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	Т	G	NA	NA	missense	K267N	0.046	benign	unknown	none
												receptor for	
												cell/cell signaling in	
												neural	
CELSR2	Autosomal D	Unaffected	Chr1: 109803819	т	С	NA	NA	missense	F1372L	0.998	probably damaging	development	none
												regulates integrin-	
												mediated signal	
ILK	Autosomal D	Unaffected	Chr11: 6631060	А	G	NA	NA	missense	N321S	0.115	benign	transduction	
												laminin for	
												extracellular	
LAMB4	Autosomal D	Unaffected	Chr7: 107706230	С	Т	NA	NA	missense	C938Y	1.000	probably damaging	interactions	none
												involved in Golgi	
GCC1	Autosomal D	Unaffected	Chr7: 127222636	Т	С	NA	NA	missense	K587R	0.001	benign	structure	none
												serine and	
												threonine	
SDS	Autosomal D	Unaffected	Chr12: 113836398	G	С	NA	NA	missense	A116G	0.004	benign	dehydratase	none
												cytochrome C	cytochrome c oxidase
COX6B1	Autosomal D	Unaffected	Chr19: 36145529	G	A	NA	NA	missense	E55K	0.143	benign	oxidase subunit	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
												receptor for IGF2	
												and other cell	
IGF2R	Autosomal D	Unaffected	Chr6: 160489364	С	Т	NA	NA	missense	P1400L	0.386	benign	signaling ligands	
												modifies alpha	
TTLL11	Autosomal D	Unaffected	Chr9: 124855120	С	G	NA	NA	missense	G193A	0.017	benign	tubulin	none
TMEM45B	Autosomal D	Unaffected	Chr11: 129728556	СТТ	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
													Dilated
												regulates molecular	cardiomyopathy 1HH;
								codon				chaperones and	Myofibrillar myopathy
BAG3	Autosomal D	Unaffected	Chr10: 121436295	CAGG	С	NA	NA	deletion	NA	NA	NA	inhibit apoptosis	6
												regulates	
												transcription and	
WAC	Autosomal L	Unaffected	Chr10: 28878666	C	T	NA	NA	missense	P128L	0.000	benign	RNA processing	none
												c c	Amelogenesis
			Ch		-				12041	0.067		formation of tooth	Imperfecta type IB
ENAM	Autosomal L	Unaffected	Chr4: /150/984	A	 	NA	NA	missense	N281Y	0.967	probably damaging	enamel	and IC
1 KIM14	Autosomal D	Unaffected	Cnr9: 100850210	A	1	NA	NA	Imissense	C2915	0.801	possibly damaging	unknown	none
						1						cnorionic	
6604		11	Ch						6275	1 000		gonadotropin	
ICGB1	Autosomal L	Unaffected	JCnr19: 49539490	C	A	INA	INA	Imissense	10271	1.000	probably damaging	Isupunit	Inone

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

	Autosomal												
A2ML1	dominant	Affected	Chr 12: 8975800	G	С	NA	NA	missense	A29P	1.000	probably damaging	proteinase inhibitor	none
	Autosomal												
VPS13C	dominant	Affected	Chr 15: 62253984	G	А	NA	NA	missense	R1238C	1.000	probably damaging	unknown	none
												transcription factor	
	Autosomal											in keratinocyte	
ELF5	dominant	Affected	Chr 11: 34501833	G	A	NA	NA	missense	R244W	1.000	probably damaging	differentiation	none
												transcription factor	Popliteal pterygium
	Autosomal											in epidermal	syndrome 1; van der
IRF6	dominant	Affected	Chr 1: 209961848	G	A	NA	NA	missense	R441C	0.134	benign	development	Woude syndrome
	Autosomal												
ХРОТ	dominant	Affected	Chr 12: 64823883	А	G	NA	NA	missense	T598A	0.001	benign	nuclear export	none
												chromosome	
												alignment and	
	Autosomal											microtubule	
DCTN2	dominant	Affected	Chr 12: 57926769	Т	С	NA	NA	missense	M258V	0.010	benign	alignment	none
	Autosomal											lamanin in	
LAMA1	dominant	Affected	Chr 18: 7009319	с	Т	NA	NA	missense	R1307Q	0.997	probably damaging	extracellular matrix	none
												anchors	Endplate
	Autosomal								c.872_873i	i		acetylcholinesteras	acetylcholinesterase
COLQ	dominant	Affected	Chr 3: 15498067	TG	Т	NA	NA	frame shift	nsG	NA	NA	e	deficiency
	Autosomal												
ABHD15	dominant	Affected	Chr 17: 27893425	Т	с	NA	NA	missense	Y187C	0.004	benign	unknown	none
	Autosomal											cell growth	
WDR6	dominant	Affected	Chr 3: 49050602	Т	G	NA	NA	missense	S545R	0.119	benign	suppression	none
	Autosomal												
LUZP2	dominant	Affected	Chr 11: 24753719	Т	С	NA	NA	missense	L79S	0.980	probably damaging	unknown	none
													Epidermolysis bullosa
													simplex with pyloric
													atrosia: Enidermolysis
													bullosa simpley. Ogna
													bullosa simplex, Oglia
													type; Muscular
													dystrophy with
												helps anchor	epidermolysis bullosa
	Autosomal											various cytoskeleta	l simplex; Limb-girdle
PLEC	dominant	Affected	Chr 8: 144996248	G	A	NA	NA	missense	R2718W	0.740	possibly damaging	elements	muscular dystrophy
	Autosomal												
RIMS2	dominant	Affected	Chr 8: 104897819	С	Т	NA	NA	missense	S331L	0.000	benign	unknown	none
												tyrosine kinase	
												receptor ligand	
	Autosomal											involved in various	
GAS6	dominant	Affected	Chr 13: 114531579	A	lG	NA	NA	lmissense	Y417H	0.130	lbenign	cell processes	Inone

												transcription factor	
	Autosomal											involved in neuron	
POU6F2	dominant	Affected	Chr 7: 39503980	G	С	NA	NA	missense	A591P	0.993	probably damaging	differentiation	none
									NM_0333				Cardiofaciocutaneous
	Autosomal								60.2:c.556				syndrome 2; Noonan
KRAS	dominant	Affected	Chr 12: 25368389	А	AT	NA	NA	frame shift	_557insT	NA	NA	GTPase	syndrome 3
								codon					
								change and	c.3064_30				
	Autosomal							codon	65insTTCC			involved in cell	
KIF14	dominant	Affected	Chr 1: 200558395	Т	TTTCCTG	NA	NA	insertion	TG	NA	NA	division	none
												component of	
	Autosomal											mitochondrial	
UQCRC1	dominant	Affected	Chr 3: 48638129	c	Т	NA	NA	missense	V371I	0.001	benign	respiratory chain	none