

Appendix Table A1. Identified variants. List of all variants identified in the study population and the percentage of each variant in the general population across the Broad Institute Exome Aggregation Consortium (Broad ExAC).

Variants Identified		Percentage in general population*
ABCC6	Deletion of Exon 23-29	N/A
ABCC6	c.1283A>G p.Asn428Ser	0.0001
ABCC6	c.3979G>A p.Gly1327Arg	0.0003
ADAMTS2	c.1883G>A p.Arg628His	0.0012
ADAMTS2	c.2015G>T p.Arg672Leu	0.0001
ATP6V0A2	c.776G>A p.Arg259Gln	<.01%
B4GALT7	c.564G>A p.Pro188Pro	0.0001
BMP1	c.2134G>A p.Gly712Ser	0.001
CBS	c.349C>T p.Arg132Cys	0.0003
CBS	c.1105C>T p.Arg369Cys	0.0033
COL1A1	c.3766G>A p.Ala1256Thr	0.0003
COL1A2	c.2711G>C p.Gly904Ala	N/A
COL1A2	c.2782-19T>G p.?	<.01%
COL11A1	c.560C>T p.Thr187Met	0.0003
COL12A1	c.3652G>A p.Val1218Met	0.0003
COL12A1	c.5369C>G p.Ser1790Cys	>.01%
COL12A1	c.6080G>A p.Gly2027Glu	>.01%
COL12A1	c.7813A>C p.Lys2605Gln	>.01%
COL5A2	c.4067A>G p.Asp1356Gly	0.0003
COL6A1	c.2809A>G p.Lys937Glu	0.0006
COL6A2	c.1661A>G p.Lys554Arg	0.0002
COL6A2	c.2254G>A p.Val752Ile	>0.01%
COL6A2	c.2950G>A p.Val984Met	>0.01%
COL6A3	c.3205G>A p.Val1069Met	0.0004
COL6A3	c.7447A>G p.Lys2483Glu	0.0006
COL6A3	c.9245C>G p.Pro3082Arg	0.001
DSE	c.1004G>A p.Arg335His	>.01%
FBN1	c.4437C>G p.Asp1479Glu	>.01%
FBN2	c.197A>G p.Glu66Gly	Never Reported
NOTCH1	c.383G>A p.Arg128His	>.01%
PRDM5	c.46C>A p.Arg16Arg	>.01%
PRDM5	c.1633T>C p.Tyr545His	0.0009
RYR1	c.12813C>T p.Gly4271Gly	>.01%

RYR1	c.13934G>A p.Arg4645Gln	>.01%
RYR1	c.14204G>A p.Arg4735Gln	>.01%
RYR1	c.2511C>A p.His837Gln	Never Reported
RYR1	c.6961A>G p.Ile2321Val	0.0004
RYR1	c.7115G>C p.Gly2372Ala	>.01%
SLC2A10	c.1324G>C p.Glu442Gln	>.01%
SLC2A10	c.251T>C p.Val84Ala	Never Reported
SMAD3	c.91T>A p.Cys31Ser	Never Reported
TGFBR1	c.883T>A p.Tyr295Asn	Never Reported
TNC	c.4243G>A p.Val1415Ile	>.01%
TNC	c.5317G>A p.Val1773Met	>.01%
TNXB	c.2387C>T p.Ala796Val	Never Reported
TNXB	c.3277G>A p.Val1093Met	0.0003
TNXB	c.11056C>G p.Arg3686Gly	>.01%
TTN	c.5740G>A p.Ala1914Thr	0.0001
TTN	c.13040G>T p.Arg4347Ile	>.01%
TTN	c.18844G>T p.Val6282Phe	>.01%
TTN	c.24115G>A p.Val8039Met	0.0002
TTN	c.41709G>T p.Trp13903Cys	0.0004
TTN	c.42910G>C p.Ala14304Pro	>.01%
TTN	c.45149G>A p.Arg15050His	0.0002
TTN	c.46400C>T p.Ala15467Val	>0.01%
TTN	c.49207G>A p.Val16403Met	>0.01%
TTN	c.51614A>G p.Glu17205Gly	0.0001
TTN	c.51887G>A p.Gly17296Asp	>.01%
TTN	c.52282C>G p.Leu17428Val	Never Reported
TTN	c.52442G>A p.Arg17481His	0.0001
TTN	c.58726G>A p.Ala19576Thr	0.0002
TTN	c.64277C>T p.Ala21426Val	>.01%
TTN	c.70003G>A p.Val23335Ile	0.0002
TTN	c.74984G>A p.Arg24995His	0.0002
TTN	c.80425C>T p.Leu26809Phe	Never Reported
TTN	c.80702C>T p.Ala26901Val	0.0001
TTN	c.81016C>T p.Arg27006Cys	0.0003
TTN	c.88685C>T p.Thr29562Met	>.01%
TTN	c.95047A>G p.Met31683Val	0.0007
ZNF469	c.10204del p.Arg3414Glyfs*59	>.01%
ZNF469	c.1913C>T p.Pro638Leu	0.0004
ZNF469	c.2270T>G p.Leu757Arg	0.0003
ZNF469	c.2653C>G p.Leu885Val	0.0013
ZNF469	c.2803G>A p.Glu935Lys	0.0024
ZNF469	c.3242C>T p.Pro1081Leu	>.01%

ZNF469	c.4825C>T p.Arg1609Trp	0.0007
ZNF469	c.523G>A p.Ala175Thr	Never Reported
ZNF469	c.8558G>A p.Arg2853His	0.0005
ZNF469	c.891C>G p.Phe297Leu	>.01%
ZNF469	c.10937C>T p.Ala3646Val	>.01%
ZNF469	c.11341G>A p.Glu3781Lys	0.0003

*observed frequency across entire Broad ExAC dataset (>60,000 individuals); N/A= not available