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Supplemental Material to:

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A human de novo mutation in *MYH10* phenocopies the loss of function mutation in mice

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Supplementary tables:

Table S1: Bioinformatics Variant Filtering

Stepwise Filtering[1]	**Proband**	**Father**	**Mother**	**Average**
No. of variants in coding regions[2]	118,337	125,974	126,536	**123,616**
No.post-removal of intergenic and 3'/5' UTR variants	86,497	88,687	88,911	**88,032**
No. post-removal of non-splice-related intronic[3] variants	22,380	23,255	23,483	**23,039**
No. post-removal of synonymous variants	11,926	12,353	12,590	**12,290**

[1]Stepwise filtering protects variants annotated within the Human Gene Mutation Database (HGMD) and/or the Online Mendelian Inheritance in Man (OMIM) databases.
[2]Variants refers to single nucleotide alterations, insertions, deletions, and indels with at least 10x base pair coverage.
[3]Intronic refers to >3 bp into the introns

Table S2: Variant Filtering based on the inheritance model and interpretation

	Inheritance Model Filtering	**Manual Review[1]**	**Notable Candidate Genes[‡]**
Autosomal Dominant Genes (Alterations)	4 (4)	3 (3)	**1 (1)**
Autosomal Recessive Genes (Alterations)	6 (10)	4 (7)	**0 (0)**
X-linked Recessive Genes (Alterations)	5 (5)	4 (4)	**1 (1)**
X-linked Dominant Genes (Alterations)	0 (0)	0 (0)	**0 (0)**
Y-linked Genes (Alterations)	0 (0)	0 (0)	**0 (0)**
TOTAL GENES (Alterations)	**15 (19)**	**11 (14)**	**2 (2)**

[1]Manual filtering involves the removal of genes unrelated to the patient's evaluated phenotype and alterations considered benign
[2]Notable Candidate Genes: Genes with disease phenotype association overlapping that of the proband

Table S3: Summary of the variants remaining after manual filtering

Gene Symbol/ Model	Novel/ Characterized Gene	Variant	Genotype			SIFT	dbSNP	Allele Frequency	
			Proband	Father	Mother			ESP	1000 Genomes
Autosomal Dominant									
MYH10	Novel	c.2722G>T (p.E908X)	+/-	-/-	-/-		N/A		
PLCB3	Novel	c.703G>C (p.A235P)	+/-	-/-	-/-	tolerated	N/A	N/A	N/A
ZNF705G	Novel	c.530G>A (p.C177Y)	+/-	-/-	-/-	damaging	N/A	N/A	0.05%
Autosomal Recessive									
CRIPAK	Novel	c.755_756insCA (p.L252PfsX178)	+/-	-/-	-/-				
		c.937_938insCG (p.S313RfsX117)	+/-	-/-	-/-				
KLHDC7A	Novel	c.46C>A (p.Q16K)	+/-	-/-	+/-	damaging	rs140809384	0.17%	0.09%
		c.493C>T (p.P165S)	+/-	+/-	-/-	tolerated	N/A	N/A	N/A
PLCB3	Novel	c.703G>C (p.A235P)	+/-	-/-	-/-	tolerated	N/A	N/A	N/A
		c.1301G>A (p.G434E)	+/-	+/-	-/-	damaging	N/A	N/A	N/A
EPSTI1	Novel	c.1184_1185insCCTGA (p.E395AfsX2)	+/+	+/-	+/-		rs111313375		
X-linked recessive									
DOCK11	Novel	c.5315C>T p.S1772F	+/+	-/-	+/-	tolerated	N/A	N/A	N/A
ODZ1	Novel	c.5093G>A p.R1698H	+/+	-/-	+/-	tolerated	rs140984539	0.03%	0.14%
ZNF630	Novel	c.568G>T p.D190Y	+/+	-/-	+/-	damaging	N/A	N/A	N/A
BCOR	Characterized	c.626C>T p.S209L	+/+	-/-	+/-	tolerated	rs143697110	0.06%	N/A

N/A = not available; blank boxes indicate not applicable