

Supplemental Table S1: Confirmed nonsynonymous SNV coverage metrics and minor allele frequencies

Fam ID	Sub ID	Gene Symbol and Name	Chr	Pos	Ref	Alt	Region of Homozygosity Size	Comb Score	AA Change	Quality Score	Allele Depth (Ref,Alt)	Phred Likelihood (0/0,0/1,1/1)	rs#	1000G Freq (%)	EVS Eur Freq (%)	EVS Afr Freq (%)	EVS Freq (%)
283	1511	DAK Dihydroxyacetone Kinase 2 Homolog	11	61113007	C	T	808kb	9.5	A474V	1892.32	3,62	1950,95,0	rs34152582	0.05	0	0.41	0.14
283	1511	CD248 CD248 Molecule, Endosialin	11	66084374	C	T	51kb	7.3	R42H	278.18	0,10	332,27,0	rs199630152	0.03	0.95	0.33	
283	1511	IFI44L Interferon-induced Protein 44-like	1	79095572	G	A	2876kb	6.0	G232E	5925.08	2,154	5892,457,0	rs75931592	0.27	0	0.61	0.21
283	1511	MKGPRD MAS-related GPR, Member D	11	68747840	G	A	-	4.8	R206W	1127.91	0,36	1185,96,0	rs146025023	0.14	0	1.05	0.35
466	2150	DLG1 Discs, Large Homolog 1	3	196792663	C	A	557kb	7.3	V717L	8023.66	0,119	4623,355,0	rs148283553	0.05	0.14	0.25	0.18
466	2239	DLG1 Discs, Large Homolog 1	3	196792663	C	A	557kb	7.3	V717L	8023.66	2,80	2989,165,0	rs148283553	0.05	0.14	0.25	0.18
466	2150	ZNF77 Zinc Finger Protein 77	19	2934694	C	A	58kb	6.6	C144F	20002.73	0,282	10556,831,0	rs76396690	1	0.03	1.91	0.67
466	2239	ZNF77 Zinc Finger Protein 77	19	2934694	C	A	58kb	6.6	C144F	20002.73	4,155	5679,324,0	rs76396690	1	0.03	1.91	0.67
466	2150	GBP7 Guanylate Binding Protein 7	1	89615023	C	G	3067kb	5.9	M368I	14158.08	1,215	7575,617,0	rs74098339	1	0	3.25	1.1
466	2239	GBP7 Guanylate Binding Protein 7	1	89615023	C	G	3067kb	5.9	M368I	14158.08	0,180	6245,515,0	rs74098339	1	0	3.25	1.1
475	2173	TMEM63A Transmembrane Protein 63A	1	226054381	T	G	81kb	7.0	N190H	5131.37	1,69	2680,202,0	rs115347439	1	0.02	3.79	1.3
475	2173	MTMR11 Myotubularin Related Protein 11	1	149902426	G	A	3613kb	6.3	T421I	18440.51	3,249	9132,619,0	rs114842926	0.18	0.22	0.02	0.15
489	2206	MMP2 Matrix Metalloproteinase 2	16	55525819	G	T	246kb	9.1	K429N	4043.24	4,115	3938,244,0	rs201083413	0.05			
489	2206	XKR9 XK, Kell Blood Group Complex Subunit-related Family,	8	71646361	T	A	11277kb	8.0	F275Y	5124.18	1,137	5079,367,0	rs74941166	0.41	0.48	0.09	0.35

Member 9																	
489	2206	ZNF696 Zinc Finger Protein 696	8	144377951	C	T	-	2.3	R36W	361.74	0,12	419,33,0	rs140398403	0.37	0.69	0.18	0.52

Chr: Chromosome; Pos: Position; Ref: Reference Allele; Alt: Alternate Allele;; CombScore: Combined Weighted Pathogenicity Score; AA Change: Amino Acid Change; rs#: Reference SNP #; 1000G Freq: 1000 Genomes Database Allele Frequency; EVS: Exome Variant Server; Eur: European; Afr: African; Freq: Allele Frequency

Supplemental Table S2: Confirmed nonsynonymous SNV pathogenicity scores

Fam ID	Gene Symbol and Name	Chr	Pos	Ref	Alt	Region of Homozygosity Size	Comb Score	PhastCons	GERP	Sift Prediction	Sift Score	PP2 Predict	PP2 Prob	PhyloP	LRT	MT
283	DAK Dihydroxyacetone Kinase 2 Homolog	11	61113007	C	T	808kb	9.5	1	5.71	TOLERATED	0.09	probably damaging	0.99	0.999311	1	0.999977
283	CD248 CD248 Molecule, Endosialin	11	66084374	C	T	51kb	7.3	1	2.06	TOLERATED	0.08	probably damaging	0.953	0.945553	0.994846	0.260201
283	IFI44L Interferon-induced Protein 44-like	1	79095572	G	A	2876kb	6.0	0.035	2.83	DAMAGING	0.01	probably damaging	0.988	0.997777	0.999999	0.171589
283	MRGPRD MAS-related GPR, Member D	11	68747840	G	A	-	4.8	0	2.24	DAMAGING	0.05	probably damaging	0.979	0.929859	0.765883	0.095107
466	DLG1 Discs, Large Homolog 1	3	196792663	C	A	557kb	7.3	1	5.08	TOLERATED	0.26	possibly damaging	0.809	0.998657	0.99972	0.867802
466	ZNF77 Zinc Finger Protein 77	19	2934694	C	A	58kb	6.6	0.003	2.31	DAMAGING	0.01	possibly damaging	0.83	0.991509	1	1
466	GBP7 Guanylate Binding Protein 7	1	89615023	C	G	3067kb	5.9	1	2.97	TOLERATED	0.12	possibly damaging	0.621	0.992397	0.999988	0.445681
475	TMEM63A Transmembrane Protein 63A	1	226054381	T	G	81kb	7.0	0.924	5.04	DAMAGING	0.01	possibly damaging	0.733	0.997424	0.999996	0.355015
475	MTMR11 Myotubularin Related Protein 11	1	149902426	G	A	3613kb	6.3	0.761	4.84	DAMAGING	0.05	benign	0.467	0.999681	0.988959	0.925617
489	MMP2 Matrix Metalloproteinase 2	16	55525819	G	T	246kb	9.1	1	4.88	TOLERATED	0.08	probably damaging	0.916	0.999399	1	0.99996
489	XKR9 XK, Kell Blood Group Complex Subunit-related Family, Member 9	8	71646361	T	A	11277kb	8.0	1	3.06	TOLERATED	0.33	probably damaging	0.998	0.964684	0.999981	0.999476
489	ZNF696 Zinc Finger Protein 696	8	144377951	C	T	-	2.3	0.002	0.61	DAMAGING	0.01	Nonsynonymous	unknown	NaN	0.892855	0.828047

Chr: Chromosome; Pos: Position; Ref: Reference Allele; Alt: Alternate Allele; CombScore: Combined Weighted Pathogenicity Score

Supplemental Table S3: Confirmed frameshift insertion and deletion (indel) coverage metrics, minor allele frequencies, and pathogenicity scores

Family ID	Subject ID	Gene Symbol and Name	Chr	Pos	Ref	Alt	Region of Homozygosity Size	Quality Score	Allele Depth (Ref,Alt)	Phred Likelihood (0/0,0/1,1/1)	PhastCons	Type	NMD
283	1511	LEPR Leptin Receptor Precursor	1	66058400	CT	C	1297kb	4413.7	2,110	4414,241,0	0.95	FRAMESHIFT	1
466	2150	LEPR Leptin Receptor Precursor	1	66038116	CA	C	1703kb	6685.95	4,103	4132,130,0	0.929	FRAMESHIFT	1
466	2239	LEPR Leptin Receptor Precursor	1	66038116	CA	C	1703kb	6685.95	0,61	2554,184,0	0.929	FRAMESHIFT	1
475	2173	ACOT4 Acyl-coenzyme A Thioesterase 4	14	74060511	T	TTCAA	94kb	28352.32	6,135	11810,299,0	0.975	FRAMESHIFT	1
475	2173	ACOT4 Acyl-coenzyme A Thioesterase 4	14	74060513	GCTTA	G	94kb	19886.95	9,137	8553,239,0	1	FRAMESHIFT	0

Chr: Chromosome; Pos: Position; Ref: Reference Allele; Alt: Alternate Allele; NMD: Nonsense Mediated Decay