Supplementary Table 4

Autosomal dominant variants in family P05: Annotation is the same as in Supplementary Table 3.

Patient 05 Dominant Analysis

Gene	Inheritance Pattern	Sibling Status	Position (hg19)	Reference	Variant	Frequency (Exome Variant Server)	dbSNP ID (if available)	Functional Annotation (SnpEff)	Amino Acid change	PolyPhen2	Prediction	Function (UniProt)	Associated Diseases (OMIM)
C1orf141	Autosomal D	Unaffected	Chr1: 67559090	T	G	NA	NA	missense	K267N	0.046	benign	unknown	none
CELSR2	Autosomal D	Unaffected	Chr1: 109803819	T	C	NA	NA	missense	F1372L	0.998	probably damaging	receptor for cell/cell signaling in neural development	none
ILK	Autosomal D	Unaffected	Chr11: 6631060	A	G	NA	NA	missense	N321S	0.115	benign	regulates integrin-mediated signal transduction	
LAMB4	Autosomal D	Unaffected	Chr7: 107706230	C	T	NA	NA	missense	C938Y	1.000	probably damaging	laminin for extracellular interactions	none
GCC1	Autosomal D	Unaffected	Chr7: 127222636	T	C	NA	NA	missense	K587R	0.001	benign	involved in Golgi structure	none
SDS	Autosomal D	Unaffected	Chr12: 113836398	G	C	NA	NA	missense	A116G	0.004	benign	serine and threonine dehydratase	none
COX6B1	Autosomal D	Unaffected	Chr19: 36145529	G	A	NA	NA	missense	E55K	0.143	benign	cytochrome C oxidase subunit	cytochrome c oxidase deficiency
KCNJ14	Autosomal D	Unaffected	Chr19: 48967513	G	A	NA	NA	missense	D264N	0.996	probably damaging	potassium channel	none
NYNRIN	Autosomal D	Unaffected	Chr14: 24877504	G	A	NA	NA	missense	G210S	0.754	possibly damaging	unknown	none
IGF2R	Autosomal D	Unaffected	Chr6: 160489364	C	T	NA	NA	missense	P1400L	0.386	benign	receptor for IGF2 and other cell signaling ligands	
TTLL11	Autosomal D	Unaffected	Chr9: 124855120	C	G	NA	NA	missense	G193A	0.017	benign	modifies alpha tubulin	none
TMEM45B	Autosomal D	Unaffected	Chr11: 129728556	CTT	C	NA	NA	frame shift	NA	NA	NA	unknown	none
RELL1	Autosomal D	Unaffected	Chr4: 37640126	G	T	NA	NA	missense	A129E	1.000	probably damaging	unknown	none
BAG3	Autosomal D	Unaffected	Chr10: 121436295	CAGG	C	NA	NA	codon deletion	NA	NA	NA	regulates molecular chaperones and inhibit apoptosis	Dilated cardiomyopathy 1HH; Myofibrillar myopathy 6
WAC	Autosomal D	Unaffected	Chr10: 28878666	C	T	NA	NA	missense	P128L	0.000	benign	regulates transcription and RNA processing	none
ENAM	Autosomal D	Unaffected	Chr4: 71507984	A	T	NA	NA	missense	N281Y	0.967	probably damaging	formation of tooth enamel	Amelogenesis imperfecta type IB and IC
TRIM14	Autosomal D	Unaffected	Chr9: 100850210	A	T	NA	NA	missense	C291S	0.801	possibly damaging	unknown	none
CGB1	Autosomal D	Unaffected	Chr19: 49539490	C	A	NA	NA	missense	C27F	1.000	probably damaging	chorionic gonadotropin subunit	none

RBBP8	Autosomal D	Unaffected	Chr18: 20548820	GA	G	NA	NA	frame shift	NA	NA	NA	DNA repair pathways	Seckel syndrome 2; Jawad Syndrome
ZAN	Autosomal D	Unaffected	Chr7: 100389666	G	A	NA	NA	missense	P2536L	0.926	probably damaging	sperm protein for binding to egg	none
CCDC15	Autosomal D	Unaffected	Chr11: 124829751	C	T	NA	NA	missense	T123I	0.001	benign	unknown	none
SUPT5H	Autosomal D	Unaffected	Chr19: 39944011	C	T	NA	NA	missense	R31W	0.830	possibly damaging	regulates transcription and RNA processing	none
TMED6	Autosomal D	Unaffected	Chr16: 69383523	C	T	NA	NA	missense	R82Q	0.998	probably damaging	unknown	none
MOGAT2	Autosomal D	Unaffected	Chr11: 75428946	G	A	NA	NA	missense	A5T	0.999	probably damaging	fatty acid metabolism	none
GPR6	Autosomal D	Unaffected	Chr6: 110300437	C	T	NA	NA	missense	A41V	0.147	benign	G-protein coupled receptor	none
NLRP4	Autosomal D	Unaffected	Chr19: 56390314	G	T	NA	NA	missense	A951S	0.085	benign	regulates inflammatory pathways	none
KIAA1432	Autosomal D	Unaffected	Chr9: 5774209	C	G	NA	NA	missense	P1412R	0.077	benign	unknown	none
LRRC61	Autosomal D	Unaffected	Chr7: 150034315	C	T	NA	NA	missense	P122L	0.014	benign	unknown	none
NEMF	Autosomal dominant	Affected	Chr 14: 50253447	G	C	NA	NA	missense	Q973E	0.425	benign	nuclear export	none
PAX8	Autosomal dominant	Affected	Chr 2: 113977674	A	G	NA	NA	missense	L424S	0.992	probably damaging	thyroid transcription factor	Congenital hypothyroidism
MPP2	Autosomal dominant	Affected	Chr 17: 41960344	G	A	NA	NA	missense	T172I	0.173	benign	unknown	none
GTF3C2	Autosomal dominant	Affected	Chr 2: 27552024	T	C	NA	NA	missense	N668S	0.002	benign	transcription complex component	none
HECW2	Autosomal dominant	Affected	Chr 2: 197187284	C	A	NA	NA	missense	A268S	0.414	benign	E3 ubiquitin ligase	none
FAM83F	Autosomal dominant	Affected	Chr 22: 40417371	C	T	NA	NA	missense	T286M	0.950	possibly damaging	unknown	none
RNF40	Autosomal dominant	Affected	Chr 16: 30780841	C	T	NA	NA	nonsense	R836*	NA	NA	E3 ubiquitin ligase complex component	none
UGP2	Autosomal dominant	Affected	Chr 2: 64118276	A	C	NA	NA	missense	N481T	0.998	probably damaging	glucosyl donor	none
ZWILCH	Autosomal dominant	Affected	Chr 15: 66811324	C	T	NA	NA	missense	S143L	0.978	probably damaging	component of mitotic checkpoint	none
TTC3	Autosomal dominant	Affected	Chr 21: 38568067	T	A	NA	NA	missense	L1770H	0.681	possibly damaging	E3 ubiquitin ligase complex component	none
ZNF710	Autosomal dominant	Affected	Chr 15: 90622959	C	G	NA	NA	missense	F631L	0.038	benign	possible role in transcription regulation	none

A2ML1	Autosomal dominant	Affected	Chr 12: 8975800	G	C	NA	NA	missense	A29P	1.000	probably damaging	proteinase inhibitor	none
VPS13C	Autosomal dominant	Affected	Chr 15: 62253984	G	A	NA	NA	missense	R1238C	1.000	probably damaging	unknown	none
ELF5	Autosomal dominant	Affected	Chr 11: 34501833	G	A	NA	NA	missense	R244W	1.000	probably damaging	transcription factor in keratinocyte differentiation	none
IRF6	Autosomal dominant	Affected	Chr 1: 209961848	G	A	NA	NA	missense	R441C	0.134	benign	transcription factor in epidermal development	Popliteal pterygium syndrome 1; van der Woude syndrome
XPOT	Autosomal dominant	Affected	Chr 12: 64823883	A	G	NA	NA	missense	T598A	0.001	benign	nuclear export	none
DCTN2	Autosomal dominant	Affected	Chr 12: 57926769	T	C	NA	NA	missense	M258V	0.010	benign	chromosome alignment and microtubule alignment	none
LAMA1	Autosomal dominant	Affected	Chr 18: 7009319	C	T	NA	NA	missense	R1307Q	0.997	probably damaging	lamanin in extracellular matrix	none
COLQ	Autosomal dominant	Affected	Chr 3: 15498067	TG	T	NA	NA	frame shift	c.872_873insG	NA	NA	anchors acetylcholinesterase	Endplate acetylcholinesterase deficiency
ABHD15	Autosomal dominant	Affected	Chr 17: 27893425	T	C	NA	NA	missense	Y187C	0.004	benign	unknown	none
WDR6	Autosomal dominant	Affected	Chr 3: 49050602	T	G	NA	NA	missense	S545R	0.119	benign	cell growth suppression	none
LUZP2	Autosomal dominant	Affected	Chr 11: 24753719	T	C	NA	NA	missense	L79S	0.980	probably damaging	unknown	none
PLEC	Autosomal dominant	Affected	Chr 8: 144996248	G	A	NA	NA	missense	R2718W	0.740	possibly damaging	helps anchor various cytoskeletal elements	Epidermolysis bullosa simplex with pyloric atresia; Epidermolysis bullosa simplex, Ogna type; Muscular dystrophy with epidermolysis bullosa simplex; Limb-girdle muscular dystrophy
RIMS2	Autosomal dominant	Affected	Chr 8: 104897819	C	T	NA	NA	missense	S331L	0.000	benign	unknown	none
GAS6	Autosomal dominant	Affected	Chr 13: 114531579	A	G	NA	NA	missense	Y417H	0.130	benign	tyrosine kinase receptor ligand involved in various cell processes	none

POU6F2	Autosomal dominant	Affected	Chr 7: 39503980	G	C	NA	NA	missense	A591P	0.993	probably damaging	transcription factor involved in neuron differentiation	none
KRAS	Autosomal dominant	Affected	Chr 12: 25368389	A	AT	NA	NA	frame shift	NM_033360.2:c.556_557insT	NA	NA	GTPase	Cardiofaciocutaneous syndrome 2; Noonan syndrome 3
KIF14	Autosomal dominant	Affected	Chr 1: 200558395	T	TTTCCTG	NA	NA	codon change and codon insertion	c.3064_3065insTTCCTG	NA	NA	involved in cell division	none
UQCRC1	Autosomal dominant	Affected	Chr 3: 48638129	C	T	NA	NA	missense	V371I	0.001	benign	component of mitochondrial respiratory chain	none