Supplementary Table 3

Autosomal dominant variants in family P04: The table lists all variants that passed filters as described in Methods and demonstrated an autosomal dominant mode of inheritance. A sibling status of "Affected" indicates that the heterozygous variant is shared by the father, patient, and sibling. A sibling status of "Unaffected" indicates that the heterozygous variant is shared by the father and patient only. All other annotations are the same as Supplementary Table 2.

Patient 04 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)
CPXM2	Autosomal dominant	Unaffected	Chr 10: 125506395	C	T	NA	NA	missense	S719N	0.006	benign	unknown	none
MLL3	Autosomal dominant	Unaffected	Chr 7: 151878026	T	C	NA	NA	missense	R2307G	0.262	benign	unknown	none
RALGPS1	Autosomal dominant	Unaffected	Chr 9: 129812366	A	T	NA	NA	missense	L115F	1.000	probably damaging	guanine nucleotide exchange factor	none
PLEKHG1	Autosomal dominant	Unaffected	Chr 6: 151161760	C	T	NA	NA	missense	H1296Y	0.229	benign	unknown	none
HIST1H1A	Autosomal dominant	Unaffected	Chr 6: 26017679	T	TCC	NA	NA	frame shift	c.282_283insCC	NA	NA	histone protein	none
DBF4B	Autosomal dominant	Unaffected	Chr 17: 42828020	T	C	NA	NA	missense	M416T	0.000	benign	role in DNA replication and cell proliferation	none
BAIAP2L2	Autosomal dominant	Unaffected	Chr 22: 38484752	C	CA	NA	NA	splice site	c.1118+3_4insA	NA	NA	cell membrane formation	none
GTPBP1	Autosomal dominant	Unaffected	Chr 22: 39112035	G	A	NA	NA	missense	R143H	0.115	benign	mRNA degradation	none
ATP11B	Autosomal dominant	Unaffected	Chr 3: 182559873	C	G	NA	NA	missense	R223G	1.000	probably damaging	P-type ATPase ion transporter	none
GLS2	Autosomal dominant	Unaffected	Chr 12: 56868396	C	G	NA	NA	missense	V386L	0.994	probably damaging	unknown	none
THOC3	Autosomal dominant	Unaffected	Chr 5: 175395149	C	T	NA	NA	missense	M21I	0.001	benign	functions in mRNA transport	none
WNT9B	Autosomal dominant	Unaffected	Chr 17: 44952729	C	G	NA	NA	missense	I199M	0.689	possibly damaging	signaling molecule in tissue development	none
PDE3A	Autosomal dominant	Unaffected	Chr 12: 20766482	T	A	NA	NA	missense	S373T	0.671	possibly damaging	phosphodiesterase	none
OSMR	Autosomal dominant	Unaffected	Chr 5: 38876314	C	T	NA	NA	missense	R29C	0.876	possibly damaging	component of IL31 receptor, activates downstream STAT proteins	Primary localized cutaneous amyloidosis 1
KIAA0141	Autosomal dominant	Unaffected	Chr 5: 141314152	G	A	NA	NA	splice site	c.1149+1G>A	NA	NA	unknown	none
KAT6A (MYST	Autosomal dominant	Unaffected	Chr 8: 41800361	C	T	NA	NA	missense	E796K	0.181	benign	histone acetyltransferase	none

MYPN	Autosomal dominant	Unaffected	Chr 10: 69905256	G	A	NA	NA	missense	G368D	0.993	probably damaging	sarcomere component	Dilated cardiomyopathy 1KK, familial restrictive cardiomyopathy 4, familial hypertrophic cardiomyopathy 22
LMTK2	Autosomal dominant	Affected	Chr 7: 97822515	G	C	NA	NA	missense	S913T	0.094	benign	kinase	none
DTWD1	Autosomal dominant	Affected	Chr 15: 49917564	TCTA	T	NA	NA	codon deletion	c.201_203 delCTA	NA	NA	unknown	none
FUT2	Autosomal dominant	Affected	Chr 19: 49206910	C	T	NA	NA	missense	R233C	0.403	benign	fucosyltransferase	none
TTC31	Autosomal dominant	Affected	Chr 2: 74710253	A	T	NA	NA	missense	I11F	0.917	possibly damaging	unknown	none
DLL4	Autosomal dominant	Affected	Chr 15: 41227163	C	T	NA	NA	missense	T363I	0.001	benign	angiogenesis	none
WDR53	Autosomal dominant	Affected	Chr 3: 196288144	T	C	NA	NA	missense	Y68C	0.998	probably damaging	unknown	none
FLRT2	Autosomal dominant	Affected	Chr 14: 86087932	G	A	NA	NA	missense	G25E	0.965	probably damaging	cell adhesion and receptor signaling	none
NPR2	Autosomal dominant	Affected	Chr 9: 35800429	G	T	NA	NA	missense	E389D	0.000	benign	natriuretic peptide receptor	Maroteaux type acromesomelic dysplasia
CC2D1A	Autosomal dominant	Affected	Chr 19: 14037355	G	A	NA	NA	missense	D656N	0.120	benign	neuronal transcription factor	Autosomal recessive mental retardation 3
BAZ2B	Autosomal dominant	Affected	Chr 2: 160194190	C	T	NA	NA	missense	E1850K	0.366	benign	transcriptional regulation	none
RFC5	Autosomal dominant	Affected	Chr 12: 118469082	A	AGAT	NA	NA	codon insertion	c.959_960i nsGAT	NA	NA	DNA replication factor	none
LYPLA2	Autosomal dominant	Affected	Chr 1: 24121184	G	A	NA	NA	missense	V220M	0.795	possibly damaging	hydrolyze fatty acid moeities	none
SGTA	Autosomal dominant	Affected	Chr 19: 2767667	C	T	NA	NA	missense	E40K	0.998	probably damaging	protein chaperone	none
ZNF774	Autosomal dominant	Affected	Chr 15: 90903968	A	G	NA	NA	missense	Q302R	0.014	benign	possible role in transcription regulation	none
MUC16	Autosomal dominant	Affected	Chr 19: 9073879	C	T	NA	NA	missense	A4523T	0.001	benign	mucin	none
NR2F6	Autosomal dominant	Affected	Chr 19: 17356004	C	A	NA	NA	missense	G9V	0.082	benign	transcription repression	none
DHX33	Autosomal dominant	Affected	Chr 17: 5359481	T	C	NA	NA	missense	I291V	0.518	possibly damaging	transcription of rRNA	none
MUC16	Autosomal dominant	Affected	Chr 19: 9072538	C	A	NA	NA	missense	A4970S	0.087	benign	mucin	none

SERINC2	Autosomal dominant	Affected	Chr 1: 31906004	G	A	NA	NA	missense	V403I	0.002	benign	unknown	none
PAH	Autosomal dominant	Affected	Chr 12: 103249019	G	A	NA	rs62517205	missense	H201Y	0.943	possibly damaging	phenylalanine hydroxylase	phenylketonuria
MYH10	Autosomal dominant	Affected	Chr 17: 8424573	G	C	NA	NA	missense	T632S	0.001	benign	myosin heavy chain	none
AFAP1L1	Autosomal dominant	Affected	Chr 5: 148699238	G	A	NA	NA	missense	V525M	1.000	probably damaging	cell projection formation	none
PIGS	Autosomal dominant	Affected	Chr 17: 26881275	G	C	NA	NA	missense	T544S	0.000	benign	transfer of phosphatidylinositol-glycan membrane anchors	none
AP2A1	Autosomal dominant	Affected	Chr 19: 50298969	G	A	NA	NA	missense	R263Q	1.000	probably damaging	vescicle transport	none
FCGBP	Autosomal dominant	Affected	Chr 19: 40364055	G	A	NA	NA	missense	H4863Y	0.987	probably damaging	maintenance of mucosa	none
HTR6	Autosomal dominant	Affected	Chr 1: 20005569	G	A	NA	NA	missense	R344H	0.000	benign	serotonin receptor	none
KIR2DL1	Autosomal dominant	Affected	Chr 19: 55285012	T	G	NA	NA	missense	C100G	1.000	probably damaging	unknown	none
HS3ST3A1	Autosomal dominant	Affected	Chr 17: 13503872	T	C	NA	NA	missense	Y192C	1.000	probably damaging	heparan sulfate sulfotransferase	none
CERKL	Autosomal dominant	Affected	Chr 2: 182521519	G	A	NA	NA	missense	P72L	0.197	benign	regulation of apoptosis	none
RAI1	Autosomal dominant	Affected	Chr 17: 17698661	G	A	NA	NA	missense	G800E	0.22	benign	transcriptional regulator	Smith-magenis syndrome