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SUPPLEMENTAL DATA

The usefulness of the genetic panel in the classification and refinement of diagnostic accuracy of Mexican patients with Marfan syndrome and other connective tissue disorders

Table S1. Considerations of the surgical group for the decision of the surgical technique

Valve	Aortic ring	Sinotubular junction	Surgical technique
Affected (advanced age)	Normal	Normal	Supracoronary replacement + valve change
Affected (young patient)	Normal	Affected	Bentall-De Bono or Ross
Normal	Normal	Affected	Remodeling (Yacoub/David)
Normal	Affected	Affected	Bentall-De Bono or Remodeling + ring plasty.
Endocarditis	Normal	Normal or affected	Bentall-De Bono

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Table S2. Dataset

ID	Diagnosis	Gen 1	Zygote	Position	Position + Exon	Mutation type	Severity	Gen 2	Zygote	Position 3	Position + Exon 4
339	Beals Hecht	*ANK2*	Hetero-zygous	rs1396114703	NM_015141.4(GPD1L):c.995A>C (p.Tyr332Ser) Exon: 15/46	Frame-shift Indels	Likely patho-genic	*FBN2*	Hetero-zygous	rs2291628	NM_001999.3c.7739C>Tp.(Ser2580Leu)Exon: 61/65
299	Beals Hecht	*FKRP*	Hetero-zygous	rs28937900	NM_024301.5(FKRP):c.826C>A (p.Leu276Ile) Exon 4/4	Stop codon	Likely patho-genic	*FBN2*	Homo-zygous	rs154001	NM_001999.3c.2893G>Ap.(Val965Ile)Exon: 23/65
415	Beals Hecht	*PRMD16*	Hetero-zygous	rs572178955	NM_022114.3c.3221C>T(p.Ser1074Leu)Exon: 14/17	Missense	Likely patho-genic	*FBN2*	Hetero-zygous	rs32209	NM_001999.3c.6931A>G(p.Met2311Val)Exon: 55/65
306	Beals Hecht	*DSP*	Hetero-zygous	rs771721410	NM_004415.4(DSP):c.4466T>G (p.Leu1489Arg) Exon: 23/24	Frame-shift Indels	Likely patho-genic	*FBN2*	Homo-zygous	rs154001	NM_001999.3c.2893G>A(p.Val965Ile)Exon: 23/65
414	Beals Hecht	*FBN2*	Hetero-zygous	rs780589159	NM_001999.4(FBN2):c.1961G>A (p.Arg654His) Exon: 14/65	Frame-shift Indels	Likely patho-genic				
247	Beals Hecht	*SOS1*	Hetero-zygous		NM_005633.3c.3724del(p.Ser1242ValfsTer37) Exon: 23/23	Missense	Likely patho-genic	*FBN2*	Homo-zygous	rs154001	NM_001999.3c.2893G>A(p.Val965Ile)Exon: 23/65
357	Beals Hecht	*APOB*	Hetero-zygous		NM_007294.4(BRCA1):c.5365G>T(p.Ala1789Ser) Exon: 14/29	Frame-shift Indels	Likely patho-genic	*FBN2*	Hetero-zygous	s190450	NM_001999.3c.7200T>Cc.7200T>C(p.(Ser2400=))Exon: 57/65
279	Beals Hecht	*LDB3*	Hetero-zygous	rs45487699	NM_007078.3(LDB3):c.566C>T (p.Ser189Leu) Exon: 5/14	Stop codon	Patho-genic	*FBN2*	Hetero-zygous	rs154001	NM_001999.3c.2893G>Ap.(Val965Ile)Exon: 23/65
359	Beals Hecht	*APOC4-APOC2*	Hetero-zygous	rs5122	NR_037932.1n.1385G>A Exon: 5/6	Stop codon	Patho-genic	*FBN2*	Homo-zyggous	rs154001	NM_001999.3c.2893G>A(p.Val965Ile)Exon: 23/65

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110	Beals Hecht	*TTN*	Hetero-zygous		33083_33086 (Tyr11029Ter) Exon: 135/363	Missense	Patho-genic	*FBN2*	Hetero-zygous	rs146849637	NM_001999.3c.829G>A(p.Val277Ile)Exon: 7/65
351	Beals Hecht	*TTN-AS1*	Hetero-zygous	rs68080670	NR_038271.1n.446+8403C>T Exon: 1/7	Missense	VUS	*FBN2*	Hetero-zygous	rs775848753	NM_001999.3c.7660T>Gp.(Phe2554Val)Exon: 60/65
177	Beals Hecht	*FBN2*	Hetero-zygous	rs757052151	NM_001999.3c.8705C>A (p.Ala2902Asp) Exon: 65/65	Missense	VUS				
293	Beals Hecht	*ALMS1*	Hetero-zygous		NM_015120.4c.72_74del(p.Glu29del) Exon: 1/23	Missense	VUS	*FBN2*	Hetero-zygous	rs2291628	NM_001999.3c.7739C>Tp.(Ser2580Leu)Exon: 61/65
196	Ehlers Danlos	*RYR2*	Hetero-zygous	rs749201569	NM_001035.2c.14217del (p.Phe4739LeufsTer15) Exon: 99/105	Frame-shift Indels	Likely patho-genic	*COL3A1*	Hetero-zygous	rs1800255	NM_000090.3c.2092G>A(p.Ala698Thr)Exon: 30/51
115	Ehlers Danlos	*APOE*	Hetero-zygous	rs769452	NM_000041.3(APOE):c.388T>C (p.Cys130Arg) Exon: 3/4	Frame-shift Indels	Likely patho-genic	*COL3A1*	Hetero-zygous	rs41263773	NM_000090.3c.2035G>A(p.Ala679Thr)Exon: 30/51
130	Ehlers Danlos	*MYLK*	Hetero-zygous	rs770373926	NM_053025.3c.472del(p.Glu158SerfsTer79) Exon: 7/34	Frame-shift Indels	Likely patho-genic	*COL3A1*	Hetero-zygous		NM_000090.3c.2744G>A(p.Gly915Asp)Exon: 39/51
215	Ehlers Danlos	*AKAP9*	Hetero-zygous	rs779447911	NM_005751.4c.4960_4961del(p.Arg1654GlyfsTer23) Exon: 19/50	Inframe deletion	Likely patho-genic	*COL5A2*	Hetero-zygous	rs555111281	NM_000393.3c.3826A>G(p.Ser1276Gly)Exon: 51/54
100	Ehlers Danlos	*APOB*	Hetero-zygous		NM_007294.4(BRCA1):c.5365G>T (p.Ala1789Ser) Exon: 14/29	Missense	Likely patho-genic	*COL5A2*	Hetero-zygous	rs35830636	NM_000393.4c.1378C>T(p.Pro460Ser)Exon: 21/54
397	Ehlers Danlos	*COL5A1*	Hetero-zygous		NM_000093.4c.4706del(p.Pro1569ArgfsTer50) Exon: 62/66	Stop codon	Patho-genic				
412	Ehlers Danlos	*COL5A2*	Hetero-zygous	rs376971803	NM_000393.3c.3733C>T(p.Pro1245Ser) Exon: 51/54	Synonymous	VUS				

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308	Ehlers Danlos	*DNAJC19*	Hetero-zygous	rs754317673	NM_145261.4(DNAJC19):c.202G>A (p.Gly68Ser)	Missense	VUS	*COL3A1*	Hetero-zygous	rs1516446	NM_000090.3c.4059=c.4059=(p.(Gln1353=))Exon: 50/51
318	Ehlers Danlos	*TTN*	Hetero-zygous		70258 C>A (Pro23420Thr) Exon: 326/363	Missense	VUS	*COL5A1*	Hetero-zygous	rs748610273	NR_103451.2n.71-4475C>TExon:
171	Ehlers Danlos	*COL5A2*	Hetero-zygous		NM_000393.3c.2355del(p.Gly786ValfsTer38) Exon: 35/54	Missense	VUS				
262	Loeys Dietz	*TGFBR1*	Hetero-zygous	rs1564161322	NM_001306210.1c.617C>T(p.Ala206Val) Exon: 4/9	Stop codon	Likely patho-genic				
178	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs34833812	NM_001024847.2c.1019C>T(p.Thr340Met) Exon: 5/8	Inframe deletion	Likely patho-genic				
165	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs79375991	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic	*FBN1*	Hetero-zigoto	rs397515762	NM_000138.4c.1837+1G>A Exon: 15/66
270	Loeys Dietz	*TGFB2*	Hetero-zygous	rs757551766	NM_001135599.2c.484G>A (p.Val162Ile) Exon: 3/8	Frame-shift Indels	Likely patho-genic				
410	Loeys Dietz	*TGFBR1*	Hetero-zygous	rs767589799	NM_004612.4(TGFBR1):c.1425G>T (p.Trp475Cys) Exon: 9/9	Frame-shift Indels	Likely patho-genic				
169	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs79375991	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
183	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs79375991	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				

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187	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
219	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
221	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
224	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
241	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
274	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
291	Loeys Dietz	*TGFBR2*		rs7937599 1	NM_001024847.2c.458del(p.Lys153SerfsTer35)Exon: 4/8	Frame-shift Indels	Likely patho-genic				
385	Loeys Dietz	*TGFBR2*	Hetero-zygous	rs7525801 04	NM_003242.5:c.383del (p.Lys128SerfsTer) Exon:	Missense	Patho-genic				
300	Loeys Dietz	*TGFBR1*	Hetero-zygous	rs8860389 19	NM_001306210.1c.769A>G(p.Met257Val) Exon: 4/9	Missense	Patho-genic				
311	Marfan	*FBN1*	Hetero-zygous	rs1121042 70	NM_000138.4c.5671+1G> A Exon: 46/66	Frame-shift	Likely patho-				

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						Indels	genic				
164	Marfan	*FBN1*	Hetero-zygous	rs112911555	NM_000138.4c.2861G>A(p.Arg954His) Exon: 25/66	Frame-shift Indels	Likely patho-genic				
123	Marfan	*FBN1*	Hetero-zygous	rs113812345	NM_000138.4c.1546C>T(p.Arg516Ter) Exon: 13/66	Stop codon	Likely patho-genic				
125	Marfan	*FBN1*	Hetero-zygous	rs113812345	NM_000138.4c.1546C>T(p.Arg516Ter) Exon: 13/66	Frame-shift Indels	Likely patho-genic				
239	Marfan	*FBN1*	Homo-zigous	rs137854466	NM_000138.4c.8326C>T(p.Arg2776Ter) Exon: 66/66	Missense	Likely patho-genic				
240	Marfan	*FBN1*	Homo-zigous	rs137854466	NM_000138.4c.8326C>T(p.Arg2776Ter) Exon: 66/66	Frame-shift Indels	Likely patho-genic				
363	Marfan	*FBN1*	Hetero-zygous	rs137854470	NM_000138.4c.3350G>A(p.Cys1117Tyr) Exon: 28/66	Frame-shift Indels	Likely patho-genic				
377	Marfan	*FBN1*	Hetero-zygous	rs140583	NM_000138.4c.2581C>T(p.Arg861Ter) Exon: 22/66	Frame-shift Indels	Likely patho-genic				
242	Marfan	*FBN1*	Hetero-zygous	rs1555394562	NM_000138.4c.7134C>G(p.Cys2378Trp) Exon: 58/66	Frame-shift Indels	Likely patho-genic				
248	Marfan	*FBN1*	Hetero-zygous	rs1555398974	NM_000138.4c.2849G>A(p.Cys950Tyr) Exon: 24/66	Frame-shift	Likely patho-				

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						Indels	genic				
260	Marfan	*FBN1*	Hetero-zygous	rs1555399963	NM_000138.4c.1760G>A(p.Cys587Tyr) Exon: 15/66	Frame-shift Indels	Likely patho-genic				
298	Marfan	*FBN1*	Hetero-zygous	rs1555401697	NM_000138.4c.577del(p.Gln193ArgfsTer137) Exon: 7/66	Frame-shift Indels	Likely patho-genic				
329	Marfan	*FBN1*	Hetero-zygous	rs1597533619	NM_000138.4c.5728G>T(p.Gly1910Cys) Exon: 47/66	Frame-shift Indels	Likely patho-genic				
296	Marfan	*FBN1*	Hetero-zygous	rs193922228	NM_000138.4c.6806T>C(p.Ile2269Thr) Exon: 56/66	Stop codon	Likely patho-genic				
265	Marfan	*FBN1*	Hetero-zygous	rs363815	NM_000138.4c.6331T>C(p.Cys2111Arg) Exon: 52/66	Stop codon	Likely patho-genic				
310	Marfan	*FBN1*	Hetero-zygous	rs397515848	NM_000138.4c.7180C>T(p.Arg2394Ter) Exon: 58/66	Frame-shift Indels	Likely patho-genic				
231	Marfan	*AKAP9*	Hetero-zygous	rs779447911	NM_005751.4c.4960_4961del(p.Arg1654GlyfsTer23) Exon: 19/50	Frame-shift Indels	Likely patho-genic	*FBN1*	Hetero-zigoto		NM_000138.4c.1427G>A(p.Cys476Tyr) Exon: 12/66
106	Marfan	*FBN1*	Hetero-zygous	rs794728162	NM_000138.4c.640G>A(p.Gly214Ser) Exon: 7/66	Non-coding exon	Likely patho-genic				
393	Marfan	*FBN1*	Hetero-zygous	rs794728228	NM_000138.4c.4621C>T(p.Arg1541Ter) Exon: 38/66	Stop codon	Likely patho-				

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							genic				
176	Marfan	*FBN1*	Hetero-zygous	rs794728335	NM_000138.4c.6425G>A(p.Cys2142Tyr) Exon: 53/66	Stop codon	Likely patho-genic				
371	Marfan	*GCKR*	Hetero-zygous	rs8179249	NM_001486.3c.1619G>A(p.Arg540Gln) Exon: 18/19	Missense	Likely patho-genic	*FBN1*	Hetero-zygous		NM_000138.4c.239G>Tp.(Cys80Phe)Exon: 3/66
347	Marfan	*FBN1*	Hetero-zygous	rs990134651	NM_000138.4c.3211A>G(p.Ile1071Val) Exon: 27/66	Missense	Likely patho-genic				
268	Marfan	*COL5A1*	Hetero-zygous		NM_000093.4c.4050del(p.Gly1351GlufsTer137) Exon: 51/66	Missense	Likely patho-genic	*FBN1*	Hetero-zygous	rs1555395189	NM_000138.4c.6491G>Ap.(Cys2164Tyr)Exon: 53/66
220	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.4383C>A(p.Cys1461Ter) Exon: 36/66	Frame-shift Indels	Likely patho-genic				
209	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.5846dup(p.Asn1949LysfsTer12) Exon: 48/66	Frame-shift Indels	Likely patho-genic				
144	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.6734G>A(p.Cys2245Tyr) Exon: 55/66	Stop codon	Likely patho-genic				
355	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.7178del(p.Gly2393AlafsTer5) Exon: 58/66	Stop codon	Likely patho-genic				
277	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.8516del(p.Lys2839ArgfsTer7) Exon: 66/66	Missense	Likely patho-				

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							genic				
379	Marfan	*FBN1*	Hetero-zygous	rs1057524458	NM_000138.4c.2945G>C(p.Cys982Ser) Exon: 25/66	Stop codon	Patho-genic				
331	Marfan	*FBN1*	Hetero-zygous	rs1060501055	NM_000138.4c.7525T>G(p.Cys2509Gly) Exon: 61/66	Splice donor	Patho-genic				
180	Marfan	*FBN1*	Hetero-zygous	rs1060501087	NM_000138.4c.5720A>G(p.Asn1907Ser) Exon: 47/66	Stop codon	Patho-genic				
161	Marfan	*FBN1*	Hetero-zygous	rs112911555	NM_000138.4c.2861G>A(p.Arg954His) Exon: 25/66	Splice donor	Patho-genic				
235	Marfan	*FBN1*	Hetero-zygous	rs113812345	NM_000138.4c.1546C>T(p.Arg516Ter) Exon: 13/66	Missense	Patho-genic				
350	Marfan	*FBN1*	Hetero-zygous	rs113812345	NM_000138.4c.1546C>T(p.Arg516Ter) Exon: 13/66	Missense	Patho-genic				
330	Marfan	*FBN1*	Homo-zigous	rs113936729	NM_000138.4c.1415=c.1415=(p.(Tyr472=)) Exon: 12/66	Missense	Patho-genic				
213	Marfan	*FBN1*	Hetero-zygous	rs140593	NM_000138.4c.3037G>A(p.Gly1013Arg) Exon: 25/66	Frame-shift Indels	Patho-genic				
345	Marfan	*FBN1*	Hetero-zygous	rs1555394406	NM_000138.4c.7376G>A(p.Cys2459Tyr) Exon: 60/66	Frame-shift Indels	Patho-genic				
361	Marfan	*FBN1*	Hetero-zygous	rs1555394406	NM_000138.4c.7376G>A(p.Cys2459Tyr) Exon: 60/66	Frame-shift Indels	Patho-genic				
153	Marfan	*FBN1*	Hetero-zygous	rs1555394562	NM_000138.4c.7134C>G(p.Cys2378Trp) Exon: 58/66	Inframe deletion	Patho-genic				
366	Marfan	*FBN1*	Hetero-	rs1555396	NM_000138.4c.5503T>C(p.Cys1835Arg)	Missense	Patho-				

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			zygous	198	Exon: 45/66		genic				
332	Marfan	*FBN1*	Hetero-zygous	rs397515830	NM_000138.4c.6119G>A(p.Cys2040Tyr) Exon: 50/66	Stop codon	Patho-genic				
356	Marfan	*FBN1*	Homo-zigous	rs794728165	NM_000138.4c.1090C>T(p.Arg364Ter) Exon: 10/66	Stop codon	Patho-genic				
365	Marfan	*FBN1*	Hetero-zygous	rs794728228	NM_000138.4c.4621C>T(p.Arg1541Ter) Exon: 38/66	Missense	Patho-genic				
204	Marfan	*FBN1*	Hetero-zygous	rs886041350	NM_000138.4c.1957_1958del(p.Val653Ter) Exon: 16/66	Missense	Patho-genic				
294	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.177T>A(p.Cys59Ter) Exon: 3/66	Stop codon	Patho-genic				
229	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.3697C>T(p.Gln1233Ter) Exon: 30/66	Missense	Patho-genic				
297	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.449G>T(p.Cys150Phe) Exon: 6/66	Missense	Patho-genic				
222	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.5742C>A(p.Cys1914Ter) Exon: 47/66	Stop codon	Patho-genic				
369	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.7684G>T(p.Gly2562Cys) Exon: 62/66	stop codon	Patho-genic				
276	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.8516del(p.Lys2839ArgfsTer7) Exon: 66/66	Missense	Patho-genic				
238	Marfan	*FBN1*	Hetero-zygous	rs113905529	NM_000138.4c.493C>T(p.Arg165Ter)Exon: 6/66	Missense	VUS				
124	Marfan	*FBN1*	Hetero-zygous	rs397515762	NM_000138.4c.1837+1G>A Exon: 15/66	Inframe deletion	VUS				
334	Marfan	*FBN1*	Hetero-	rs3975158	NM_000138.4c.6119G>A(p.Cys2040Tyr)	Missense	VUS				

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			zygous	30	Exon: 50/66						
333	Marfan	*FBN1*	Hetero-zygous	rs794728165	NM_000138.4c.1090C>T(p.Arg364Ter) Exon: 10/66	Missense	VUS				
203	Marfan	*FBN1*	Hetero-zygous	rs794728319	NM_000138.4c.7039_7040del(p.Met2347ValfsTer19) Exon: 58/66	Missense	VUS				
326	Marfan	*FBN1*	Hetero-zygous	rs794728335	NM_000138.4c.6425G>A(p.Cys2142Tyr) Exon: 53/66	Missense	VUS				
390	Marfan	*FBN1*	Hetero-zygous	rs794728335	NM_000138.4c.6425G>A(p.Cys2142Tyr) Exon: 53/66	Missense	VUS				
295	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.1427G>A(p.Cys476Tyr) Exon: 12/66	Missense	VUS				
225	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.2238T>A(p.Tyr746Ter) Exon: 19/66	Missense	VUS				
309	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.4970_4984del(p.Ile1657_Gly1661del) Exon: 41/66	Missense	VUS				
302	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.5988C>G(p.Tyr1996Ter) Exon: 49/66	Missense	VUS				
141	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.6734G>A(p.Cys2245Tyr) Exon: 55/66	Missense	VUS				
244	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.6772T>A(p.Cys2258Ser) Exon: 56/66	Inframe deletion	VUS				
374	Marfan	*FBN1*	Hetero-zygous		NM_000138.4c.8047C>T(p.Gln2683Ter) Exon: 64/66	Missense	VUS				
286	NCTD	*RYR1*	Hetero-zygous	rs1064794572	NM_000540.2c.14770_14772del(p.Phe4924del) Exon: 102/106	Frame-shift Indels	Likely patho-genic				

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250	NCTD	*SDHA*	Hetero-zygous	rs112307877	NM_004168.3c.1945_1946del(p.Leu649GlufsTer4) Exon: 15/15	Missense	Likely patho-genic				
256	NCTD	*SDHA*	Hetero-zygous	rs112307877	NM_004168.3c.1945_1946del(p.Leu649GlufsTer4) Exon: 15/15	Missense	Likely patho-genic				
349	NCTD	*ZBTB17*		rs138897393	NM_001287603.1c.317C>T(p.Thr106Met) Exon: 4/16	Frame-shift Indels	Likely patho-genic				
264	NCTD	*ZHX3*	Hetero-zygous	rs150841576	NM_015035.3c.233C>T(p.Ser78Phe) Exon: 3/4	Frame-shift Indels	Likely patho-genic				
173	NCTD	*HFE*	Heterozygous	rs1800562	NM_000410.3c.845G>A(p.Cys282Tyr) Exon: 4/6	Stop codon	Likely patho-genic				
343	NCTD	*TNNT2*	Hetero-zygous	rs200754249	NM_000364.3(TNNT2):c.113C>T (p.Ala38Val) Exon: 6/16	Frame-shift Indels	Likely patho-genic				
375	NCTD	*TTN-AS1*	Hetero-zygous	rs534881266	NR_038271.1n.447-22390C>T Exon: 1/7	Frame-shift Indels	Likely patho-genic				
228	NCTD	*MYBPC3*	Hetero-zygous	rs727504288	NM_000256.3c.2441_2443del (p.Lys814del) Exon: 25/35	Frame-shift Indels	Likely patho-genic				
280	NCTD	*MYBPC3*	Hetero-zygous	rs727504288	NM_000256.3c.2441_2443del (p.Lys814del) Exon: 25/35	Missense	Likely patho-genic				

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122	NCTD	*RYR2*	Hetero-zygous	rs749201569	NM_001035.2c.14217del (p.Phe4739LeufsTer15) Exon: 99/105	Frame-shift Indels	Likely patho-genic				
271	NCTD	*RYR2*	Hetero-zygous	rs749201569	NM_001035.2c.14217del (p.Phe4739LeufsTer15) Exon: 99/105	Frame-shift Indels	Likely patho-genic				
179	NCTD	*DES*	Hetero-zygous	rs769096434	NM_001927.3c.525_526del(p.Val176ArgfsTer48) Exon: 1/9	Frame-shift Indels	Likely patho-genic				
198	NCTD	*DES*	Hetero-zygous	rs769096434	NM_001927.3c.525_526del(p.Val176ArgfsTer48) Exon: 1/9	Frame-shift Indels	Likely patho-genic				
289	NCTD	*DES*	Hetero-zygous	rs769096434	NM_001927.3c.525_526del(p.Val176ArgfsTer48) Exon: 1/9	Frame-shift Indels	Likely patho-genic				
230	NCTD	*SMAD3*	Hetero-zygous		NM_005902.3c.205A>Cc.205A>C(p.(Arg69=)) Exon: 1/9	Missense	Likely patho-genic				
237	NCTD	*SDHA*	Hetero-zygous	rs112307877	NM_004168.3c.1945_1946del(p.Leu649GlufsTer4) Exon: 15/15	Missense	Patho-genic				
214	NCTD	*SDHA*	Hetero-zygous	rs112307877	NM_004168.3c.1945_1946del(p.Leu649GlufsTer4) Exon: 15/15	Missense	Patho-genic				
236	NCTD	*RYR1*	Hetero-zygous	rs1375580585	NM_000540.2c.4495T>G(p.Phe1499Val) Exon: 31/106	Frame-shift Indels	Patho-genic				
208	NCTD	*TNNT2*	Hetero-zygous	rs397516470	NM_000364.3c.517_519del(p.Glu173del) Exon: 12/16	Stop codon	Patho-genic				

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199	NCTD	*AKAP9*	Hetero-zygous	rs779447911	NM_005751.4c.4960_4961del(p.Arg1654GlyfsTer23) Exon: 19/50	Inframe deletion	Patho-genic				
272	NCTD	*AKAP9*	Hetero-zygous	rs779447911	NM_005751.4c.4960_4961del(p.Arg1654GlyfsTer23) Exon: 19/50	Frame-shift Indels	Patho-genic				
275	NCTD	*KCNH2*	Hetero-zygous	rs794728455	NM_000238.3c.2775del(p.Pro926ArgfsTer48) Exon: 12/15	Missense	Patho-genic				
223	NCTD	*SGCB*	Hetero-zygous		NM_000232.4c.552T>A(p.Tyr184Ter) Exon: 4/6	Missense	Patho-genic				
193	NCTD	*TPM1*	Hetero-zygous		NM_001018005.1c.230del(p.Lys77ArgfsTer9) Exon: 2/10	Missense	Patho-genic				
288	NCTD	*ACTN2*	Hetero-zygous		NM_001103.3c.674T>C(p.Ile225Thr) Exon: 7/21	Missense	Patho-genic				
273	NCTD	*SOS1*	Hetero-zygous		NM_005633.3c.3724del(p.Ser1242ValfsTer37) Exon: 23/23	Stop codon	Patho-genic				
233	NCTD	*NOTCH1*	Hetero-zygous	rs761020817	NM_017617.4c.4732_4734del (p.Val1578del) Exon: 26/34	Missense	VUS				
257	NCTD	*AKAP9*	Hetero-zygous	rs779447911	NM_005751.4c.4960_4961del(p.Arg1654GlyfsTer23) Exon: 19/50	Missense	VUS				
170	NCTD	*HADHA*	Hetero-zygous		NM_000182.4c.658C>T(p.Gln220Ter) Exon: 7/20	Missense	VUS				
413	NCTD	*TTN*	Hetero-zygous		NM_001267550.2c.91097_91100del(p.Arg30366IlefsTer24) Exon: 335/363	Missense	VUS				
282	NCTD	*MYPN*	Hetero-zygous		NM_032578.3c.2671del(p.Ile891Ter) Exon: 12/20	Missense	VUS				
195	Weill Marchesa	*TGFB3*	Hetero-zygous		NM_003239.3c.884del(p.Gly295ValfsTer74) Exon: 5/7	Missense	VUS				

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NCTD: Nonspecific connective tissue disease; VUS: Variants of unknown significance