

Supplementary Table 1: Filtered variants for affected members of RDHM-05

Chr	Start	Ref	Alt	Gene name	Amino acid change	1000G_ALL	REVEL	ClinPred	Comment
1	145532549	T	C	<i>ITGA10</i>	p.(Phe 191Phe)	.	.	.	Synonymous SNV
2	109381202	A	G	<i>RANBP2</i>	p. (Asn1403 Asp)	.	0.033	0.02	Not conserved
2	109545803	G	A	<i>EDAR</i>	p.(Tyr 69 Tyr)	0.0014	0.0014	0.001	Synonymous SNV
8	8185918	C	T	<i>PRAG1</i>	p.(Val792Ile)	0.0016	0.172	0.058	Not conserved, 2 homozygous reported in ExAC
8	10469039	G	A	<i>RP1L1</i>	p.(Pro 857 Ser)	.	0.085	0.615	Not conserved
8	12947891	T	C	<i>DLC1</i>	p. (Asn 804Ser)	.	0.015	0.019	Not conserved, very low REVEL and ClinPred scores
10	47000004	T	C	<i>GPRIN2</i>	p.(Val375Ala)	.	0.042	0.002	Not conserved, not present in region of homozygosity
10	47087680	C	T	<i>NPY4R</i>	p. (His 299 His)	.	.	.	Synonymous SNV
10	47087731	C	T	<i>NPY4R</i>	p. (Cys 316 Cys)	.	.	.	Synonymous SNV
14	20482735	-	CTT	<i>OR4K14</i>	p.(Leu 206_Leu 207ins Arg)	.	.	.	Non-frameshift insertion, Olfactory receptor
14	20766966	T	C	<i>TTC5</i>	p.(Tyr210Cys)	.	0.828	0.99	Conserved, High Revel and ClinPred scores, Segregates
21	10920098	T	C	<i>TPTE</i>	p. (Lys 248Glu)	.	0.329	0.009	Not conserved, not present in region of homozygosity

Chr, chromosome; Ref, reference; Alt, alternative; 1000G-ALL, 1000 genomes allele frequency; REVEL and ClinPred