-	Microcephaly, strabismus	1 stop htz	1T (Najmabadi et al., Nature, 2011)	351
KCNJ10	602208	SESAME syndrome (MIM 612780)	Seizures, deafness, ataxia	1 stop htz	2T, 11M	1 180
KDM5A	180202	-	DF	-	1M (Najmabadi et al., Nature, 2011)	6 165
LAMA1	150320	-	Strabismus	3 stops htz, 2 splices in 2 htz & 1 hmz	1T (Najmabadi et al., Nature, 2011)	11 772
LAMC3	604349	Cortical malformations, occipital (MIM 614115)	Seizures associated with transient loss of vision	2 stops htz, 1 splice in 2 htz	3T, 1M	5 941
LINS	610350	-	Microcephaly	2 stops in 3 htz	1T (Najmabadi et al., Nature, 2011)	2 649
MAN1B1	604346	Mental retardation, autosomal recessive 15 (MIM 614202)	NS-ID, overweight	-	1T, 2M	2 663
MANBA	609489	Mannosidosis, beta (MIM 248510)	Developmental delay	2 stops htz, 4 splices in 5 htz	7T, 1M	3 320

MCPH1	607117	Microcephaly 1, primary, autosomal recessive (MIM 251200)	-	6 stops htz, 2 splices in 3 htz	3T, 3M, 1 large exon deletion	3 076
MED23	605042	Mental retardation, autosomal recessive 18 (MIM 614249)	NS-ID	2 stops htz	1M	5 386
MPI	154550	Congenital disorder of glycosylation, type Ib (MIM 602579)	Diarrhea, vomiting, hypoglycemia, convulsions, liver defects	-	1T, 4M	1 592
MTHFR	607093	Homocystinuria due to MTHFR deficiency (MIM 236250)	Psychiatric matters, muscle weakness, seizures	-	3T, 7M	2 411
NPC2	601015	Niemann-pick disease, type C2 (MIM 607625)	Neurodegeneration, respiratory defects, viscera defects	4 stops htz	7T, 3M	656
NRXN1	600565	Pitt-Hopkins-like syndrome 2 (MIM 614325)	Hyperventilation, autistic traits, developmental delay/regression, constipation, DF	-	1T, 3 large exon deletion	5 863
NSUN2	610916	Mental retardation, autosomal recessive 5 (MIM 611091)	Poor postnatal growth, DF, microcephaly, developmental delay	1 splice htz	4T, 1M	3 133
PARP1	173870	-	Spasticity, hyperreflexia, pes cavus	1 stop htz	1M (Najmabadi et al., Nature, 2011)	3 942
PDHX	608769	Lacticacidemia due to PDX1 deficiency (MIM 245349)	Psychomotor retardation, hypotonia, ataxia, lactic acidosis	2 stops htz (1 in alternative isoform), 1 splice htz	6T, 1M, 1 in-frame deletion, 2 large exon deletions	2 089
PIGV	610274	Hyperphosphatasia with mental retardation syndrome 1 (MIM 239300)	Hypotonia, seizures	1 stop htz	5M	1 602
PMM2	601785	Congenital disorder of glycosylation, type Ia (MIM 212065)	Pshychomotor retardation, hypotonia, hypotreflexia, ataxia, strasbismus	1 stop htz, 1 splice in 2 htz	2T, 20M, 1 large exon deletion	1 061
PRKRA	603424	Dystonia 16 (MIM 612067)	Dystonia, parkinsonism, bradykinesia	-	1T, 1M	1 286
PRSS12	606709	Mental retardation, autosomal recessive 1 (MIM 249500)	NS-ID	3 stops htz, 3 splices in 8 htz	1T	3 148
RABL6	610615	-	NS-ID	1 stop htz	1M (Najmabadi et al., Nature, 2011)	3 188
RALGDS	601619	-	NS-ID	-	1M (Najmabadi et al., Nature, 2011)	3 504
RELN	600514	Lissencephaly 2 (Norman-Roberts type, MIM 257320)	Microcephaly, DF, postnatal growth deficiency, seizures, hypertonia	2 stops htz, 1 splice htz	2T	13 154
SARS	607529	-	-	1 stop htz	1M (Puettmann et al., abstract, 15th MRX workshop, Berlin, 2011)	1 974
SCAPER	611611	-	NS-ID	3 stops htz, 1 in an alternative isoform, 1 splice htz	1T (Najmabadi et al., Nature, 2011)	5 554
SLC46A1	611672	Folate malabsorption, hereditary (MIM 229050)	Megaloblastic anemia, diarrhea, immune deficiency, infections, convulsions	-	4T, 6M	1 581
SOBP	613667	Mental retardation, anterior maxillary protrusion, and strabismus (MIM 613671)	-	-	1T	2 862

SRD5A3	611715	Congenital disorder of glycosylation, type Iq (MIM 612379); Kahrizi syndrome (MIM 612713)	Colobomas, ichthyosis, cerebellar anomalies, kyphosis, DF	1 splice htz	7T	1 152
STIL	181590	Microcephaly 7, primary, autosomal recessive (MIM 612703)	Strabismus, ataxia, seizures	-	3T	4 507
TECR	610057	Mental retardation, autosomal recessive 14 (MIM 614020)	NS-ID	-	1M	1 747
TMEM135	-	-	Microcephaly, congenital cataract	1 stop htz	1M (Najmabadi et al., Nature, 2011)	1 962
TRAPPC9	611966	Mental retardation, autosomal recessive 13 (MIM 613192)	Microcephaly, white matter abnormalities, obesity, autistic traits (hand-flapping)	3 stops htz	4T	4 661
TTI2	614426	Mental retardation, autosomal recessive 39 (MIM 615541)	NS-ID	1 splice htz	1M	1 800
TUSC3	601385	Mental retardation, autosomal recessive 7 (MIM 611093)	NS-ID	1 splice in 3 htz	2T	1 503
UPB1	606673	Beta-ureidopropionase deficiency (MIM 613161)	Hypotonia, microcephaly, seizures	1 stop in 2 htz, 2 splices in 30 htz	2T, 2M	1 555
UROC1	613012	Urocanase deficiency (MIM 276880)	Atypical behaviour, ataxia	1 stop htz	2M	3 111
WDR45L	609226	-	Microcephaly	1 stop htz	1M (Najmabadi et al., Nature, 2011)	1 425
WDR62	613583	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations (MIM 604317)	Psychomotor delay	1 stop htz, 4 stops htz	8T, 5M	6 185
ZC3H14	613279	-	NS-ID	-	1T, 1 large deletion (Pak et al., PNAS, 2011)	3 316
ZNF526	614387	-	NS-ID	1 stop htz	2M (Najmabadi et al., Nature, 2011)	2 052
ZNF697	-	-	NS-ID	-	1M (Puettmann et al., abstract, 15th MRX workshop, Berlin, 2011)	1 716
Autosomal Dominant genes						
AFF3	601464	-	Cerebellar anomalies	-	1 large deletion (Steichen-Gersdorf et al., Clin Genet, 2008)	4 744
ANKRD11	611192	KBG syndrome (MIM 148050)	Macrodontia, DF, short stature, skeletal anomalies, seizures	-	3T	8 517
ARHGEF4	605216	-	ADHD, epilepsy, behavioral abnormalities	2 stops htz	1 recurrent large deletion or reciprocal duplication in 2q21.1 (Dharmadhikari et al., Hum Mol Genet, 2012)	2 662
ARID1B	614556	Mental retardation, autosomal dominant 12 (MIM 614562)	Psychomotor delay, hypotonia, short stature	-	10T	7 550
CACNG2	602911	Mental retardation, autosomal dominant 10 (MIM 614256)	NS-ID	-	1M	1 132

CDH15	114019	Mental retardation, autosomal dominant 3 (MIM 612580)	Limb anomalies, DF	1 frequent stop (in 11 hmz, 451 htz), 1 splice htz	3M, 1 translocation	3 005
CIC	612082	-	Development delay	-	1M (Vissers et al., Nat genet, 2010)	6 225
CREBBP	600140	Rubinstein-Taybi syndrome (MIM 180849)	Talon cusps, glaucoma, broad thumbs and toes, DF	1 stop htz	4T, 4M	8 569
DEAF1	602635	-	NS-ID, speech impairment	-	3M (Vissers et al., Nat genet, 2010; Rauch et al., Lancet, 2012)	2 178
DISC1	605210	Schizophrenia, susceptibility (MIM 181500 & 604906)	Schizophrenia	4 stops htz, 1 in an alternative isoform	-	4 154
DLG2	603583	-	Epilepsy, ASD	1 stop htz, 1 stop loss htz	1 large exon deletion (Nillesen et al., 15th MRX workshop, Berlin, 2011)	4 642
DYRK1A	600855	Mental retardation, autosomal dominant 7 (MIM 614104)	Epilepsy, microcephaly, speech delay, feeding anomalies, stereotypic features	-	4T, 1 large exon deletion	2 805
EPB41L1	602879	Mental retardation, autosomal dominant 11 (MIM 614257)	Hypotonia	-	1M	3 486
EHMT1	607001	Kleefstra syndrome (MIM 610253)	Hypotonia, brachy/microcephaly, epilepsy, synophrys, cardiac defects, DF	-	3T, 1M	5 062
FOXG1	164874	Rett syndrome, congenital variant (MIM 613454)	Progressive microcephaly, hypotonia, unresponsiveness, apraxia,autistic traits, no speech, corpus callosum hypoplasia	-	9T, 1M	1 510
FOXP1	605515	Mental retardation with language impairment and autistic features (MIM 613670)	-	-	1T, 1 large exon deletion	3 037
GRIN1	138249	Mental retardation, autosomal dominant 8 (MIM 614254)	Epilepsy, speech delay	1 stop htz	1M, 1 in-frame duplication	4 235
GRIN2A	138253	Epilepsy, focal, with speech disorder and with or without mental retardation (MIM 245570)	Hypotonia, behavioral abnormalities	-	1T, 3M, 1 translocation	4 667
GRIN2B	138252	Mental retardation, autosomal dominant 6 (MIM 613970)	NS-ID	-	6T, 3M, 2 translocations	4 935
HDAC4	605314	Brachydactyly-mental retardation syndrome (MIM 600430)	Short stature, eczema, behavioral abnormalities	-	2T	4 338
HRAS	190020	Congenital myopathy with excess of muscle spindles (MIM 218040); Costello syndrome (MIM 218040)	Short stature, feeding difficulties, growth retardation, cardiac defects, DF	2 stops in 3 htz	13M, 2 in-frame duplications	887
KCNK9	605874	Birk-Barel mental retardation dysmorphism syndrome (MIM Birk-Barel mental retardation dysmorphism syndrome)	Hypotonia, hyperactive, feeding difficulties, DF	-	1M	1 205
KCNQ2	602235	Epileptic encephalopathy, early infantile, 7 (MIM 613720)	Hypotonia, dystonia, spasticity	-	1T, 5M	3 363

KIRREL3	607761	Mental retardation, autosomal dominant 4 (MIM 612581)	DF	-	3M, 1 translocation	3 124
MBD5	611472	Mental retardation, autosomal dominant 1 (MIM 156200)	Developmental delay, seizures, autistic traits, ataxia	-	1 large exon deletion	4 885
MEF2C	600662	Chromosome 5q14.3 deletion syndrome / Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations (MIM 613443)	ADHD, autistic traits, poor eye contact, speech delay, hypotonia, DF	-	3T, 2M	2 019
NRXN2	600566	-	ASD	2 splice htz	1T (Gauthier et al., Hum Genet, 2011)	6 324
PACS1	607492	Mental retardation, autosomal dominant 17 (MIM 615009)	Cranofacial anomalies, genital abnormalities	-	1M	3 966
PAFAH1B1	601545	Lissencephaly 1, Subcortical laminar heterotopia (MIM 607432)	Spastiticty, seizures, hypotonia, microcephaly, DF	-	3T, 7M	1 633
PGA5	169730	-	Hypotonia, developmental delay	1 splice htz	1M (Vissers et al., Nat genet, 2010)	1 589
RAI1	607642	Smith-Magenis syndrome (MIM 182290)	Hypotonia, speech delay, hearing loss, sleep disturbances, DF	-	5T, 2M	5 881
SCN1A	182389	Dravet syndrome (MIM 607208); Epilepsy, generalized, with febrile seizures plus, type 2 (MIM 604403)	Absences, ataxia, mental decline	-	5T, 15M, 2 large exon deletions	7 070
SCN2A	182390	Epileptic encephalopathy, early infantile, 11 (MIM 613721)	Seizures, spasticity, developmental delay	-	1T, 3M	7 245
SHANK1	604999	-	ASD with high functioning	-	2 large exon deletions (Sato et al., AJHG, 2012)	7 383
SHANK2	603290	Autism susceptibility 17 (MIM 613436)	ASD	-	1T, 2 large exon deletions	6 328
SHANK3	606230	Phelan-McDermid syndrome (MIM 606232)	Neonatal hypotonia, developmental delay, speech delay, autistic traits	-	3T, 1M	6 403
SLC2A1	138140	GLUT1 deficiency syndrome 1 (MIM 606777); GLUT1 deficiency syndrome 2 (MIM 612126)	Epileptic encephalopathy, developmental delay, microcephaly, ataxia, spasticity, seizures, sleep disturbances	-	2T, 10M, 1 entire gene deletion, 1 in-frame deletion, 1 in-frame insertion	1 931
STXBP1	602926	Epileptic encephalopathy, early infantile, 4 (MIM 612164)	Seizures, spasticity, brain malformations	-	3T, 4M	2 695
SYNGAP1	603384	Mental retardation, autosomal dominant 5 (MIM 612621)	Developmental delay, seizures, autistic traits	-	8T, 2M	4 811
TCF4	602272	Pitt-Hopkins syndrome (MIM 610954)	Hyperventilation, sleep apneas, intestinal abnormalities, clubbed toes & fingers, DF	2 stops htz	3T, 3M	
UBE3A	601623	Angelman syndrome (MIM 105830)	Gait disorder, abnormal behavior, epilepsy, speech limitations, DF	-	9T, 2M	3 189
YY1	600013	-	Microcephaly, develomental delay	-	1M (Vissers et al., Nat genet, 2010)	1 445

ZBTB20	606025	-	ASD, NS-ID	-	2M, 1 translocation, 1 large deletion (Srivastava et al., asbtract, ICHG, Montreal, 2011 and ASHG, Boston, 2013)	2 382
ZEB2	605802	Mowat-Wilson syndrome (MIM 235730)	Developmental delay, epilepsy, neurocristopathy	-	12T, 3M, 1 in-frame deletion, 1 large exon deletion	4 005
ZNF599	-	-	Hypotonia, developmental delay	4 stops in 47 htz, 1 hmz; 1 splice htz	1M (Vissers et al., Nat genet, 2010)	1 927
Others						
DOCK8	611432	Mental retardation, autosomal dominant 2 (MIM 614113) ; Hyper-IgE recurrent infection syndrome, autosomal recessive (MIM 243700)	Developmental delay, poor speech	1 stop htz, 1 splice htz	1 large deletion, 1 translocation	8 220
KIF1A	601255	Mental retardation, autosomal dominant 9 (MIM 614255); Spastic paraplegia 30, autosomal recessive (MIM 610357)	Hypotonia, spasticity, cerebellar anomalies, hyperrefkexia,	2 stops htz	1M ; 2M	7 036
MED13L	608771	Transposition of the great arteries, dextro-looped 1, autosomal dominant (MIM 608808); ID with cardiac anomalies, autosomal dominant; NS-ID, autosomal recessive;	Hypotonia, cardiac defects, ataxia and DF; NS-ID;	-	1 translocation, 3M (Munckle et al., circulation, 2003); 2 large exon deletions, 1 triplication (Asadollahi, EJHG, 2013) 1 M (Najmabadi et al., Nature, 2011);	7 842
PRODH	606810	Hyperprolinemia, type I (MIM 239500); Schizophrenia, susceptibility to, 4 (MIM 600850)	Renal abnormalities, hearing loss, epilepsy	-	8M, 1 large exon deletion	2 466
PRRT2	614386	Convulsions, familial infantile, with paroxysmal choreoathetosis (MIM 602066) autosomal dominant; Episodic kinesigenic dyskinesia 1 (MIM 128200) autosomal dominant; BFIS/PKD with ID: autosomal recessive	Ataxia, absences	-	1T (Labate et al., Epilepsia, 2012)	1 140
DIP2B	611379	Mental retardation, FRA12A type (MIM 136630)	NS-ID	-	1 CGGn 5'UTR expansion	6 347
CYFIP1	606322	-	-	2 stops in 17 htz	-	

T: truncating mutation, M: missense mutation

Table S9: list of variants passing our filtering criteria in each patient with corresponding annotations by VaRank

DNA ID	Sex	MOI	Segregation	Gene	Omim #	Transcript#	Coverage (X)	Reads variation (%)	Nomenclature (g.)	Nomenclature (c.)	Nomenclature (p.)	rs #	Clinical Significance	EVS carriers	EVS MAF	DMES (%)	DSSF (%)	DNNS (%)	Local splice effect (prediction)	Sift	PPH2	phyloP	Phast Cons	Grant ham	All Count	Conclusion
		AD		*SHANK1*	604999	NM_016148.2	24	58	g.51171270C>T	c.3947G>A	p.Gly1316Asp	-	-	-	-	0	0	0	-	Tolerated	neutral	1.174	0.992	94	1 htz	Possibly benign
APN-1	M	AR		*LAMA1*	150320	NM_005559.2	273	44	g.7017272C>T	c.2808+5G>A	p.?	rs201030108	-	24 (htz)	0,18%	-63.5	-14.8	-88.3	-	NA	NA	3.837	1,00	NA	1 htz	VOUS
				SLC46A1	611672	NM_080669.3	139	48	g.6137A>T	c.623A>T	p.Tyr208Phe	rs201837257	-	-	-	0	0	0	-	Deleterious	neutral	NA	0.098	22	1 htz	VOUS
		?		*CYFIP1*	606322	NM_001033028.1	310	47	g.22956361A>G	c.305A>G	p.His102Arg	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.44	0,00	29	1 htz	Possibly benign
		XL	inherited (Mo)	*IQSEC2*	300522	NM_001111125.1	163	99	g.53265591G>A	c.3364C>T	p.Arg1122Cys	-	-	-	-	0	0	0	-	Tolerated	deleterious	3.03	1,00	180	1 hemz	VOUS
				ALDH18A1	138250	NM_002860.3	311	46	g.97393376C>G	c.589G>C	p.Glu197Gln	rs201691969	-	-	-	0	0	0	-	Deleterious	deleterious	5.371	1,00	29	1 htz	VOUS
APN-2	M			*RELN*	600514	NM_005045.3	110	52	g.103629739C>A	c.65G>T	p.Arg22Met	rs139532757	-	1	0,01%	0	0	0	-	Deleterious	neutral	1.739	1,00	91	1 htz	VOUS
		AR		*LAMA1*	150320	NM_005559.2	412	46	g.6982519T>C	c.5867A>G	p.Asn1956Ser	rs117433399	-	16 (htz)	0,12%	0	0	0	-	Tolerated	neutral	0.044	0,00	46	1 htz	Possibly benign
				TUSC3	601385	NM_006765.3	142	54	g.15397977C>T	c.38C>T	p.Ala13Val	rs200808372	-	4 (htz)	0,03%	0	0	0	-	Deleterious	neutral	0.69	0.976	64	2 htz	VOUS
				ZNF526	614387	NM_133444.1	176	43	g.42729310_42729312del	c.755_757del	p.Glu252del	rs138669870	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	2 htz	VOUS
		XL		*MID1*	300552	NM_001193278.1	223	99	g.10469476C>T	c.880G>A	p.Glu294Lys	rs111428432	-	-	-	0	0	0	-	Notscored	neutral	-1.409	0,00	56	1 hemz	VOUS
		AD		*ARHGEF4*	605216	NM_032995.1	144	51	g.131802007G>A	c.1735G>A	p.Ala579Thr	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	-1.328	0,00	58	1 htz	Possibly benign
				AP4E1	607244	NM_007347.3	368	49	g.51260460G>A	c.1852G>A	p.Val618Ile	rs142215198	-	4	0,03%	-23.7	-4.8	-11.7	-	Tolerated	neutral	4.483	1,00	29	1 htz	Possibly benign
APN-3	M			*HAL*	609457	NM_002108.2	219	49	g.96388732G>A	c.287C>T	p.Ser96Phe	rs142371886	-	12 (htz)	0,09%	NA	0	0	-	Deleterious	neutral	2.95	0.858	155	1 htz	VOUS
		AR		*NRXN1*	600565	NM_001135659.1	281	53	g.50847195G>A	c.1405C>T	p.Pro469Ser	rs78540316	-	34 (htz)	0,28%	0	0	0	-	NA	deleterious	6.42	1,00	74	1 htz	VOUS
				TRAPPC9	611966	NM_031466.5	229	51	g.141461333C>T	c.434G>A	p.Arg145Gln	rs147182402	-	7 (htz)	0,05%	0	0	0	-	Tolerated	neutral	3.111	0.992	43	1 htz	Possibly benign
				TTI2	614426	NM_025115.3	135	50	g.33361318G>A	c.1063C>T	p.Arg355Cys	rs138108276	-	50 (htz)	0,38%	0	0	0	-	Deleterious	deleterious	2.788	1,00	180	1 htz	VOUS
				PMM2	601785	NM_000303.2	475	48	g.8905014T>G	c.426T>G	p.Ile142Met	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.198	0.937	10	1 htz	Possibly benign
APN-4	M	AR		*KDM5A*	180202	NM_001042603.1	365	40	g.432809G>A	c.2107C>T	p.Pro703Ser	-	-	-	-	0	0	0	-	Tolerated	neutral	2.385	1,00	74	1 htz	Possibly benign
				CACNA1G	604065	NM_198396.1	360	49	g.48653594G>T	c.1831G>T	p.Ala611Ser	-	-	-	-	0	0	0	-	Tolerated	neutral	3.111	0.992	99	1 htz	Possibly benign
APN-7	M	AD	inherited (Fa)	*CREBBP*	600140	NM_004380.2	405	45	g.3827646G>A	c.2126C>T	p.Ser709Phe	rs147740753	other	-	-	0	0	-0.7	-	Deleterious	deleterious	5.371	1,00	155	1 htz	Benign
		AD		*CIC*	612082	NM_015125.3	173	47	g.42797323C>G	c.3685C>G	p.Pro1229Ala	-	-	-	-	0	0	0	-	Tolerated	neutral	0.69	0.677	27	1 htz	Possibly benign
				DEAF1	602635	NM_021008.2	329	52	g.679660G>A	c.1126+28C>T	p.?	-	-	-	-	0	0	0	Cryptic ASS	NA	NA	-1.247	0,00	NA	1 htz	Possibly benign
APN-8	M			*LAMA1*	150320	NM_005559.2	263	34	g.6947145_6947161del	c.8844+1_8844+17del	p.?	-	-	-	-	-100	-100	-100	-	NA	NA	NA	NA	NA	1 htz	VOUS
		AR		*TRAPPC9*	611966	NM_031466.5	278	41	g.140922432C>T	c.3217G>A	p.Gly1073Ser	-	-	-	-	NA	NA	NA	-	Tolerated	neutral	2.546	0.984	56	1 htz	Possibly benign
				SCAPER	611611	NM_001145923.1	593	47	g.76994121C>T	c.1748G>A	p.Arg583His	-	-	-	-	0	0	0	-	Tolerated	NA	5.694	0.992	29	1 htz	VOUS
		XL		*NLGN3*	300336	NM_181303.1	86	97	g.70367619C>T	c.20C>T	p.Pro7Leu	rs199925687	-	3 (2F, 1M)	0,03%	0	0	0	-	Tolerated	neutral	-0.117	0.016	98	1 hemz	Possibly benign
		AD	inherited (Mo)	*MBD5*	611472	NM_018328.4	415	46	g.149226102T>C	c.590T>C	p.Val197Ala	-	-	-	-	0	0	0	-	Deleterious	neutral	0.125	0.976	64	1 htz	Benign
APN-9	M			*CENPJ*	609279	NM_018451.3	509	51	g.25478104T>C	c.2785A>G	p.Lys929Glu	rs141844033	-	9 (htz)	0,07%	0	0	0	-	Deleterious	deleterious	4.483	1,00	56	1 htz	VOUS
		AR		*PARP1*	173870	NM_001618.3	180	48	g.226570748G>T	c.1148C>A	p.Ser383Tyr	rs3219062	-	27 (htz)	0,21%	0	0	0	-	Deleterious	neutral	4.887	0.362	144	1 htz	VOUS
				CACNA1G	604065	NM_198397.1	231	42	g.48683469G>A	c.4507G>A	p.Gly1503Ser	-	-	-	-	0	0	0	New ASS	Deleterious	neutral	-1.651	0,00	56	1 htz	Possibly benign
				RABL6	610615	NM_001173988.1	260	46	g.139733769C>T	c.1592C>T	p.Pro531Leu	rs143011384	-	38 (htz)	0,30%	0	0	0	-	Tolerated	NA	0.851	0,00	98	1 htz	VOUS
		XL	inherited (Mo)	*HUWE1*	300697	NM_031407.4	158	96	g.53596663G>C	c.6437C>G	p.Thr2146Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.595	1,00	71	1 hemz	Possibly benign
		AD	Absent from Mo	*GRIN2A*	138253	NM_001134407.1	178	48	g.9862749C>T	c.2554G>A	p.Val852Met	rs150316865	-	-	-	0	0	0	-	Deleterious	deleterious	2.788	1,00	21	1 htz	VOUS
APN-10	M			*PMM2*	601785	NM_000303.2	349	44	g.8905010G>A	c.422G>A	p.Arg141His	rs28936415	**pathogenic**	50 (htz)	0,39%	0	0	0	-	Deleterious	deleterious	5.694	1,00	29	2 htz	VOUS
		AR		*LAMA1*	150320	NM_005559.2	346	39	g.6959394G>A	c.7724C>T	p.Thr2575Met	rs76482057	-	42 (htz)	0,32%	0	0	0	Cryptic ASS	Deleterious	neutral	-0.44	0,00	81	1 htz	VOUS
				RABL6	610615	NM_001173988.1	234	50	g.139726282G>A	c.568G>A	p.Asp190Asn	rs199805458	-	3 (htz)	0,02%	0	0	0	Cryptic DSS	Tolerated	deleterious	5.29	1,00	23	1 htz	VOUS
APN-11	M	AD		*ANKRD11*	611192	NM_001256182.1	76	50	g.89346825T>C	c.6125A>G	p.Asp2042Gly	-	-	-	-	0	0	0	-	Tolerated	NA	0.205	0.039	94	1 htz	VOUS
		AR		*CDK5RAP2*	608201	NM_018249.4	286	43	g.123230241T>A	c.2003A>T	p.Tyr668Phe	-	-	-	-	0	0	0	-	Tolerated	deleterious	2.707	0.315	22	1 htz	VOUS
		XL		*SLC9A6*	300231	NM_001042537.1	31	38	g.135067686G>T	c.25G>T	p.Ala9Ser	rs201523857	-	-	-	NA	0	NA	-	Tolerated	neutral	0.044	0,00	99	1 htz	Possibly benign
APN-12	F	AD		*KCNQ2*	602235	NM_172107.2	129	51	g.62038152C>T	c.2464G>A	p.Ala822Thr	-	-	-	-	0	NA	0	-	Tolerated	neutral	1.255	0.921	58	1 htz	Possibly benign
		AR		*WDR62*	613583	NM_001083961.1	136	52	g.36594882C>T	c.4152C>T	p.=	-	-	-	-	-7.3	0.5	-2.7	-	NA	NA	-0.117	0.441	NA	1 htz	VOUS
		XL	inherited (Mo)	*SRPX2*	300642	NM_014467.2	84	94	g.99920309del	c.602del	p.Ala201Valfs*10	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 hemz	Possibly benign
				LAMA1	150320	NM_005559.2	168	49	g.7049112C>G	c.733G>C	p.Glu245Gln	-	-	-	-	0	0	0	-	Tolerated	deleterious	6.259	1,00	29	1 htz	VOUS
APN-13	M	AR		*SCAPER*	611611	NM_001145923.1	275	44	g.77067396T>C	c.97A>G	p.Thr33Ala	rs142461932	-	12 (htz)	0,10%	0	0	0	New ASS	Tolerated	NA	0.528	0.984	58	1 htz	Possibly benign
				STIL	181590	NM_001048166.1	264	46	g.47726135T>C	c.2906A>G	p.His969Arg	rs148193936	-	21 (htz)	0,16%	0	0	0	-	Tolerated	neutral	0.69	0.024	29	1 htz	Possibly benign
				RALGDS	601619	NM_001042368.1	247	45	g.135983956T>C	c.613+104A>G	p.?	rs111236084	-	-	-	0	0	0	New DSS	NA	NA	-0.36	0,00	NA	2 htz	VOUS
			de novo	*MED13L*	608771	NM_015335.4	239	35	g.116406845_116406852del	c.6118_6125del	p.Gly2040Asnfs*32	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	Pathogenic
		AD		*ARID1B*	614556	NM_020732.3	83	45	g.157100380G>C	c.1317G>C	p.Glu439Asp	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.117	0.937	45	1 htz	Possibly benign
APN-14	M			*YY1*	600013	NM_003403.3			g.100705788_100705790del	c.222_224del	p.His80del	-	-	-	-	0.0	0.0	0.0	-	NA	NA	NA	NA	NA	1 htz	VOUS
				PMM2	601785	NM_000303.2	285	51	g.8905010G>A	c.422G>A	p.Arg141His	rs28936415	**pathogenic**	50 (htz)	0,39%	0	0	0	-	Deleterious	deleterious	5.694	1,00	29	2 htz	VOUS
		AR		*HRAS*	190020	NM_005343.2	222	50	g.532627T>A	c.*5+4A>T	p.?	-	-	-	-	-37	-11.2	-20.3	-	NA	NA	2.707	1,00	NA	1 htz	VOUS
				CC2D1A	610055	NM_017721.4	153	52	g.14030696G>A	c.1288G>A	p.Glu430Lys	-	-	-	-	0	0	0	-	Tolerated	neutral	3.514	1,00	56	1 htz	Possibly benign
APN-15	M																								VOUS
		XL	inherited (Mo)	*MAOA*	NA	NM_000240.2	95	93	g.43590942G>T	c.797G>T	p.Cys266Phe	-	-	-	-	-0.7	0	18.1	-	Deleterious	NA	3.434	1,00	205	1 hemz	Pathogenic
APN-16	M	AD	inherited (Mo)	*HDAC4*	605314	NM_006037.3	45	57	g.240016711C>T	c.2260G>A	p.Val754Ile	rs151043798	-	1	0,01%	0	0	0.1	-	Tolerated	neutral	2.465	1,00	29	1 htz	Benign
			inherited (Fa)	*DOCK8*	611432	NM_203447.3	276	43	g.161649A>G	c.1954A>G	p.Ile652Val	-	-	-	-	0	0	0	-	Tolerated	deleterious	NA	1,00	29	1 htz	Benign
		XL	inherited (Mo)	*FLNA*	300017	NM_001110556.1	280	98	g.153588207G>A	c.3872C>T	p.Pro1291Leu	rs137853319	**pathogenic**	1	0,01%	0	0	0	-	Deleterious	neutral	1.739	0.039	98	1 hemz	VOUS
APN-17	M	AD	inherited (Mo)	*PACS1*	607492	NM_018026.2	406	52	g.66010561C>T	c.2776+59C>T	p.?	-	-	-	-	0	0	0	New DSS	NA	NA	-0.117	0.016	NA	1 htz	Benign
		AR		*CENPJ*	609279	NM_018451.3	477	50	g.25480913C>G	c.1263G>C	p.Gln421His	rs201088712	-	1	0,01%	0	0	0	-	Deleterious	deleterious	1.255	1,00	24	1 htz	VOUS
		AD		*SHANK2*	603290	NM_133266.3	318	47	g.70331576C>T	c.3058G>A	p.Val1020Ile	-	-	-	-	0	0	0	-	Tolerated	NA	0.205	0.024	29	1 htz	VOUS
				PIGV	610274	NM_001202554.1	283	45	g.27117322C>G	c.16C>G	p.Pro6Ala	-	-	-	-	0	0	0	-	Tolerated	neutral	2.465	0.984	27	1 htz	Possibly benign
APN-18	M	AR		*AP4M1/TAF6*	602296	NM_004722.3	256	44	g.99704100G>A	c.1100G>A	p.Arg367Gln	rs139861201	-	5 (htz)	0,04%	0	0	0	New ASS	Deleterious	deleterious	4.725	1,00	43	1 htz	VOUS
				UROC1	613012	NM_001165974.1	292	49	g.126227314G>C	c.356C>G	p.Pro119Arg	rs200128917	-	2 (htz)	0,02%	0	0	0.6	-	Deleterious	deleterious	4.887	1,00	103	1 htz	VOUS
APN-19	M																								VOUS
		XL		*NSDHL*	300275	NM_001129765.1	120	98	g.152037637C>T	c.1099C>T	p.Arg367Cys	rs199567542	-	-	-	0	0	0	-	Tolerated	neutral	2.223	0.984	180	1 hemz	Possibly benign
APN-24	M	AR		*TTI2*	614426	NM_025115.3	268	47	g.33361016C>T	c.1190G>A	p.Gly397Glu	rs150984360	-	14 (htz)	0,11%	0	0	0	-	Tolerated	neutral	1.255	0.992	98	2 htz	Possibly benign
				C12orf57/ATN1	615140	NM_138425.2	233	48	g.7053298C>G	c.14C>G	p.Ser5Trp	-	-	-	-	0	0	0	-	Deleterious	neutral	3.434	0.717	177	1 htz	VOUS
				PGA5	169730	NM_014224.2	16	43	g.61013499G>A	c.538G>A	p.Asp180Asn	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.918	0.984	23	2 htz	VOUS
		AD		*DISC1*	605210	NM_001164537.1	127	51	g.231762651C>G	c.38C>G	p.Ala13Gly	rs79978593	-	-	-	0	0	0	-	Tolerated	neutral	-0.279	0,00	60	1 htz	Possibly benign
APN-25	M			*ZNF599*	NA	NM_001007248.2	295	50	g.35250893C>T	c.813G>A	p.=	rs201514857	-	1	0,01%	0	0	0	New ASS	NA	NA	0.69	1,00	NA	1 htz	VOUS
		AR		*UPB1*	606673	NM_016327.2	265	47	g.24896113C>G	c.143C>G	p.Ser48Cys	rs145989498	-	6	0,05%	0	0	0	-	Tolerated	neutral	0.448	0,00	112	2 htz	Possibly benign
				LAMA1	150320	NM_005559.2	227	45	g.7046445T>C	c.769-79A>G	p.?	rs517111	-	-	-	0	0	0	New DSS	NA	NA	-0.36	0,00	NA	1 htz	VOUS
APN-26	M	XL		*RELN*	600514	NM_005045.3	343	46	g.103143571A>G	c.8381T>C	p.Leu2794Ser	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.483	1,00	145	1 htz	VOUS
		AD		*HDAC4*	605314	NM_006037.3	493	49	g.240036862C>T	c.1663G>A	p.Gly555Ser	rs199936119	-	1	0,01%	0	0	0	-	Tolerated	neutral	0.69	0.008	56	1 htz	Possibly benign
APN-28	M		inherited (Mo)	*ANKRD11*	611192	NM_001256182.1	265	50	g.89345850G>C	c.7100C>G	p.Thr2367Ser	-	-	-	-	0	0	0	New ASS	Tolerated	NA	3.999	0.52	58	1 htz	Benign
		AR		*NRXN1*	600565	NM_138735.2	150	34	g.50574009_50574023del	c.65_79del	p.Gly22_Gly26del	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
				CDK5RAP2	608201	NM_018249.4	318	52	g.123230079A>T	c.2106+59T>A	p.?	rs75136083	-	-	-	0	0	0	Cryptic DSS	NA	NA	0.367	0.016	NA	1 htz	VOUS
		inherited (Mo)		*ARID1B*	614556	NM_017519.2	489	46	g.157522410G>A	c.4643G>A	p.Arg1548His	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.451	1,00	29	1 htz	Benign
		AD		*SHANK2*	603290	AB208026.1	509	48	g.70319434T>C	c.3953A>G	p.Tyr1318Cys	-	-	-	-	0	0	0	-	Tolerated	deleterious	0.609	0.567	194	1 htz	VOUS
				KCNQ2	602235	NM_172107.2	306	46	g.62050979G>A	c.1294C>T	p.Arg432Cys	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	1.82	1,00	180	1 htz	Possibly benign
APN-29	M			*KCNK9*	605874	NM_016601.2	426	47	g.140630506C>T	c.1120G>A	p.Val374Ile	rs149047706	-	1	0,01%	0	0	65.8	-	Tolerated	neutral	-0.037	0.866	29	1 htz	Possibly benign
				UROC1	613012	NM_144639.2	348	48	g.126219691G>A	c.992C>T	p.Thr331Met	rs142914763	-	2	0,02%	0	0	0	-	Tolerated	deleterious	0.77	0.63	81	1 htz	VOUS
		AR		*MPI*	154550	NM_002435.1	475	48	g.75189471A>G	c.964A>G	p.Arg322Gly	-	-	-	-	0	0	0	-	Tolerated	neutral	1.416	1,00	125	1 htz	Possibly benign
				PRSS12	606709	NM_003619.3	236	47	g.119273848G>C	c.28C>G	p.Leu10Val	rs143520192	-	50 (htz)	0,38%	0	0	0	-	Tolerated	neutral	0.609	0.835	32	1 htz	Possibly benign
				FTCD	606806	NM_006657.2	298	50	g.47558456C>T	c.1409G>A	p.Arg470Gln	rs61735839	-	7	0,06%	0	0	0	-	Tolerated	neutral	-0.521	0,00	43	1 htz	Possibly benign

APN-30	M	AR		*CC2D1A*	610055	NM_017721.4	269	46	g.14040420G>A	c.2657G>A	p.Arg886His	rs201921029	-	-	12	0,10%	0	0	0	-	Deleterious	neutral	1.013	1,00	29	1 htz	VOUS
			inherited (Fa)	*FTCD*	606806	NM_006657.2	324	53	g.47571578C>A	c.530G>T	p.Gly177Val	-	-	-	-		0	0	0	-	Deleterious	deleterious	5.532	1,00	109	1 htz	Benign
			inherited (Fa)	*FTCD*	606806	NM_006657.2	170	52	g.47566194C>G	c.954G>C	p.Lys318Asn	-	-	-	-		0	0	0	-	Deleterious	deleterious	0.205	0.472	94	1 htz	Benign
APN-31	M	AR		*SCAPER*	611611	NM_001145923.1	509	48	g.77067254T>C	c.239A>G	p.Glu80Gly	rs186407258	-	-	29	0,24%	0	0	0	Cryptic ASS	Deleterious	NA	2.304	0.583	98	1 htz	VOUS
				SOBP	613667	NM_018013.3	202	48	g.107955777C>T	c.1729C>T	p.Pro577Ser	rs200685194	-	-	8	0,07%	0	0	0	-	Tolerated	neutral	0.69	0.417	74	1 htz	Possibly benign
				RELN	600514	NM_005045.3	414	45	g.103214642C>T	c.4408G>A	p.Val1470Ile	rs143213152	-	-	33	0,25%	0	0	0	-	Tolerated	neutral	1.981	1,00	29	1 htz	Possibly benign
			inherited (Fa)	*HDAC4*	605314	NM_006037.3	556	51	g.240036898G>A	c.1627C>T	p.Arg543Trp	rs140814864	-	-	1	0,01%	0	0	0	-	Deleterious	deleterious	3.837	1,00	101	1 htz	Benign
APN-32	M	AD	inherited (Mo)	*SHANK2*	NA	NM_012309.3	561	48	NA	c.227C>T	p.Pro76Leu	rs199717803	-	-	1	-	NA	NA	NA	-	NA	deleterious	NA	NA	NA	1 htz	Benign
				SHANK3	606230	NM_001080420.1	243	47	g.51169586_51169588del	c.5090_5092del	p.His1697del	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
		AR		*RELN*	600514	NM_005045.3	726	52	g.103151360G>A	c.8212C>T	p.Arg2738Trp	rs202166176	-	-	-	-	0	0	0	-	Deleterious	deleterious	1.577	1,00	101	1 htz	VOUS
				NHS	300457	NM_198270.2	65	98	g.17394091C>T	c.211C>T	p.Pro71Ser	-	-	-	-		0	0	0	-	Deleterious	neutral	-0.117	0.016	74	1 hemz	VOUS
		XL		*IGBP1*	300139	NM_001551.2	294	99	g.69370146A>C	c.845A>C	p.Asp282Ala	-	-	-	-		0	0	0	-	Tolerated	neutral	2.223	1,00	126	1 hemz	Possibly benign
APN-33	M			*PORCN*	300651	NM_203473.1	238	98	g.48375565C>G	c.1159-6C>G	p.?	-	-	-	-		-26.3	-1.2	-14	-	NA	NA	1.255	0.071	NA	1 hemz	VOUS
		AR		*AP4B1*	607245	NM_006594.2	624	48	g.114442873G>A	c.767C>T	p.Thr256Ile	rs143286419	-	-	14	0,11%	0	0	0	-	Tolerated	neutral	2.788	1,00	89	2 htz	Possibly benign
				MED23	605042	NM_004830.2	478	49	g.131939442T>C	c.780+105A>G	p.?	rs6923189	-	-	-	-	0	0	0	Cryptic DSS	NA	NA	0.125	0.008	NA	1 htz	VOUS
		XL	de novo	*KDM5C*	314690	NM_004187.5	162	100	g.53228250C>G	c.2152G>C	p.Ala718Pro	-	-	-	-		0	0	0	-	Deleterious	deleterious	4.967	1,00	27	1 hemz	Pathogenic
		AD		*CDH15*	114019	NM_004933.2	312	48	g.89253963C>A	c.770C>A	p.Ala257Asp	-	-	-	-		0	0	0	-	Deleterious	neutral	2.627	0.961	126	1 htz	VOUS
APN-34	M			*DOCK8*	611432	NM_203447.3	299	49	g.176467A>G	c.2779A>G	p.Ile927Val	-	-	-	-		11	4.7	7.2	-	Tolerated	neutral	NA	0.024	29	1 htz	Possibly benign
				CC2D1A/PODNL1	610055	NM_017721.4	144	51	g.14040915A>G	c.2735A>G	p.Gln912Arg	-	-	-	-		0	0	0	-	Deleterious	neutral	3.03	0.992	43	1 htz	VOUS
		AR		*RABL6*	610615	NM_001173988.1	173	39	g.139734799_139734801del	c.2018_2020del	p.Lys673del	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
		AD		*CREBBP*	600140	NM_004380.2	243	51	g.3778320G>A	c.6728C>T	p.Ala2243Val	rs190153828	-	-	1	0,01%	0	0	0	-	Deleterious	neutral	2.707	0.992	64	1 htz	VOUS
APN-35	M		inherited (Mo)	*SHANK2*	NA	NM_012309.3	263	53	NA	NA	NA	.	-	-	-	-	NA	NA	NA	-	NA	NA	NA	NA	NA	1 htz	Benign
		AR	inherited (Fa)	*RELN*	600514	NM_005045.3	289	50	g.103207043C>T	c.4747+5G>A	p.?	-	-	-	-	-	-41.8	-13.3	-8	Cryptic ASS	NA	NA	1.416	0.11	NA	1 htz	VOUS
		XL		*TSPAN7*	300096	NM_004615.3	188	99	g.38530697G>A	c.338G>A	p.Arg113His	-	-	-	-		0	0	0	-	Tolerated	neutral	3.999	1,00	29	1 hemz	Possibly benign
			inherited (Fa)	*ANK3*	600465	NM_020987.3	335	48	g.61898845C>T	c.2615G>A	p.Gly872Asp	-	-	-	-		-3.8	-4.4	-16.2	-	Deleterious	deleterious	4.241	1,00	94	1 htz	Benign
APN-36	M	AR		*ACY1*	104620	NM_000666.2	259	49	g.52021045C>T	c.625C>T	p.Arg209Cys	rs140738368	-	-	5	0,04%	0	0	0	-	Deleterious	deleterious	1.497	0,00	180	1 htz	VOUS
				CACNA1G	604065	NM_198396.1	269	46	g.48653642G>A	c.1879G>A	p.Gly627Arg	rs200317339	-	-	4	0,03%	0	0	0	New DSS	Tolerated	neutral	0.932	0.354	125	1 htz	Possibly benign
		AD	inherited (Fa)	*ZBTB20*	606025	NM_015642.4	463	51	g.114057962C>T	c.1897G>A	p.Val633Met	-	-	-	-		0	0	0	-	Tolerated	deleterious	3.514	1,00	21	1 htz	Benign
APN-37	M			*PRODH*	606810	NM_016335.4	284	42	g.18905859G>A	c.1397C>T	p.Thr466Met	rs2870984	**pathogenic**	34	0,26%	0	0	0	0	-	Deleterious	neutral	1.577	1,00	81	1 htz	VOUS
		AR		*SLC46A1*	611672	NM_080669.3	491	51	g.6026T>A	c.512T>A	p.Val171Asp	rs189103810	-	-	-	-	0	0	0	-	Deleterious	deleterious	NA	0,00	152	1 htz	VOUS
				ADRA2B	104260	NM_000682.5	570	46	g.5858G>C	c.858G>C	p.Glu286Asp	-	-	-	-	NA	NA	NA	NA	-	Tolerated	neutral	NA	0,00	45	1 htz	Possibly benign
APN-38	M	AD	de novo	*SLC2A1*	138140	NM_006516.2	209	47	g.43395407G>A	c.724C>T	p.Gln242*	-	-	-	-		0	0	0	-	NA	NA	2.546	1,00	NA	1 htz	Pathogenic
		XL		*MED12*	300188	NM_005120.2	304	99	g.70347021C>T	c.2849+39C>T	p.?	-	-	-	-		0	0	0	New DSS	NA	NA	-0.198	0,00	NA	1 hemz	ossibly benign
APN-39	M			*LAMA1*	150320	NM_005559.2	939	51	g.6965341G>A	c.7141C>T	p.Arg2381Cys	rs142063208	-	-	16	0,12%	0	0	0	-	Deleterious	deleterious	3.353	1,00	180	1 htz	VOUS
		AR		*TMEM135*	NA	NM_022918.3	458	47	g.87017001G>A	c.722G>A	p.Arg241Lys	rs146860039	-	-	17	0,13%	0	0	0	-	Tolerated	deleterious	4.079	1,00	26	1 htz	VOUS
		AD		*MED13L*	608771	NM_015335.4	659	50	g.116446341G>A	c.1877C>T	p.Pro626Leu	rs186297695	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	0.69	0.031	98	1 htz	Possibly benign
APN-40	M	AR		*GAMT/NDUFS7*	601240	NM_000156.4	410	48	g.1397462G>A	c.607C>T	p.Arg203Trp	-	-	-	-		0	0	0	-	Deleterious	neutral	1.013	0.984	101	1 htz	VOUS
		XL		*HUWE1*	300697	NM_031407.4	310	99	g.53600750T>C	c.6272A>G	p.Asn2091Ser	-	-	-	-		0	0	0	New ASS	Tolerated	neutral	1.9	1,00	46	1 hemz	Possibly benign
		AD	de novo	*TCF4*	602272	NM_001083962.1	680	43	g.53017622_53017625del	c.514_517del	p.Lys172Phefs*61	-	-	-	-		0	0	-1	-	NA	NA	NA	NA	NA	1 htz	Pathogenic
APN-41	M			*SYNGAP1*	603384	NM_006772.2	308	36	g.33411686_33411694del	c.3357_3365del	p.Ser1121_Gly1123del	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
		AR		*RABL6*	610615	NM_001173988.1	552	49	g.139734210C>T	c.1826C>T	p.Pro609Leu	-	-	-	1	0,01%	0	0	0	-	Tolerated	deleterious	-0.521	0,00	98	1 htz	VOUS
				HAL	609457	NM_002108.2	523	49	g.96370239C>T	c.1801G>A	p.Glu601Lys	-	-	-	1	0,01%	NA	0	NA	-	Deleterious	neutral	6.016	1,00	56	1 htz	VOUS
		XL	inherited (Mo)	*DMD*	300377	NM_004010.3	275	94	g.31164440del	c.10520del	p.Arg3507Glnfs*27	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 hemz	Pathogenic
		AD	inherited (Mo)	*KIRREL3*	607761	NM_032531.3	519	49	g.126306852G>A	c.1406C>T	p.Thr469Met	-	-	-	1	0,01%	0	0	0	-	Deleterious	deleterious	5.371	1,00	81	1 htz	Benign
APN-42	M			*AP4M1/TAF6*	602296	NM_004722.3	673	50	g.99702938G>A	c.803G>A	p.Arg268Gln	-	-	-	-		0	0	0	-	Deleterious	deleterious	4.967	1,00	43	1 htz	VOUS
		AR		*RELN*	600514	NM_005045.3	724	49	g.103180699C>T	c.6875G>A	p.Arg2292His	-	-	-	-		0	0	0	-	Deleterious	deleterious	5.694	1,00	29	1 htz	VOUS
				WDR62	613583	NM_001083961.1	471	49	g.36595425C>A	c.4159C>A	p.Leu1387Ile	rs147652186	-	-	5	0,04%	0	0	0.3	-	Tolerated	neutral	0.69	0,00	5	1 htz	Possibly benign
		XL	inherited (Mo)	*SLC9A6*	300231	NM_001042537.1	84	90	g.135080258_135080262del	c.526-9_526-5del	p.?	-	-	-	-		-32	-10.4	-77.8	-	NA	NA	NA	NA	NA	1 hemz	Pathogenic
				AP4M1/MCM7	602296	NM_004722.3	147	39	g.99701053A>G	c.373A>G	p.Thr125Ala	-	-	-	-		0	0	0	-	Deleterious	deleterious	3.837	1,00	58	1 htz	VOUS
				TTI2	614426	NM_025115.3	209	48	g.33361016C>T	c.1190G>A	p.Gly397Glu	rs150984360	-	-	14	0,11%	0	0	0	-	Tolerated	neutral	1.255	0.992	98	2 htz	Possibly benign
APN-43	M	AR		*CACNA1G*	604065	NM_198397.1	153	56	g.48683415G>C	c.4453G>C	p.Val1485Leu	rs59226503	-	-	49	0,38%	0	0	0	-	Tolerated	neutral	0.044	0,00	32	1 htz	Possibly benign
				LAMC3	604349	NM_006059.3	36	38	g.133884640_133884648dup	c.39_47dup	p.Pro15_Ala17dup	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
				RALGDS	601619	NM_001042368.1	193	46	g.135983956T>C	c.613+104A>G	p.?	rs111236084	-	-	-	-	0	0	0	New DSS	NA	NA	-0.36	0,00	NA	2 htz	VOUS
		AD	de novo	*RAI1*	607642	NM_030665.3	212	39	g.17698594_17698598del	c.2332_2336del	p.Gly778Glnfs*7	-	-	-	-		0	0	0	-	NA	NA	NA	NA	NA	1 htz	Pathogenic
			inherited (Mo)	*KIRREL3*	607761	NM_032531.3	200	46	g.126319042G>A	c.859C>T	p.Arg287Trp	-	-	-	-		0	0	-8.2		Deleterious	deleterious	0.367	0.969	101	1 htz	Benign
APN-46	M			*ELP2*	NA	NM_018255.1	336	50	g.33722872A>G	c.739A>G	p.Thr247Ala	-	-	-	-		0	0	0	-	Tolerated	neutral	1.255	0.969	58	1 htz	Possibly benign
		AR		*ELP2*	NA	NM_018255.1	321	48	g.33734736G>C	c.1126-77G>C	p.?	-	-	-	-		0	0	0	Cryptic ASS	NA	NA	-2.7	0,00	NA	1 htz	VOUS
				AP4B1	607245	NM_006594.2	292	53	g.114442885A>G	c.755T>C	p.Val252Ala	rs141417436	-	-	25	0,19%	0	0	0	-	Deleterious	neutral	2.465	1,00	64	2 htz	VOUS
				PMM2	601785	NM_000303.2	354	44	g.8904982A>G	c.394A>G	p.Ile132Val	-	-	-	-		0	0	0	-	Deleterious	neutral	-0.037	0.016	29	1 htz	VOUS
		AD		*SHANK1*	604999	NM_016148.2	203	47	g.51169797G>A	c.5420C>T	p.Pro1807Leu	rs184259943	-	-	-	-	0	0	0	-	Tolerated	neutral	0.932	0,00	98	2 htz	Possibly benign
APN-47	M	AR		*AP4B1/DCLRE1B*	607245	NM_006594.2	194	45	g.114447327C>T	c.13G>A	p.Gly5Ser	-	-	-	1	0,01%	0	0	0	-	Deleterious	deleterious	5.451	1,00	56	1 htz	VOUS
				MAN1B1	604346	NM_016219.3	320	50	g.139982611A>C	c.304A>C	p.Ile102Leu	rs149322865	-	-	11	0,08%	0	0	0	-	Tolerated	neutral	0.044	0.11	5	1 htz	Possibly benign
APN-48	M	AR		*CC2D1A*	610055	NM_017721.4	199	47	g.14024308C>T	c.605C>T	p.Ala202Val	-	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	0.125	0,00	64	1 htz	Possibly benign
		AD		*PGA5*	169730	NM_014224.2	23	60	g.61013499G>A	c.538G>A	p.Asp180Asn	-	-	-	-		0	0	0	-	Deleterious	deleterious	3.918	0.984	23	2 htz	VOUS
			inherited (Mo)	*NRXN2*	600566	NM_138734.2	79	45	g.64410248C>G	c.28G>C	p.Gly10Arg	-	-	-	-		0	0	0	-	Deleterious	neutral	0.205	1,00	125	1 htz	Benign
APN-49	M			*ACY1*	104620	NM_000666.2	318	50	g.52022837C>T	c.1057C>T	p.Arg353Cys	rs121912698	**pathogenic**	33	0,25%	0	0	8.4	-	Deleterious	deleterious	2.465	0.961	180	1 htz	VOUS
		AR		*SLC46A1*	611672	NM_080669.3	283	48	g.11542C>T	c.1366C>T	p.Pro456Ser	rs186143284	-	-	-	-	0	0	0	-	Tolerated	neutral	NA	0,00	74	1 htz	Possibly benign
		AD	inherited (Fa)	*SHANK1*	604999	NM_016148.2	129	51	g.51170463G>A	c.4754C>T	p.Thr1585Met	-	-	-	-		0	0	0	-	Deleterious	deleterious	0.205	0.992	81	1 htz	Benign
APN-50	M	AR		*MTHFR*	607093	NM_005957.4	187	49	g.11852412G>A	c.1555C>T	p.Arg519Cys	rs45496998	-	-	33	0,25%	0	0	0	-	Tolerated	deleterious	3.03	1,00	180	1 htz	VOUS
		AD		*DOCK8*	611432	NM_203447.3	170	38	g.231494C>T	c.5296C>T	p.Arg1766Trp	rs201776049	-	-	1	-	0	0	0	-	Deleterious	deleterious	NA	1,00	101	1 htz	Benign
APN-45	M	AR		*WDR62/THAP8*	613583	NM_001083961.1	116	41	g.36545955C>T	c.82C>T	p.Arg28Trp	rs200283315	-	-	-	-	0	0	0	-	Deleterious	deleterious	0.932	1,00	101	2 htz	VOUS
APN-51	M																												
APN-53	M	XL		*PHF8*	300560	NM_015107.2	89	98	g.54012358T>C	c.2020A>G	p.Met674Val	-	-	-	-		0	0	0	-	Tolerated	neutral	0.205	0.992	21	1 hemz	Possibly benign
		AD		*PACS1*	607492	NM_018026.2	207	48	g.66000459G>A	c.1760G>A	p.Arg587Gln	rs201700614	-	-	2	0,02%	0	0	0	-	Tolerated	neutral	1.335	1,00	43	1 htz	Benign
		XL		*CCDC22*	300859	NM_014008.3	51	100	g.49103368G>C	c.891G>C	p.Glu297Asp	-	-	-	-		0	0	0	-	Tolerated	neutral	4.644	1,00	45	1 hemz	Possibly benign
		AD	inherited (Fa)	*NRXN2*	600566	NM_015080.3	38	39	g.64481101G>A	c.71C>T	p.Ala24Val	-	-	-	-		0	0	0	New DSS	Deleterious	neutral	0.044	0.102	64	1 htz	Benign
APN-54	M			*ASPM*	605481	NM_018136.4	158	46	g.197091341A>G	c.3689T>C	p.Ile1230Thr	-	-	-	-		0	0	0	-	Deleterious	deleterious	4.725	1,00	89	1 htz	VOUS
		AR		*CDK5RAP2*	608201	NM_018249.4	118	56	g.123287277G>A	c.1079C>T	p.Thr360Ile	rs145165171	-	-	13	0,10%	0	0	0	-	Tolerated	neutral	0.77	0,00	89	1 htz	Possibly benign
				RELN	600514	NM_005045.3	129	55	g.103175844C>T	c.7268G>A	p.Arg2423His	rs140660860	-	-	5	0,04%	0	0	0	-	Deleterious	neutral	1.174	0.866	29	1 htz	VOUS
				FTCD	606806	NM_006657.2	101	42	g.47570220C>A	c.775-56G>T	p.?	rs186916919	-	-	-	-	0	0	0	Cryptic ASS	NA	NA	-0.36	0,00	NA	1 htz	VOUS
		XL		*IQSEC2*	300522	NM_001111125.1	29	96	g.53263460C>T	c.4408G>A	p.Ala1470Thr	rs191886831	-	-	-	-	0	0	0	-	Tolerated	NA	0.125	0.425	58	1 hemz	Possibly benign
				NSUN2	610916	NM_017755.5	147	54	g.6600149C>T	c.2194G>A	p.Val732Met	rs143551642	-	-	6	0,05%	0	0	0	-	Tolerated	neutral	-0.117	0,00	21	1 htz	Possibly benign
APN-56	M	AR		*FTCD*	606806	NM_006657.2	65	47	g.47565840dup	c.990dup	p.Pro331Alafs*2	rs35208133	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	2 htz	VOUS
				LAMA1	150320	NM_005559.2	128	55	g.7044753G>A	c.944C>T	p.Pro315Leu	rs201386303	-	-	1	0,01%	0	0	0	-	Deleterious	deleterious	5.774	1,00	98	1 htz	VOUS
		AR		*ANK3*	600465	NM_020987.3	111	45	g.61846461T>C	c.3722A>G	p.Asn1241Ser	-	-	-	-		0	0	0	-	Tolerated	neutral	1.497	0.969	46	1 htz	Possibly benign
APN-57	M			*CDK5RAP2*	608201	NM_018249.4	97	49	g.123165239G>C	c.5152C>G	p.Leu1718Val	rs141004029	-	-	9	0,07%	0	0	0	-	Deleterious	neutral	-0.44	0.134	32	1 htz	VOUS
		?		*DIP2B*	611379	NM_173602.2	81	54	g.51080389C>A	c.1475C>A	p.Ser492Tyr	rs148830732	-	-	5	0,04%	0	0	0	-	Deleterious	neutral	3.918	1,00	144	1 htz	VOUS
		AD	de novo	*DYRK1A*	600855	NM_001396.3	54	42	g.38858865C>T	c.613C>T	p.Arg205*	-	-	-	-		0	0	0	-	NA	NA	4.483	1,00	NA	1 htz	Pathogenic
APN-58	M	AR		*UROC1*	613012	NM_001165974.1	33	48	g.126229555A>G	c.209T>C	p.Leu70Pro	rs137852796	**pathogenic**	-	-	-	0	0	0	-	Tolerated	neutral	0.286	0.992	98	1 htz	VOUS
				AP4S1	607243	NM_007077.3	64	45	g.31535431A>G	c.29A>G	p.Lys10Arg	rs147135554	-	-	9	0,07%	0	0	0	-	Tolerated	neutral	1.82	1,00	26	1 htz	Possibly benign
		AD		*ANKRD11*	611192	NM_001256182.1	88	40	g.89346906T>G	c.6044A>C	p.Tyr2015Ser	-	-	-	-		0	0	0	-	Deleterious	NA	1.174	0.992	144	1 htz	Possibly benign
APN-60	M	AR		*MCPH1*	607117	NM_024596.3	113	42	g.6338338C>T	c.2077C>T	p.Arg693Cys	-	-	-	4	0,03%	0	0	0	-	Deleterious	deleterious	4.079	0.992	180	1 htz	VOUS

		AR		*WDR62/THAP8*	613583	NM_001083961.1	71	47	g.36545955C>T	c.82C>T	p.Arg28Trp	rs200283315	-	-	-	0	0	0	-	Deleterious	deleterious	0.932	1,00	101	2 htz	VOUs
		AD	de novo	*GRIN1*	138249	NM_007327.3	108	51	g.140056661C>G	c.1670C>G	p.Pro557Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.483	1,00	103	1 htz	Pathogenic
APN-63	M			*CDK5RAP2*	608201	NM_018249.4	161	39	g.123210388C>T	c.2810G>A	p.Arg937His	rs200811652	-	-	-	0	0	-0.5	-	Tolerated	deleterious	3.111	0.992	29	1 htz	VOUS
		AR		*KDM5A*	180202	NM_001042603.1	145	42	g.427530A>G	c.2639T>C	p.Met880Thr	-	-	1	0,01%	0	0	-100	-	Tolerated	neutral	3.272	1,00	81	1 htz	Possibly benign
				TUSC3	601385	NM_006765.3	117	53	g.15397977C>T	c.38C>T	p.Ala13Val	rs200808372	-	4	0,03%	0	0	0	-	Deleterious	neutral	0.69	0.976	64	2 htz	VOUS
		XL		*OPHN1*	300127	NM_002547.2	52	100	g.67414426C>T	c.1105-86G>A	p.?	rs145196238	-	-	-	0	0	0	New ASS	NA	NA	-0.602	0,00	NA	1 hemz	VOUS
APN-65	M	AD	inherited (Fa)	*DOCK8*	611432	NM_203447.3	161	42	g.186973G>A	c.3023G>A	p.Arg1008Gln	rs145844320	-	-	-	0	0	0	-	Deleterious	deleterious	NA	1,00	43	1 htz	Benign
			inherited (Fa)	*DOCK8*	611432	NM_203447.3	164	42	g.195131G>C	c.3312G>C	p.Glu1104Asp	rs138810908	-	-	-	0	0	0	-	Deleterious	deleterious	NA	0.827	45	1 htz	Benign
		XL	de novo	*IQSEC2*	300522	NM_001111125.1	106	99	g.53268395G>A	c.3097C>T	p.Gln1033*	-	-	-	-	0	0	0	-	NA	NA	5.613	1,00	NA	1 hemz	Pathogenic
APN-68	M	AR		*C12orf57/PTPN6*	615140	NM_138425.2	153	48	g.7053770C>T	c.184C>T	p.Gln62*	-	-	-	-	0	0	0	-	NA	NA	5.048	1,00	NA	1 htz	VOUS
		AD		*RAI1*	607642	NM_030665.3	289	45	g.17697991G>A	c.1729G>A	p.Gly577Ser	rs141458634	-	-	-	0	0	0	-	Tolerated	neutral	-0.037	0.992	56	1 htz	Possibly benign
				CRBN	609262	NM_016302.3	364	53	g.3194210G>A	c.1078C>T	p.Leu360Phe	-	-	-	-	0	0	0	-	Tolerated	deleterious	6.259	1,00	22	1 htz	VOUS
APN-69	M			*ASPM*	605481	NM_018136.4	381	51	g.197111913C>T	c.1469G>A	p.Arg490His	-	-	-	-	0	0	0	-	Tolerated	deleterious	1.174	0.976	29	1 htz	VOUS
		AR		*UPB1*	606673	NM_016327.2	163	52	g.24896195G>T	c.225G>T	p.=	rs150740488	-	1	0,01%	0	0	0	Cryptic ASS	NA	NA	0.69	1,00	NA	1 htz	VOUS
		XL	inherited (Mo)	*PQBP1*	300463	NM_005710.2	100	99	g.48760294C>T	c.731C>T	p.Pro244Leu	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.129	0.992	98	1 hemz	Possibly pathogenic
		AD		*DEAF1*	602635	NM_021008.2	170	45	g.678739C>T	c.1210G>A	p.Ala404Thr	-	-	-	-	0	0	0	-	Tolerated	neutral	0.286	0.268	58	1 htz	Possibly benign
APN-70	M			*YY1*	600013	NM_003403.3	70	41	g.100705806_100705814del	c.225_233del	p.His78_His80del	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	Possibly benign
		AR		*RELN*	600514	NM_005045.3	242	45	g.103193992C>T	c.5988G>A	p.Met1996Ile	-	-	-	-	0	0	92.7	-	Deleterious	deleterious	5.774	1,00	10	1 htz	VOUS
				ASPM	605481	NM_018136.4	245	46	g.197071470T>C	c.6911A>G	p.His2304Arg	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.36	0,00	29	1 htz	Possibly benign
APN-72	M	AD	inherited (Fa)	*CIC*	612082	NM_015125.3	402	50	g.42798112G>A	c.4066G>A	p.Glu1356Lys	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.209	1,00	56	1 htz	Benign
		AR		*RABL6*	610615	NM_001173988.1	342	49	g.139734218C>T	c.1834C>T	p.Pro612Ser	-	-	-	-	0	0	0	-	Tolerated	deleterious	0.69	0,00	74	1 htz	VOUS
		AD		*EHMT1*	607001	NM_024757.4	346	47	g.140611519C>T	c.527C>T	p.Pro176Leu	-	-	-	-	0	0	0	-	Deleterious	neutral	4.402	0.512	98	1 htz	VOUS
APN-73	M			*SHANK1*	604999	NM_016148.2	299	47	g.51169797G>A	c.5420C>T	p.Pro1807Leu	rs184259943	-	-	-	0	0	0	Cryptic ASS	Tolerated	neutral	0.932	0,00	98	2 htz	Possibly benign
		AR		*PRODH*	606810	NM_016335.4	4	25	g.18923839_18923841del	c.-40-1_-39del	p.?	-	-	-	-	NA	-100	-100	-	NA	NA	NA	NA	NA	1 htz	VOUS
APN-74	M	XL	inherited (Mo)	*MECP2*	300005	NM_001110792.1	4	100	g.153363068C>T	c.55G>A	p.Glu19Lys	-	-	-	-	0	0	0	-	Deleterious	neutral	2.223	1,00	56	1 hemz	Benign
				PHF8	300560	NM_015107.2	87	97	g.54013472C>T	c.1995+39G>A	p.?	-	-	5	0,05%	0	0	0	New ASS	NA	NA	0.528	0.843	NA	1 hemz	Possibly benign
		AD	inherited (Fa)	*EHMT1*	607001	NM_024757.4	329	51	g.140611260C>T	c.268C>T	p.Arg90Trp	-	-	-	-	0	0	0	-	Deleterious	deleterious	0.448	0.055	101	1 htz	Benign
			inherited (Fa)	*RELN*	600514	NM_005045.3	394	48	g.103162588G>A	c.7549C>T	p.Gln2517*	-	-	-	-	0	0	0	-	NA	NA	5.774	1,00	NA	1 htz	Benign
APN-76	M		inherited (Fa)	*RELN*	600514	NM_005045.3	463	47	g.103417058G>A	c.490C>T	p.Arg164Trp	-	-	-	-	0	0	-0.1	-	Deleterious	deleterious	1.093	1,00	101	1 htz	Benign
		AR		*CACNA1G*	604065	NM_198385.1	354	48	g.48687255A>T	c.4718A>T	p.Asp1573Val	-	-	-	-	0	0	-0.1	-	Deleterious	deleterious	2.223	1,00	152	1 htz	VOUS
				SOBP	613667	NM_018013.3	479	47	g.107954923T>G	c.875T>G	p.Val292Gly	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.272	1,00	109	1 htz	VOUS
		?		*DIP2B*	611379	NM_173602.2	237	45	g.51102417G>T	c.2649+72G>T	p.?	-	-	-	-	0	0	0	Cryptic ASS	NA	NA	-0.602	0,00	NA	1 htz	VOUS
APN-77	F	XL	inherited (Mo)	*BCOR*	300485	NM_001123385.1	137	47	g.39921622G>A	c.4198C>T	p.Arg1400Trp	-	-	-	-	0	0	0	-	Deleterious	deleterious	2.062	1,00	101	1 htz	VOUS
		AD		*RAI1*	607642	NM_030665.3	243	41	g.17700043_17700045del	c.3781_3783del	p.Glu1261del	rs149716029	-	-	-	0	0	0	-	NA	NA	NA	NA	2 htz	Possibly benign
		AD		*DOCK8*	611432	NM_203447.3	280	45	g.94782C>T	c.470C>T	p.Thr157Met	-	-	-	-	0	0	0	-	Deleterious	neutral	NA	0.945	81	1 htz	VOUS
				LAMA1	150320	NM_005559.2	172	54	g.7037576C>A	c.1737+1G>T	p.?	-	-	-	-	-100	-100	-100	-	NA	NA	5.451	1,00	NA	1 htz	VOUS
APN-80	M			*CACNA1G*	604065	NM_198396.1	245	47	g.48703802T>C	c.6722T>C	p.Leu2241Pro	-	-	2	0,02%	0	0	0	-	Deleterious	deleterious	2.142	0.992	98	1 htz	VOUS
		AR		*PARP1*	173870	NM_001618.3	196	54	g.226570772G>A	c.1124C>T	p.Ser375Leu	-	-	-	-	0	0	0	-	Tolerated	neutral	1.497	0,00	145	1 htz	Possibly benign
				NRXN1	600565	NM_001135659.1	285	50	g.50733596T>C	c.2617+37A>G	p.?	rs202221361	-	5	0,04%	0	0	0	New ASS	NA	NA	0.205	0,00	NA	1 htz	VOUS
APN-81	M	AR		*SRD5A3*	611715	NM_024592.4	208	50	g.56212579T>A	c.76T>A	p.Phe26Ile	-	-	-	-	0	0	0	-	Tolerated	deleterious	2.304	1,00	21	1 htz	VOUS
		XL	inherited (Mo)	*IL1RAPL1*	300206	NM_014271.3	137	86	g.29935696_29935705del	c.894_903del	p.Trp299Thrfs*18	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 hemz	Pathogenic
APN-82	M	AD		*KIF1A*	601255	NM_004321.5	248	48	g.241697865G>A	c.2467C>T	p.Pro823Ser	-	-	-	-	0	0	0	-	Tolerated	deleterious	4.16	1,00	74	1 htz	VOUS
		AR		*L1CAM*	308840	NM_000425.3	89	100	g.153137798G>A	c.209C>T	p.Thr70Met	-	-	-	-	0	0	-0.1	-	Deleterious	neutral	0.448	0.307	81	1 hemz	VOUS
				PRODH	606810	NM_016335.4	180	51	g.18904466T>C	c.1463A>G	p.Asn488Ser	rs139903009	-	24	0,18%	0	0	0	-	Tolerated	neutral	0.851	0.677	46	2 htz	Possibly benign
		AD	inherited (Fa)	*GRIN2A*	138253	NM_001134407.1	669	49	g.10032128G>A	c.695C>T	p.Ser232Phe	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.774	1,00	155	1 htz	Benign
APN-84	M			*TRAPPC9*	611966	NM_031466.5	360	46	g.141461436G>A	c.331C>T	p.His111Tyr	-	-	-	-	0	0	0	-	Tolerated	deleterious	4.241	1,00	83	1 htz	VOUS
		AR		*NRXN1*	600565	NM_001135659.1	378	53	g.51254660C>A	c.752G>T	p.Arg251Leu	-	-	-	-	0	0	0	Cryptic ASS	NA	neutral	2.869	0.992	102	1 htz	VOUS
		AD		*YY1*	600013	NM_003403.3	297	44	g.100705875C>T	c.294C>T	p.=	-	-	-	-	0	0	0	Cryptic ASS	NA	NA	1.013	1,00	NA	1 htz	VOUS
				CNTNAP2	604569	NM_014141.5	275	51	g.145814041G>A	c.73G>A	p.Ala25Thr	rs200866893	-	8	0,06%	0	0	0	-	Deleterious	neutral	2.627	1,00	58	1 htz	VOUS
APN-83	M			*RABL6*	610615	NM_001173988.1	480	45	g.139732024G>A	c.1039G>A	p.Ala347Thr	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.198	0,00	58	1 htz	Possibly benign
		AR		*UPB1*	606673	NM_016327.2	222	45	g.24896113C>G	c.143C>G	p.Ser48Cys	rs145989498	-	6	0,05%	0	0	0	-	Tolerated	neutral	0.448	0,00	112	2 htz	Possibly benign
		XL	inherited (Mo)	*SHROOM4*	300579	NM_020717.3	166	98	g.50345803G>A	c.3772C>T	p.Gln1258*	-	-	-	-	0	0	-0.9	-	NA	NA	4.079	1,00	NA	1 hemz	VOUS
APN-86	M	AD		*DYRK1A*	600855	NM_001396.3	398	50	g.38884589C>T	c.2047C>T	p.Arg683Cys	rs201554841	-	-	-	0	0	0	-	Deleterious	neutral	3.999	1,00	180	1 htz	VOUS
		AR		*CACNA1G*	604065	NM_198384.1	246	42	g.48653234G>A	c.1471G>A	p.Val491Ile	rs201788352	-	4	0,03%	0	0	0	-	Tolerated	deleterious	1.335	0.866	29	1 htz	VOUS
		XL		*CACNA1F*	300110	NM_005183.2	201	99	g.49061743G>C	c.5788C>G	p.Arg1930Gly	-	-	-	-	0	0	0	-	Tolerated	neutral	1.255	1,00	125	1 hemz	Possibly benign
		AD	de novo	*DYRK1A*	600855	NM_001396.3	367	41	g.38858873_38858876delinsGAA	c.621_624delinsGAA	p.Glu208Asnfs*3	-	-	-	-	0	0	0	-	NA	NA	NA	NA	1 htz	Pathogenic
			inherited (Fa)	*EPB41L1*	602879	NM_012156.2	337	48	g.34766583A>G	c.479A>G	p.Lys160Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.353	1,00	26	1 htz	Benign
				CC2D1A	610055	NM_017721.4	408	50	g.14037530C>A	c.2065C>A	p.Pro689Thr	rs200663646	-	-	-	0	0	0	-	Tolerated	deleterious	4.967	1,00	38	1 htz	VOUS
APN-87	M			*SARS*	607529	NM_006513.3	367	49	g.109778021C>T	c.937C>T	p.Arg313Cys	-	-	-	-	0	0	0	-	Deleterious	deleterious	2.304	1,00	180	1 htz	VOUS
				STIL	181590	NM_001048166.1	387	49	g.47726025G>C	c.3016C>G	p.Leu1006Val	-	-	-	-	0	0	0	-	Deleterious	neutral	2.707	1,00	32	1 htz	VOUS
		AR		*PRSS12*	606709	NM_003619.3	241	41	g.119273436G>A	c.440C>T	p.Pro147Leu	rs72903215	-	23	0,18%	0	0	0	-	Deleterious	deleterious	2.385	1,00	98	1 htz	VOUS
				RELN	600514	NM_005045.3	389	51	g.103205873G>C	c.5062C>G	p.Leu1688Val	-	-	-	-	0	0	0	-	Tolerated	neutral	0.851	0.992	32	1 htz	Possibly benign
				RELN	600514	NM_005045.3	312	38	g.103132368T>C	c.9443+32A>G	p.?	-	-	-	-	0	0	0	New ASS	NA	NA	-0.198	0,00	NA	2 htz	VOUS
		XL	inherited (Mo)	*NLGN3*	300336	NM_181303.1	376	99	g.70389249C>T	c.1849C>T	p.Arg617Trp	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.837	1,00	101	1 hemz	Possibly pathogenic
APN-99	M	AD		*SHANK3*	606230	NM_001080420.1	4	50	g.51136080G>A	c.1483G>A	p.Val495Met	-	-	-	-	0	0	0	-	Tolerated	neutral	0.609	0.142	21	1 htz	Possibly benign
		AR		*MAN1B1*	604346	NM_016219.3	248	45	g.139981510T>G	c.59T>G	p.Leu20Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	0.851	0.992	102	1 htz	VOUS
				ASPM	605481	NM_018136.4	861	48	g.197111601G>C	c.1781C>G	p.Thr594Arg	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.521	0,00	71	1 htz	Possibly benign
		AD		*SHANK1*	604999	NM_016148.2	14	43	g.51171571T>G	c.3646A>C	p.Thr1216Pro	-	-	-	-	0	0	0	-	Tolerated	neutral	0.205	0.992	38	1 htz	Possibly benign
				ANK3	600465	NM_020987.3	84	54	g.61994624T>C	c.799-80A>G	p.?	rs138975531	-	-	-	0	0	0	New DSS	NA	NA	-0.521	0,00	NA	1 htz	VOUS
APN-100	M			*TRAPPC9*	611966	NM_031466.5	119	47	g.141370271G>A	c.1667C>T	p.Ala556Val	rs147214217	-	1	0,01%	0	0	NA	-	Deleterious	deleterious	6.178	1,00	64	1 htz	VOUS
		AR		*KDM5A*	180202	NM_001042603.1	243	46	g.461384T>C	c.1136A>G	p.Asn379Ser	rs201675393	-	11	0,09%	0	0	0	New ASS	Tolerated	neutral	4.644	1,00	46	1 htz	Possibly benign
				UPB1	606673	NM_016327.2	88	52	g.24891397T>C	c.26T>C	p.Leu9Pro	-	-	-	-	0	0	0	-	Deleterious	neutral	3.111	1,00	98	1 htz	VOUS
				PRSS12	606709	NM_003619.3	60	52	g.119273839C>T	c.37G>A	p.Gly13Arg	rs140334007	-	8	0,06%	0	0	0	-	Tolerated	neutral	0.851	0.008	125	1 htz	Possibly benign
				DEAF1	602635	NM_021008.2	472	44	g.674690G>A	c.1349C>T	p.Pro450Leu	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.763	0.268	98	1 htz	Possibly benign
		AD	Absent from Mo	*TCF4*	602272	NM_001083962.1	518	48	g.52899907C>T	c.1487-5G>A	p.?	-	-	-	-	-11.6	-100	0.3	New ASS	NA	NA	0.286	0,00	NA	1 htz	Possibly pathogenic
APN-101	M			*MED13L*	608771	NM_015335.4	705	49	g.116434379T>C	c.2898A>G	p.Ile966Met	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.521	0.819	10	1 htz	Possibly benign
				STIL	181590	NM_001048166.1	1003	52	g.47717077T>C	c.3598A>G	p.Thr1200Ala	rs200980713	-	4	0,03%	0	0	0	-	Deleterious	deleterious	3.676	1,00	58	1 htz	VOUS
		AR		*LAMA1*	150320	NM_005559.2	797	49	g.6976021G>A	c.6404C>T	p.Ser2135Phe	-	-	-	-	0	0	0	-	Tolerated	deleterious	1.416	0.22	155	1 htz	VOUS
		XL		*MECP2*	300005	NM_004992.3	118	100	g.153296095C>G	c.1184G>C	p.Ser395Thr	-	-	-	-	0	0	0	-	Tolerated	neutral	1.658	1,00	58	1 hemz	Possibly benign
		AD		*AFF3*	601464	NM_002285.2	560	49	g.100170859G>A	c.3473C>T	p.Ala1158Val	-	-	-	-	0	0	0	-	Tolerated	deleterious	6.097	1,00	64	1 htz	VOUS
				EPB41L1	602879	NM_012156.2	103	58	g.34782048C>T	c.1301-86C>T	p.?	rs139014428	-	-	-	0	0	0	New DSS	NA	NA	0.125	0,00	NA	2 htz	VOUS
APN-102	M			*MCPH1*	607117	NM_024596.3	627	44	g.6302738G>A	c.1495G>A	p.Val499Met	rs146586991	-	34	0,28%	0	0	0	-	Tolerated	deleterious	-5.041	0,00	21	1 htz	VOUS
		AR		*CDK5RAP2*	608201	NM_018249.4	574	46	g.123205924C>T	c.3122G>A	p.Ser1041Asn	-	-	-	-	0	0	0	-	Tolerated	neutral	-1.57	0,00	46	1 htz	Possibly benign
		?		*CYFIP1*	606322	NM_001033028.1	565	49	g.22956276T>C	c.220T>C	p.Trp74Arg	-	-	-	-	0	0	0	-	Deleterious	neutral	-2.054	0,00	101	1 htz	VOUS
		XL		*KDM5C*	314690	NM_004187.3	148	99	g.53246446C>T	c.536G>A	p.Arg179His	rs201805773	-	3 (1M)	0,03%	0	0	9.5	-	Tolerated	neutral	1.739	1,00	29	1 hemz	Possibly benign
APN-103	M	AR		*CDK5RAP2*	608201	NM_018249.4	590	46	g.123215872G>C	c.2655C>G	p.Phe885Leu	rs112600265	-	38	0,29%	0	0	0	-	Tolerated	neutral	1.255	0,00	22	1 htz	Possibly benign
		XL	inherited (Mo)	*PHF8*	300560	NM_015107.2	366	100	g.54028583C>G	c.1141+5G>C	p.?	-	-	-	-	-41.3	-17.4	-98.8	-	NA	NA	0.851	0.606	NA	1 hemz	Pathogenic
		AD	de novo	*DOCK8*	NA	NM_203447.3	746	55	g.197171G>T	c.3496G>T	p.Glu1166*	-	-	-	-	0	0	0	-	NA	NA	NA	1,00	NA	1 htz	VOUS
				FTCD	606806	NM_006657.2	45	56	g.47565840dup	c.990dup	p.Pro331Alafs*2	rs35208133	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	2 htz	VOUS
APN-105	M	AR		*LAMC3*	604349	NM_006059.3	251	41	g.133967094C>G	c.4648C>G	p.Leu1550Val	-	-	-	-	0	0	0	-	Deleterious	deleterious	1.577	0.858	32	1 htz	VOUS
				CENP1	609279	NM_018451.3	578	49	g.25481285A>C	c.1021T>G	p.Tyr341Asp	rs143258862	-	33	0,25%	0	0	0	-	Deleterious	deleterious	1.416	1,00	160	2 htz	VOUS
		AD	inherited (Mo)	*ARID1B*	NA	NM_017519.2	788	52	g.157527645G>C	c.5331G>C	p.Lys1777Asn	-	-	-	-	0	0	0	-	Deleterious	deleterious	2.062	0.984	94	1 htz	Benign

APN-104	M	AD		*PARP1*	173870	NM_001618.3	674	47	g.226550829T>C	c.2819A>G	p.Lys940Arg	rs3219145	-	13	0,10%	0	0	0	-	Tolerated	deleterious	5.129	1,00	26	1 htz	VOUS
		AR		*ADRA2B*	NA	NM_000682.5	544	46	g.6055T>C	c.1055T>C	p.Ile352Thr	-	-	-	-	NA	NA	NA	-	NA	neutral	NA	0,00	89	1 htz	Possibly benign
APN-106	M	AD		*AFF3*	601464	NM_002285.2	981	49	g.100194796C>T	c.2911G>A	p.Asp971Asn	-	-	-	-	0	0	0	-	Tolerated	deleterious	2.304	1,00	23	1 htz	VOUS
				KIF1A	NA	NM_001244008.1	823	49	g.241710418C>T	c.1311G>A	p.=	-	-	-	-	0	0	0	New ASS	NA	NA	-1.005	0.472	NA	1 htz	VOUS
		AR		*CENP1*	609279	NM_018451.3	1022	50	g.25481285A>C	c.1021T>G	p.Tyr341Asp	rs143258862	-	33	0,25%	0	0	0	-	Deleterious	deleterious	1.416	1,00	160	2 htz	Possibly damaging
APN-107	M	AD	inherited (Fa)	*KCNQ2*	602235	NM_172107.2	678	51	g.62039761C>T	c.1887+5G>A	p.?	-	-	-	-	-63.5	-15.3	-69.6	-	NA	NA	2.788	0.26	NA	1 htz	Benign (does not
			inherited (Mo)	*EHMT1*	607001	NM_024757.4	654	45	g.140674130G>A	c.2236G>A	p.Ala746Thr	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.241	0.992	58	1 htz	Benign
				ASPM	605481	NM_018136.4	1054	47	g.197060113T>C	c.9503A>G	p.Lys3168Arg	-	-	-	-	0	0	0	-	Deleterious	neutral	0.286	0,00	26	1 htz	VOUS
		AR		*ASPM*	605481	NM_018136.4	1403	49	g.197071579G>C	c.6802C>G	p.Leu2268Val	-	-	-	-	0	0	0	-	Tolerated	neutral	-0.924	0,00	32	1 htz	Possibly benign
APN-108	M																												
APN-109	M	AD	inherited (Mo)	*DEAF1*	602635	NM_021008.2	697	48	g.691601G>C	c.290-3C>G	p.?	-	-	-	-	-100	-12.1	-99.8	-	NA	NA	2.546	1,00	NA	1 htz	VOUS
				NSUN2	610916	NM_017755.5	1223	50	g.6605449C>A	c.1674G>T	p.Arg558Ser	-	-	-	-	0	0	0	-	Deleterious	deleterious	0.448	1,00	110	1 htz	VOUS
		AR		*CRBN*	609262	NM_016302.3	1146	46	g.3195735C>A	c.860G>T	p.Cys287Phe	-	-	-	-	0	0	0	-	Tolerated	deleterious	5.613	1,00	205	1 htz	VOUS
APN-110	M	XL	inherited (Mo)	*SLC16A2*	300095	NM_006517.3	422	100	g.73749067T>C	c.1412T>C	p.Leu471Pro	rs122456132	**pathogenic**	-	-	0	0	0.9	-	Deleterious	deleterious	4.725	1,00	98	1 hemz	Pathogenic
		AR		*RELN*	600514	NM_005045.3	664	47	g.103124188C>T	c.10093G>A	p.Val3365Ile	rs115035120	-	8	0,06%	0	0	0	-	Deleterious	deleterious	2.062	0.984	29	1 htz	VOUS
				RABL6	NA	NM_024718.4	1358	50	g.139734176G>A	c.1789G>A	p.Asp597Asn	rs141843004	-	-	-	0	0	0	Cryptic DSS	Tolerated	deleterious	4.322	0.913	23	1 htz	VOUS
APN-111	M	AR		*UPB1*	606673	NM_016327.2	342	45	g.24919586G>A	c.917-1G>A	p.?	rs143493067	-	28	0,22%	-100	-100	-100	-	NA	NA	5.855	1,00	NA	1 htz	Possibly benign
APN-112	M	AD		*CICPAFAH1B3*	612082	NM_015125.3	91	48	g.42799299T>C	c.4783T>C	p.Ser1595Pro	-	-	-	-	0	0	0	-	Deleterious	neutral	2.627	0.102	74	1 htz	VOUS
		AR		*LAMA1*	150320	NM_005559.2	998	49	g.7023207G>A	c.2657C>T	p.Ala886Val	rs144738522	-	34	0,26%	0	0	0	-	Tolerated	deleterious	1.981	0.953	64	1 htz	VOUS
				AP4M1/TAF6	602296	NM_004722.3	367	51	g.9970408C>T	c.1084C>T	p.Arg362Cys	-	-	-	-	0	0	0	-	Deleterious	neutral	1.9	0.331	180	1 htz	VOUS
APN-113	M	XL	inherited (Mo)	*HCFC1*	300019	NM_005334.2	439	99	g.153220153G>A	c.218C>T	p.Ala73Val	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.451	1,00	64	1 hemz	Pathogenic
			inherited (Mo)	*ATRX*	300032	NM_000489.3	826	100	g.76939735G>C	c.1013C>G	p.Ser338Cys	-	-	-	-	0	0	0	-	Deleterious	deleterious	1.981	0.425	112	1 hemz	Possibly pathogenic
		AR		*FTCD*	606806	NM_006657.2	867	49	g.47557244G>A	c.1448C>T	p.Ala483Val	rs145609043	-	3	0,02%	0	0	-6.6	-	Deleterious	deleterious	2.223	0.992	64	1 htz	VOUS
APN-114	M	AD		*EPB41L1*	602879	NM_012156.2	73	56	g.34782048C>T	c.1301-86C>T	p.?	rs139014428	-	-	-	0	0	0	New DSS	NA	NA	0.125	0,00	NA	2 htz	VOUS
		AR		*PRSS12*	606709	NM_003619.3	433	50	g.119204127T>C	c.2179A>G	p.Ile727Val	rs146362932	-	3	0,02%	0	0	0	-	Deleterious	Deleterious	2.304	1,00	29	1 htz	Possibly damaging
				RABL6	610615	NM_001173988.1	145	51	g.139734624C>T	c.1952C>T	p.Pro651Leu	rs199511338	-	5	0,04%	0	0	-0.5	-	Tolerated	NA	1.416	0.331	98	1 htz	Possibly benign
APN-115	M	XL		*ARHGEF6*	300267	NM_004840.2	365	98	g.135754192T>G	c.2122A>C	p.Ile708Leu	-	-	-	-	0	0	0	-	Tolerated	neutral	0.77	0.976	5	1 hemz	Possibly benign
				KIAA2022	300524	NM_001008537.2	532	98	g.73962891C>T	c.1501G>A	p.Gly501Ser	-	-	-	-	0	0	0	-	Tolerated	neutral	1.82	1,00	56	1 hemz	Possibly benign
		AD		*SHANK1*	604999	NM_016148.2	45	42	g.51171345C>T	c.3872G>A	p.Gly1291Asp	-	-	-	-	0	0	0	-	Tolerated	deleterious	3.514	1,00	94	1 htz	VOUS
				ANK3	600465	NM_020987.3	1077	49	g.61831298A>G	c.9341T>C	p.Val3114Ala	rs181031970	-	-	-	0	0	0	-	Deleterious	neutral	2.223	0.819	64	1 htz	VOUS
		AR	inherited (Mo)	*PRRT2*	614386	NM_145239.2	451	39	g.29825024dup	c.649dup	p.Arg217Profs*8	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
				MCPH1	607117	NM_001172574.1	276	45	g.6264211G>A	c.22+1G>A	p.?	-	-	-	-	-100	-100	-100	-	NA	NA	1.013	0.969	NA	1 htz	VOUS
				PIGV	610274	NM_001202554.1	594	46	g.27124130C>T	c.1277C>T	p.Pro426Leu	rs146969255	-	20	0,15%	0	0	0	-	Deleterious	neutral	2.062	0.969	98	1 htz	VOUS
APN-116	F	XL		*SLC6A8*	300036	NM_005629.3	299	51	g.152960226C>G	c.1649C>G	p.Thr550Ser	rs199635059	-	1	0,01%	0	0	0	-	Tolerated	neutral	1.9	1,00	58	1 htz	Possibly benign
		AD		*EHMT1*	607001	NM_024757.4	281	48	g.140611140G>A	c.148G>A	p.Ala50Thr	rs78104547	-	-	-	0	0	0	-	Tolerated	neutral	0.286	0,00	58	1 htz	Possibly benign
		AR		*RELN*	600514	NM_005045.3	771	47	g.103136238C>G	c.9301G>C	p.Asp3101His	-	-	-	-	0	0	0	-	Deleterious	deleterious	6.016	1,00	81	1 htz	VOUS
		?		*DIP2B*	611379	NM_173602.2	1017	50	g.51108269T>C	c.2741T>C	p.Ile914Thr	-	-	-	-	0	0	0	-	Tolerated	neutral	4.806	1,00	89	1 htz	Possibly benign
APN-117	F	XL		*MBTPS2*	300294	NM_015884.3	789	45	g.21896786C>T	c.1237C>T	p.His413Tyr	rs200298161	-	3 (2F, 1M)	0,03%	0	0	0	-	Tolerated	neutral	1.739	1,00	83	1 htz	Possibly benign
		AD	de novo	*TCF4*	602272	NM_001083962.1	944	49	g.53017619G>A	c.520C>T	p.Arg174*	-	-	-	-	0	0	0	-	NA	NA	0.932	1,00	NA	1 htz	Pathogenic
				HDAC4	605314	NM_006037.3	599	47	g.240036871C>T	c.1654G>A	p.Ala552Thr	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	-0.521	0,00	58	1 htz	Possibly benign
				MCPH1	607117	NM_024596.3	595	49	g.6479014C>G	c.2254C>G	p.Arg752Gly	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.564	0.992	125	1 htz	VOUS
		AR		*ASPM*	605481	NM_018136.4	947	47	g.197112706G>A	c.676C>T	p.Pro226Ser	rs147830520	-	1	0,01%	0	0	0	-	Tolerated	neutral	1.82	0.772	74	1 htz	Possibly benign
				MTHFR	607093	NM_005957.4	578	40	g.11854556_11854558del	c.1204_1206del	p.Asp402del	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
APN-118	F	XL		*IQSEC2*	300522	NM_001111125.1	672	44	g.53283945T>G	c.1168A>C	p.Met390Leu	-	-	-	-	0	0	0	-	Tolerated	neutral	2.627	1,00	15	1 htz	Possibly benign
				FLNA	300017	NM_001110556.1	465	50	g.153588472C>T	c.3691G>A	p.Val1231Ile	-	-	-	-	0	0	0	-	Tolerated	NA	0.69	0.803	29	1 htz	VOUS
		AD		*SYNGAP1*	603384	NM_006772.2	252	57	g.33411567G>A	c.3238G>A	p.Ala1080Thr	-	-	-	-	0	0	0	-	Deleterious	neutral	1.739	0.984	58	1 htz	VOUS
				LAMC3	604349	NM_006059.3	173	57	g.133948055G>A	c.3250G>A	p.Glu1084Lys	rs146221263	-	42	-	0	0	0	-	Tolerated	neutral	0.367	0,00	56	1 htz	Possibly benign
				PRODH	606810	NM_016335.4	326	50	g.18904466T>C	c.1463A>G	p.Asn488Ser	rs139903009	-	24	0,18%	0	0	0	-	Tolerated	neutral	0.851	0.677	46	2 htz	Possibly benign
		AR		*TRAPPC9*	611966	NM_031466.5	271	42	g.141468584_141468592del	c.72_80del	p.Ser27_Ala29del	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
				SCAPER	611611	NM_001145923.1	919	49	g.76914159T>A	c.1919A>T	p.Tyr640Phe	rs184003295	-	45	0,38%	0	0	0.1	-	Tolerated	neutral	1.416	1,00	22	1 htz	Possibly benign
APN-119	M	AD																											
APN-120	M	AR		*NPC2*	601015	NM_006432.3	1107	46	g.74951210C>T	c.271G>A	p.Asp91Asn	rs148607507	-	8	0,06%	0	0	0	-	Tolerated	deleterious	4.564	1,00	23	1 htz	VOUS
				PRSS12	606709	NM_003619.3	1579	49	g.119203216C>T	c.2503G>A	p.Gly835Arg	rs139833696	-	2	0,02%	0	NA	0	-	Deleterious	NA	3.272	0.819	125	1 htz	VOUS
APN-121	M	XL	inherited (Mo)	*IL1RAPL1*	300206	NM_014271.3	532	100	g.29973600A>G	c.1754A>G	p.Gln585Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.322	1,00	43	1 hemz	VOUS
		AD		*DOCK8*	611432	NM_203447.3	500	46	g.102102C>G	c.541C>G	p.His181Asp	rs200684000	-	-	-	0	0	0.2	-	Tolerated	neutral	NA	0.008	81	1 htz	Possibly benign
		AR		*CC2D1A/C19orf57*	610055	NM_017721.4	65	26	g.14017269_14017277dup	c.15_23dup	p.Pro10_Gly12dup	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	VOUS
APN-122	F	XL		*HCCS*	300056	NM_005333.4	509	46	g.11139827A>G	c.704A>G	p.Asn235Ser	-	-	-	-	0	0	0	-	Tolerated	neutral	1.82	0.433	46	1 htz	Possibly benign
		AD	de novo	*SHANK3*	606230	NM_001080420.1	51	23	g.51159168_51159183dup	c.2955_2970dup	p.Pro992Argfs*325	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	Pathogenic
				RAI1	607642	NM_030665.3	344	48	g.17696363G>A	c.101G>A	p.Ser34Asn	-	-	-	-	0	0	0	-	Tolerated	neutral	2.142	1,00	46	1 htz	Possibly benign
			inherited (Fa)	*SCN2A*	182390	NM_021007.2	490	50	g.166183279T>G	c.2017-83T>G	p.?	-	-	-	-	0	0	0	New DSS	NA	NA	1.416	0,00	NA	1 htz	VOUS
APN-123	M	AR		*MPI*	154550	NM_002435.1	186	43	g.75182424C>T	c.10C>T	p.Pro4Ser	rs143982014	-	17	0,13%	0	0	0	-	Tolerated	neutral	0.528	0,00	74	2 htz	Possibly benign
APN-124	M	AD		*ANKRD11*	611192	NM_001256182.1	478	51	g.89347214G>T	c.5736C>A	p.Asp1912Glu	-	-	-	-	0	0	0	-	Deleterious	NA	0.044	0.992	45	1 htz	VOUS
				FOLR1	136430	NM_016724.2	584	49	g.71906793T>C	c.493+2T>C	p.?	rs144637717	-	27 (htz)	0,21%	-100	-10.9	-100	-	NA	NA	2.385	1,00	NA	1 htz	Possibly benign
		AR		*LAMC3*	604349	NM_006059.3	65	38	g.133884672C>T	c.71C>T	p.Ala24Val	-	-	-	-	0	0	0	-	Deleterious	neutral	3.192	1,00	64	1 htz	VOUS
				AP4S1	607243	NM_007077.3	870	47	g.31553981A>G	c.373A>G	p.Ile125Val	rs200969079	-	3	0,02%	0	0	0.5	-	Tolerated	neutral	-0.844	0,00	29	1 htz	Possibly benign
		?		*CYFIP1*	606322	NM_014608.2	670	51	g.22998426G>A	c.3118G>A	p.Gly1040Arg	-	-	-	-	3	0	5.5	-	Deleterious	deleterious	4.402	1,00	125	1 htz	VOUS
APN-125	F	XL		*RPS6KA3*	300075	NM_004586.2	682	50	g.20213116T>C	c.406+67A>G	p.?	-	-	-	-	0	0	0	New DSS	NA	NA	-0.602	0,00	NA	1 htz	VOUS
		AD	inherited (Fa)	*KIRREL3*	607761	NM_032531.3	708	48	g.126343221C>T	c.574G>A	p.Gly192Arg	-	-	-	-	0	0	0	-	Deleterious	deleterious	3.272	0.992	125	1 htz	Benign
APN-126	F	AR		*FOLR1*	136430	NM_016724.2	805	46	g.71906438C>T	c.292C>T	p.Arg98Trp	rs76191655	-	25	0,19%	0	0	0	-	Deleterious	neutral	1.658	0.772	101	1 htz	VOUS
				MCPH1	607117	NM_001172574.1	1150	47	g.6302847C>G	c.1604C>G	p.Pro535Arg	-	-	-	-	0	0	0	-	Tolerated	deleterious	-0.924	0,00	103	1 htz	VOUS
APN-127	M	AR		*NSUN2*	610916	NM_017755.5	1433	50	g.6600143C>G	c.2200G>C	p.Glu734Gln	-	-	-	-	0	0	0	-	Tolerated	neutral	0.932	0,00	29	1 htz	Possibly benign
				GRIN2A	138253	NM_001134407.1	653	51	g.10032401G>T	c.422C>A	p.Thr141Lys	-	-	-	-	0	0	-17.5	-	Tolerated	neutral	0.77	0.811	78	1 htz	Possibly benign
				LAMA1	150320	NM_005559.2	1115	45	g.6974948C>A	c.6577G>T	p.Asp2193Tyr	rs149080936	-	6	0,05%	0	0	0	-	Tolerated	deleterious	2.142	0.976	160	1 htz	VOUS
				PIGV	610274	NM_001202554.1	1523	47	g.27121036G>A	c.511G>A	p.Ala171Thr	-	-	-	-	0	0	0	-	Tolerated	neutral	3.514	1,00	58	1 htz	Possibly benign
				ZNF526	614387	NM_133444.1	579	44	g.42729310_42729312del	c.755_757del	p.Glu252del	rs138669870	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	2 htz	Possibly benign
APN-128	M	XL		*HUWE1*	300697	NM_031407.4	156	108	g.53655440G>A	c.1242+4C>T	p.?	-	-	4 (1M)	0,05%	-66.5	0.9	NA	-	NA	NA	0.286	0.992	NA	1 htz	Possibly benign
				MED12	300188	NM_005120.2	460	60	g.70361169_70361170insCAC	c.6357_6358insCAC	p.Gln2119_Gln2120insHi	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	2 htz	VOUS
				PRRT2/MAZ	614386	NM_145239.2	423	54	g.29824442G>A	c.67G>A	p.Glu23Lys	rs140383655	-	19	0,15%	0	0	0	-	Tolerated	neutral	-0.037	0.669	56	1 htz	Possibly benign
APN-130	F		de novo	*MECP2*	300005	NM_004992.3	589	69	g.153296363G>A	c.916C>T	p.Arg306Cys	rs28935468	-	-	-	0	0	0	-	Deleterious	deleterious	5.29	1,00	180	1 htz	Pathogenic
		XL		*MECP2*	300005	NM_004992.3	350	56	g.153296174G>A	c.1105C>T	p.His369Tyr	-	-	-	-	0	0	0	-	Tolerated	neutral	4.322	1,00	83	1 htz	Possibly benign
				KIAA2022	300524	NM_001008537.2	602	83	g.73963628T>C	c.764A>G	p.Asn255Ser	-	-	1	0,01%	0	0	0	-	Deleterious	neutral	0.932	1,00	46	1 htz	Possibly benign
		AD		*ZNF599*	NA	NM_001007248.2	600	80	g.35249955A>G	c.1751T>C	p.Ile584Thr	-	-	-	-	0	0	0	-	Deleterious	neutral	1.658	0.984	89	1 htz	VOUS
				ZNF526	614387	NM_133444.1	458	66	g.42730555C>T	c.2000C>T	p.Thr667Met	-	-	-	-	0	0	0	-	Deleterious	deleterious	2.385	0.984	81	1 htz	VOUS
		AR		*MTHFR*	607093	NM_005957.4	581	76	g.11854525G>A	c.1237C>T	p.Pro413Ser	-	-	-	-	0	0	NA	-	Tolerated	neutral	2.465	1,00	74	1 htz	Possibly benign
APN-131	M		inherited (Fa)	*SLC2A1*	138140	NM_006516.2	714	48	g.43392779del	c.1412del	p.Gly471Glufs*37	-	-	-	-	0	0	NA	-	NA	NA	NA	NA	NA	1 htz	Possibly pathogenic
		AD	inherited (Mo)	*ANKRD11*	NA	NM_013275.5	587	86	g.89348867G>T	c.4083C>A	p.His1361Gln	-	-	-	-	0	0	0	-	Deleterious	deleterious	-0.198	0.945	24	1 htz	Possibly pathogenic
				ANKRD11	NA	NM_001256182.1	592	67	g.89345674C>T	c.7276G>A	p.Ala2426Thr	-	-	-	-	0	0	0	-	Tolerated	NA	5.694	1,00	58	1 htz	VOUS
				SHANK1	604999	NM_016148.2	296	66	g.51205864C>G	c.1607G>C	p.Gly536Ala	-	-	-	-	0	0	NA	-	Tolerated	neutral	0.367	0.992	60	1 htz	Possibly benign
		AR		*RELN*	600514	NM_173054.2	600	78	g.103163890C>T	c.7438G>A	p.Gly2480Ser	rs150236371	-	29	0,22%	0	0	0	-	Deleterious	deleterious	4.079	0.992	56	1 htz	VOUS
		XL	inherited (Mo)	*FMR1*	309550	NM_002024.5	293	104	g.147024687G>A	c.1312G>A	p.Asp438Asn	-	-	-	-	0	0	0	-	Deleterious	deleterious	5.048	1,00	23	1 hemz	Benign

APN-132	M	AR		*AP4B1*	607245	NM_006594.2	528	59	g.114442885A>G	c.755T>C	p.Val252Ala	rs141417436	-	25	0,19%	0	0	0	-	Deleterious	neutral	2.465	1,00	64	2 htz	VOUS
				LINS	610350	NM_001040616.2	595	78	g.101115269T>C	c.554A>G	p.Asn185Ser	rs146704559	-	4	0,03%	0	0	0	-	Tolerated	neutral	0.448	0,00	46	1 htz	Possibly benign
		?		*CYFIP1*	606322	NM_014608.2	469	53	g.22969215G>A	c.2441G>A	p.Arg814Gln	rs147972810	-	2	0,02%	0	0	0	-	Tolerated	neutral	1.658	1,00	43	1 htz	Possibly benign
APN-134	M	AR		*RABL6*	NA	NM_001173989.2	599	64	g.139728202T>G	c.736T>G	p.Cys246Gly	rs187468519	-	-	-	0	0	0	-	Tolerated	neutral	0.448	0.051	159	1 htz	Possibly benign
		XL	inherited (Mo)	*KDM5C*	314690	NM_004187.3	707	91	g.53240784dup	c.1296dup	p.Glu433*	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 hemz	Pathogenic
APN-135	M	AD		*DISC1*	605210	NM_018662.2	582	83	g.231935893A>G	c.1729A>G	p.Lys577Glu	-	-	-	-	0	0	0	-	Deleterious	deleterious	1.174	0.26	56	1 htz	VOUS
		AR		*UPB1*	606673	NM_016327.2	545	46	g.24919670C>T	c.1000C>T	p.Arg334Trp	-	-	1	0,01%	0	0	0	-	Deleterious	deleterious	1.497	0,00	101	1 htz	VOUS
		XL	inherited (Mo)	*CUL4B*	300304	NM_003588.3	798	95	g.119681009_119681010del	c.811_812del	p.Gln271Aspfs*11	-	-	-	-	NA	NA	NA	-	NA	NA	NA	NA	NA	1 hemz	Pathogenic
				LAMA1	150320	NM_005559.2	594	85	g.6978321C>T	c.6064G>A	p.Ala2022Thr	rs140764072	-	13	0,10%	0	0	0	-	Tolerated	deleterious	2.707	0.827	58	1 htz	VOUS
APN-137	M	AR		*NRXN1*	600565	NM_001135659.1	547	60	g.51255090G>A	c.322C>T	p.Pro108Ser	rs199784029	-	18	0,14%	0	0	0	-	NA	neutral	2.788	0.984	74	1 htz	Possibly benign
				RALGDS	601619	NM_001042368.1	327	52	g.135977121G>A	c.2075C>T	p.Pro692Leu	-	-	-	-	0	0	0	-	Tolerated	neutral	2.304	1,00	98	1 htz	Possibly benign
				NPC2	601015	NM_006432.3	579	90	g.74951269T>C	c.212A>G	p.Lys71Arg	rs142075589	-	2	0,02%	0	0	0	-	Tolerated	neutral	0.932	0.764	26	1 htz	Possibly benign
		XL	inherited (Mo)	*ATRX*	300032	NM_000489.3	604	110	g.76972632G>A	c.109C>T	p.Arg37*	rs122445108	**pathogenic**	-	-	0	0	0	-	NA	NA	1.658	1,00	NA	1 hemz	Pathogenic
			inherited (Mo)	*CDH15*	114019	NM_004933.2	303	46	g.89251647G>C	c.569G>C	p.Arg190Pro	-	-	-	-	0	0	0	-	Deleterious	deleterious	2.062	0.984	103	1 htz	Benign
		AD		*DISC1*	605210	NM_001164550.1	572	87	g.231856789G>T	c.1214G>T	p.Gly405Val	-	-	-	-	0	0	0	New DSS	Notscored	deleterious	-0.037	0.02	109	1 htz	VOUS
APN-138	M			*MED13L*	608771	NM_015335.4	603	70	g.116446347G>A	c.1871C>T	p.Ser624Leu	rs200545513	-	1	0,01%	0	0	0	-	Tolerated	neutral	0.528	0.969	145	1 htz	Probably benign
				LAMA1	150320	NM_005559.2	562	79	g.6977814T>G	c.6257A>C	p.Lys2086Thr	rs142934543	-	8	0,06%	0	0	0	-	Tolerated	deleterious	4.402	1,00	78	1 htz	VOUS
		AR		*KDM5A*	180202	NM_001042603.1	583	84	g.402319T>G	c.4472A>C	p.Glu1491Ala	rs201998974	-	1	0,01%	0	0	-1.3	-	Tolerated	neutral	3.595	1,00	107	1 htz	Possibly benign
		XL		*IQSEC2*	300522	NM_015075.1	474	104	g.53272587C>A	c.2201G>T	p.Arg734Leu	-	-	-	-	0	0	0	-	Tolerated	deleterious	5.532	1,00	102	1 hemz	VOUS
				KIAA2022	300524	NM_001008537.2	580	120	g.73964127G>A	c.265C>T	p.His89Tyr	-	-	-	-	0	0	0	-	Deleterious	neutral	5.129	1,00	83	1 hemz	VOUS
			de novo	*SYNGAP1*	603384	NM_006772.2	11	100	g.33414346G>A	c.3583-6G>A	p.?	-	-	-	-	-93.6	-100	NA	New ASS	NA	NA	2.869	1,00	NA	1 htz	Pathogenic
APN-139	M	AD	inherited (Mo)	*MED13L*	608771	NM_015335.4	572	91	g.116413113C>T	c.5594G>A	p.Arg1865Gln	-	-	-	-	0	0	0.1	-	Deleterious	deleterious	4.241	1,00	43	1 htz	Benign
				ARID1B	NA	NM_020732.3	293	35	g.157099186dupCGCGGCGGCA	c.133_141dup	p.Ala45_Ala47dup	-	-	-	-	0	0	0	-	NA	NA	NA	NA	NA	1 htz	Possibly benign
				CC2D1A	610055	NM_017721.4	596	87	g.14020663G>T	c.88G>T	p.Asp30Tyr	-	-	-	-	0	0	0	-	Deleterious	deleterious	4.564	0.976	160	1 htz	VOUS
		AR		*CC2D1A*	610055	NM_017721.4	587	87	g.14020723G>T	c.148G>T	p.Ala50Ser	-	-	1	0,01%	0	0	0	-	Tolerated	neutral	3.03	0.992	99	1 htz	Possibly benign
		XL		*CUL4B*	300304	NM_003588.3	314	106	g.119694175G>C	c.373C>G	p.Leu125Val	-	-	-	-	0	0	0	-	Tolerated	neutral	0.125	0.992	32	1 hemz	Possibly benign
				DOCK8	NA	NM_203447.3	588	78	g.197135C>T	c.3460C>T	p.Arg1154Cys	rs34390308	-	-	-	0	0	0	-	Deleterious	deleterious	NA	1,00	180	1 htz	VOUS
		AD		*PGA5*	169730	NM_014224.2	475	53	g.61015898A>C	c.664A>C	p.Lys222Gln	rs200053998	-	-	-	0	0	5.5	-	Tolerated	neutral	-1.812	0,00	53	1 htz	Possibly benign
APN-141	M			*DISC1*	605210	NM_001164537.1	388	49	g.231881156C>T	c.1124C>T	p.Thr375Met	-	-	-	-	0	0	-0.4	-	Notscored	neutral	-0.682	0.004	81	1 htz	VOUS
				NRXN1	600565	NM_001135659.1	595	75	g.51254883G>A	c.529C>T	p.Arg177Trp	-	-	-	-	0	0	0	-	NA	neutral	2.062	1,00	101	1 htz	VOUS
		AR		*LAMC3*	604349	NM_006059.3	546	53	g.133951317G>C	c.3594G>C	p.Arg1198Ser	rs199612934	-	-	-	0	0	0	-	Tolerated	neutral	-2.297	0,00	110	1 htz	Possibly benign
				MPI	154550	NM_002435.1	356	54	g.75182424C>T	c.10C>T	p.Pro4Ser	rs143982014	-	17	0,13%	0	0	0	-	Tolerated	neutral	0.528	0,00	74	2 htz	Possibly benign
				CENPJ	609279	NM_018451.3	600	82	g.25486989T>C	c.175A>G	p.Thr59Ala	rs138732534	-	26	0,20%	0	0	0	-	Tolerated	neutral	-0.36	0,00	58	1 htz	Possibly benign
			de novo	*MECP2*	300005	NM_004992.3	596	84	g.153296777G>A	c.502C>T	p.Arg168*	rs61748421	-	-	-	0	0	0	New DSS	NA	NA	1.82	1,00	NA	1 htz	Pathogenic
				FLNA	300017	NM_001110556.1	289	48	g.153599601G>A	c.13C>T	p.His5Tyr	-	-	-	-	0	0	0	-	Deleterious	neutral	4.564	1,00	83	1 htz	VOUS
		XL		*IDS*	300823	NM_000202.5	589	82	g.148579816T>C	c.530A>G	p.Glu177Gly	-	-	-	-	0	0	0	-	Tolerated	neutral	0.286	0.984	98	1 htz	Possibly benign
APN-142	F			*SLC9A6*	300231	NM_001042537.1	589	83	g.135076986C>T	c.367C>T	p.Pro123Ser	-	-	-	-	0	0	0	-	Tolerated	neutral	5.048	1,00	74	1 htz	Possibly benign
				OCRL	300535	NM_000276.3	609	83	g.128696622C>A	c.1103C>A	p.Thr368Asn	-	-	-	-	0	0	NA	-	Tolerated	neutral	3.514	1,00	65	1 htz	Possibly benign
		AR		*ANK3*	600465	NM_001204403.1	128	56	g.62493041C>T	c.37G>A	p.Glu13Lys	-	-	-	-	0	0	0	-	Deleterious	neutral	-0.521	0,00	56	1 htz	VOUS
				RELN	600514	NM_005045.3	586	74	g.103197603G>A	c.5618C>T	p.Thr1873Ile	rs41275239	-	30	0,23%	0	0	0.1	-	Tolerated	neutral	2.869	1,00	89	2 htz	Possibly benign

MOI: mode of inheritance; XL: X-linked, AD: autosomal dominant, AR: autosomal recessive
M: Male, F: Female
Fa: father, Mo: mother
VOUS: Variant of unknown significance

possibly causative mutation
certainly causative mutation