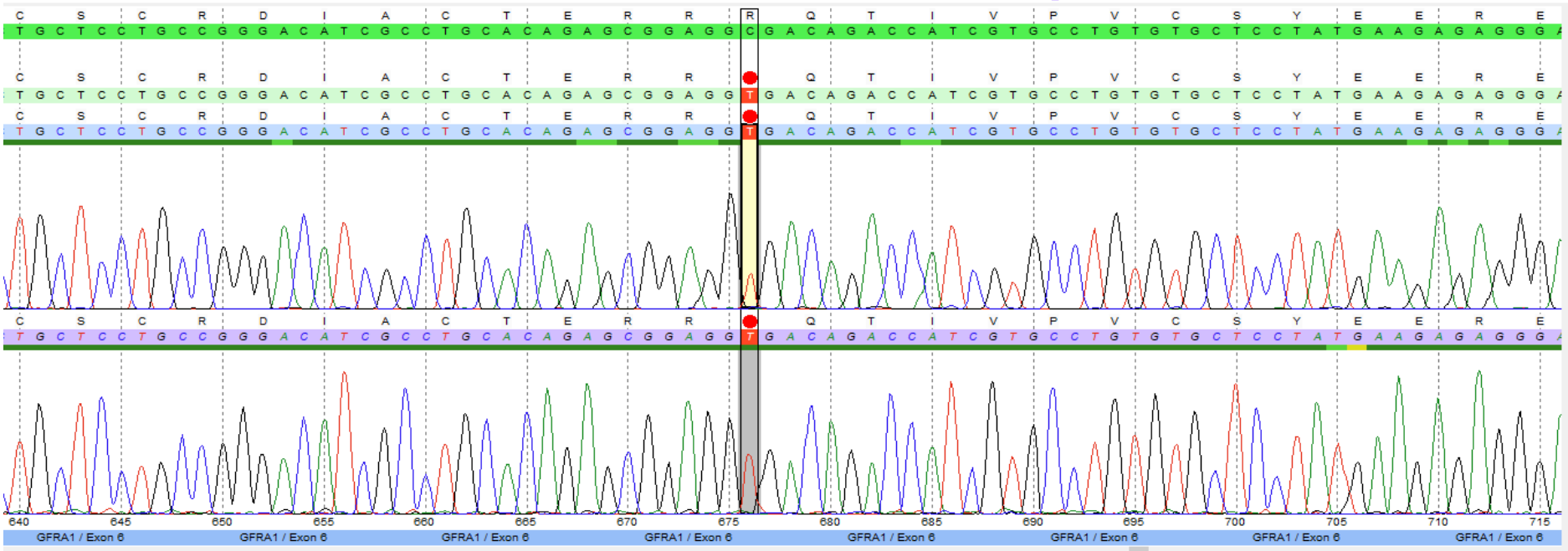
Supplementary table: Homozygous variants found in raw data analysis. Rare homozygous coding variants remaining as candidates in family 1 and 2. Homozygous variants detected in other samples in our or external data repositories (healthy individuals or affected with a distinct phenotype) were excluded.

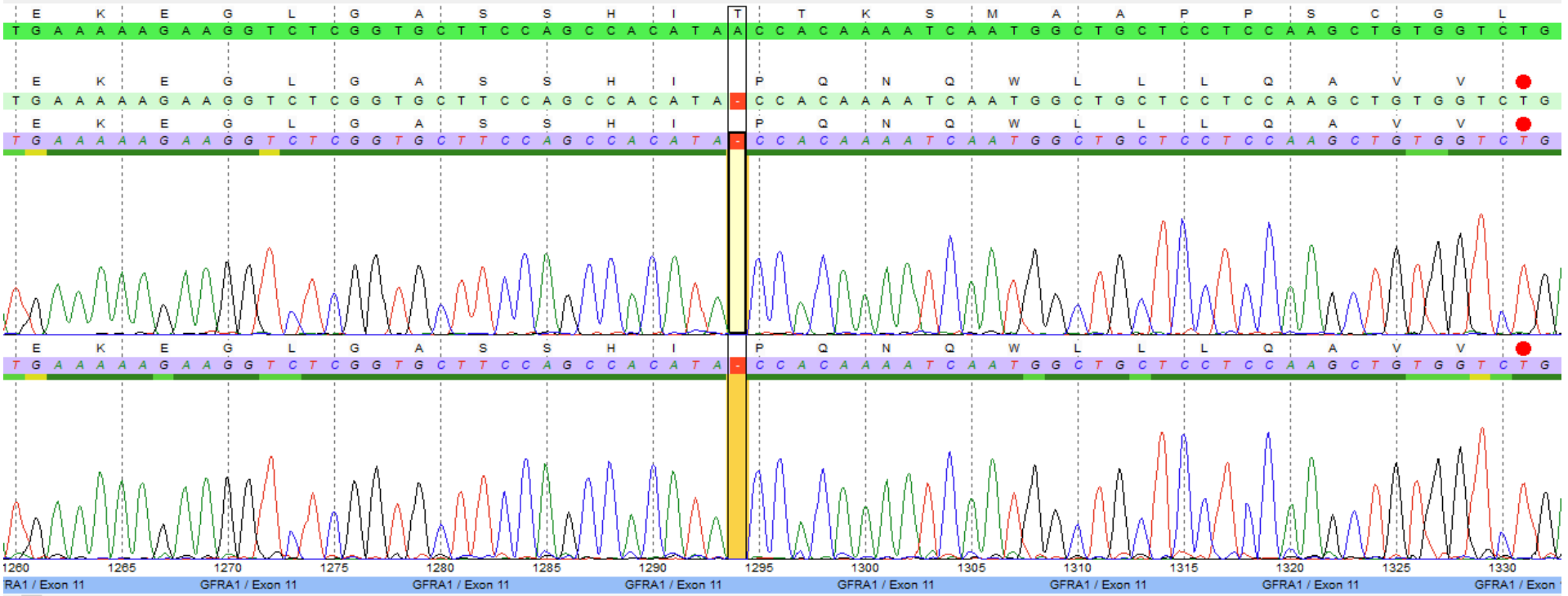
Chr	Genomic coordinate	Gene	Reference seq: nucleotide change	Reference seq.: protein change	Variant_type	dbSNP	OMIM	phyloP	Cadd_raw	PopFreq Max
chr10	**117884826**	***GFRA1***	**NM_005264.5:c.676C>T**	**NM_005264.5:p.Arg226***	**Nonsense**	**rs191814086**	**601496**	**6.762**	**14.5264**	**0.0008**
chr17	3653725	*ITGAE*	NM_002208.4:c.1945A>G	NM_002208.4:p.Met649Val	Missense	rs770556756		-0.126	-3.68351	0
chr1	94685934	*ARHGAP29*	NM_001328664.1:c.220G>C	NM_001328664.1:p.Val74Leu	Missense	rs574016258	610496	3.205	0.932488	0.002
chr10	102849625	*TLX1NB*	NM_001085398.1:c.38G>C	NM_001085398.1:p.Arg13Pro	Missense	rs151326673	612734		1.6072	0.0002
chr10	111886247	*ADD3*	NM_001320591.1:c.1594G>A	NM_001320591.1:p.Asp532Asn	Missense	rs757097119	601568 [Cerebral palsy, spastic quadriplegic, 3]	9.602	6.98095	0.0002
chr10	**117824012**	***GFRA1***	**NM_001348097.1:c.1294delA**	**NM_001348097.1:p.Thr432fs**	**Frameshift**		**601496**			**0**
chr10	124336125	*DMBT1*	NM_007329.2:c.494C>T	NM_007329.2:p.Ala165Val	Missense	rs200063826	601969	0.647	0.619438	0.0031
chr10	124339277	*DMBT1*	NM_007329.2:c.863A>G	NM_007329.2:p.Gln288Arg	Missense	rs764477516	601969	-0.888	-2.18407	0.0044
chr10	127530438	*DHX32*	NM_018180.2:c.1417A>C	NM_018180.2:p.Asn473His	Missense	rs756666817	607960; 611883	6.158	5.70413	0.0011
chr2	21360860	*TDRD15*	NM_001306137.1:c.521C>T	NM_001306137.1:p.Ser174Phe	Missense	rs530287177		0.423	4.85385	0.001
chr2	26717916	*OTOF*	NM_001287489.1:c.791G>A	NM_001287489.1:p.Arg264Gln	Missense	rs370475628	603681 [Auditory neuropathy type 1,Deafness type 9]	6.127	6.54147	0.0002
chr2	186671879	*FSIP2*	NM_173651.3:c.17846T>G	NM_173651.3:p.Leu5949Arg	Missense	rs762024267	615796 [Spermatogenic failure 34]	4.056	3.82643	0.0017
chr2	201501670	*AOX1*	NM_001159.3:c.2383A>C	NM_001159.3:p.Lys795Gln	Missense		602841	3.405	4.68488	0
chr8	11970471	*ZNF705D*	NM_001039615.3:c.707C>G	NM_001039615.3:p.Ala236Gly	Missense	rs773967266		0.666	1.26954	0
chrX	50053245	*CCNB3*	NM_033031.2:c.2076G>C	NM_033031.2:p.Glu692Asp	Missense	rs782385072	300456	1.724	-0.76569	0.0001
chrX	55248225	*PAGE5*	NM_130467.4:c.167G>T	NM_130467.4:p.Arg56Leu	Missense	rs753525619	301009	-1.629	0.638329	0.0004
chrX	78618155	*ITM2A*	NM_004867.4:c.475A>G	NM_004867.4:p.Asn159Asp	Missense	rs780824997	300222	6.368	4.85555	0.0002
chrX	134988296	*SAGE1*	NM_018666.2:c.568A>G	NM_018666.2:p.Ile190Val	Missense	rs782222781	300359	-0.125	-1.31576	0.001

PopMaxFreq (Population maximum frequency): indicates the highest frequency of the variant observed in databases. PhyloP scores indicate evolutionary conserved positions (high positive). CADD (Combined Annotation-Dependent Depletion) ranks genetic variants, including single nucleotide variants (SNVs) and short inserts and deletions (InDels), throughout the human genome reference assembly. RAW score above 4 are considered most as likely pathogenic (ref. PMID: 30371827).

Supplementary figure 1 : Chromatograph of variant detected in family 1 NM_005264.5: c.676C>T, (p.Arg226*)



Supplementary figure 2: chromatograph od variant detected in family 2 NM_005264.5: c.1294delA, (p.Thr432Profs*)



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