

Gene Result		Count	DMD	p.Val2159Serfs*4	1	ASL	p.Arg12Gln	1	CYP21A2	p.Ile173Asn	1
Class Ia		51	GBA	p.Asn409Ser	1	ASPA	p.Ala305Glu	1	CYP21A2	p.Pro454Ser	6
BRCA1	p.Tyr1853*	1	HADHA	p.Glu641Serfs*12	1	ASPA	p.Glu285Ala	1	CYP21A2	p.Val282Leu	6
BRCA2	p.Gln1408*	1	HBB	p.Glu7Val (HbS)	1	ATP6V1B1	p.Leu12Serfs*8	1	DBT	p.Arg301Cys	1
BRCA2	p.Gln2859*	1	MEFV	p.Val744Ser	1	ATP7B	p.Arg1319*	1	DHCR7	c.964-1G>C (SA)	11
BRCA2	p.Glu2846Glyfs*22	1	MEFV	p.Val726Ala	3	ATP7B	p.Asp765Asn	1	DHCR7	p.Trp151*	1
BRCA2	p.Ile2588Phefs*60	1	SERPINA1	p.Arg63Cys (I allele)	2	ATP7B	p.Gly869Arg	2	DHCR7	p.Val326Leu	1
BRIP1	p.Arg798*	1	SERPINA1	p.Asp280Val (Null Cardiff allele)	1	ATP7B	p.Thr991Met	1	DHDDS	c.441-24A>G (I)	2
BRIP1	p.Ser895*	1				BBS1	p.Met390Arg	3	DNAH5	exon 57-59 deletion	
CHEK2	p.Ile157Thr	3	SERPINA1	p.Glu366Lys (Z allele)	9	BBS10	p.Cys91Leufs*5	2	DNAH5	p.Arg4127His	1
CHEK2	p.Thr367Metfs*15	2	WNT10A	p.Cys107*	2	BBS10	p.Tyr559*	1	DNAI1	c.48+2dup (I)	1
DSC2	c.631-2A>G (SA)	1	WNT10A	p.Phe228Ile (LP)	9	BCKDHA	p.Arg117Cys (LP)	1	DNAI2	p.Asp597Valfs*8	1
DSG2	c.523+1G>A (SD)	1	Class If		17	BLM	c.3558+1G>T	1	DYSF	c.1053+1G>A	1
DSG2	p.Glu1020Alafs*18	1	CFTR	p.Phe508del and c.1210-34TG[11]T[5] (I) (bi-allelic)	1	BLM	p.Arg899*	1	ERCC6	p.Arg1087*	1
F2	c.*97G>A (c.20210G>A)	5				BLM	p.Gln627Arg	1	ETFDH	p.Leu334Pro	1
F5	p.Arg534Gln (Leiden)	14	HFE	p.Cys282Tyr (homozygous)	3	BTDD	p.Asp444His (PD)	23	EYS	c.2137+1G>A (SD)	1
HOXB13	p.Gly84Glu	1	HFE	p.Cys282Tyr and p.His63Asp (bi-allelic)	4	BTDD	p.Gly34Valfs*35	1	EYS	p.Gln171*	1
LDLR	c.2547+1G>A (SD)	1	HFE	p.His63Asp (homozygous)	7	CACNA1S	p.Cys245Alafs*89	1	EYS	p.His263Thrfs*15	1
LMNA	p.Thr655Asnfs*49	1	HFE	p.His63Asp (homozygous) (LP)	1	CACNA1S	p.Gln7*	1	EYS	p.Thr2805Asnfs*7	1
MITF	p.Glu318Lys	3	SERPINA1	p.Glu366Lys (Z allele) and p.Glu288Val (S allele) (bi-allelic)	1	CACNA1S	p.Leu1612Trpfs*22	1	F11	p.Cys146*	1
MYBPC3	c.3628-41_3628-17del (I)	1				CAPN3	p.Arg489Gln	1	F11	p.Glu135*	1
MYBPC3	p.Glu542Gln (missense and splice)	1	WNT10A	p.Phe228Ile (homozygous) (LP)	1	CAPN3	p.Arg490Trp	1	F11	p.Glu19Asnfs*3	1
MYH7	p.Lys865Arg	1				CAV3	p.Ala93Thr	1	FAH	c.1062+5G>A (I)	1
NF1	p.Leu1183Arg (mosaic?)	1	Class Ig		1	CBS	p.Ile278Thr	2	FAH	c.554-1G>T (SD)	1
PKP2	p.Thr50Serfs*61	1	FLNC	deleted gene	1	CBS	p.Pro49Leu	1	FANCA	deletion (exons 11-29)	1
PMS2	p.Lys706Serfs*19	1	Class Ila		504	CDH23	c.7482+2T>C	1	FANCG	c.1480+1G>C (SD)	1
PRKAR1A	c.709-7_709-2del	1	ABCB11	p.Arg487His	1	CDH23	p.Arg1746Gln	1	FH	p.Lys477dup	3
SDHD	p.Pro81Leu	1	ABCC8	c.3989-9G>A (I)	1	CDH23	p.Leu1218Phefs*27	1	FKRP	p.Leu276Ile	1
SERPINC1	p.Pro73Leu	2	ACADM	p.Lys329Glu	3	CDH23	p.Thr1587Cysfs*4	1	FKTN	p.Cys137*	1
TP53	p.Pro151Arg (mosaic?)	1	ACADM	p.Tyr337Ser	1	CERKL	p.Pro287Leufs*10	1	G6PC	p.Arg83Cys	1
Class Ib		1	ACADVL	p.Glu277del	1	CFTR	c.1210-34TG[11]T[5] (I) (LP)	13	G6PC	p.Gln27Argfs*9	1
APOB	p.Phe2656Thrfs*10	1	ACAT1	p.Gly183Arg	1				G6PC	p.Gln319*	1
Class Ic		2	ACSF3	p.Arg558Trp	4	CHRNAE	p.Tyr35His	1	G6PC	p.Gln347*	1
G6PD	p.[Val68Met Asn126Asp] (A- haplotype)	2	ACSF3	p.Glu359Lys	1	CLN3	p.Arg405Trp	1	GAA	c.-32-13T>G (I)	3
Class Id		49	AGXT	p.Phe152Ile	1	CLN5	p.Trp224*	1	GAA	p.Arg281Gln	1
ATM	p.Arg2034*	1	AGXT	p.Trp108Arg	1	CNGB3	p.Glu336*	1	GAA	p.Arg281Trp	1
ATM	p.Glu522Ilefs*43	1	AIRE	p.Leu323Serfs*51	1	CNGB3	p.Pro94Leufs*30	1	GAA	p.Asp404Asn	1
ATM	p.Leu2463fs	1	ALDOB	p.Ala150Pro	3	COL7A1					

Supplemental Table S1: All gene results, by clinical class.

<i>GBE1</i>	p.Tyr329Cys	1	<i>MUTYH</i>	p.Arg274Gln	2	<i>SERPINA1</i>	p.Glu288Val (S allele) (LP)	27	<i>CYP21A2</i>	c.293-13C>G (I) and	1
<i>GCDH</i>	p.Met191Thr	1	<i>MUTYH</i>	p.Gly396Asp	4	<i>SGCB</i>	p.Ser114Phe	2		p.Gly111Valfs*21 and	
<i>GJB2</i>	p.Gly12Valfs*2	4	<i>MUTYH</i>	p.Tyr179Cys	2	<i>SGCG</i>	Deleted gene	1		p.[Ile237Asn Val238Glu	
<i>GJB2</i>	p.Leu56Argfs*26	2	<i>MYO7A</i>	p.Arg1240Gln	1	<i>SGSH</i>	p.Met1?	1		Met240Lys] and	
<i>GJB2</i>	p.Leu90Pro	2	<i>MYO7A</i>	p.Arg634*	1	<i>SLC12A3</i>	c.506-1G>A	1		p.Ile173Asn and	
<i>GJB2</i>	p.Met34Thr	12	<i>NBN</i>	p.Lys233Serfs*5	1	<i>SLC22A5</i>	c.-149G>A (NC)	2		p.Leu308Phefs*6 and	
<i>GJB2</i>	p.Val37Ala	1	<i>NEB</i>	c.12331-1G>A (SA)	1	<i>SLC22A5</i>	p.Ala214Val	1		p.Pro31Leu (in cis)	
<i>GLDC</i>	deletion (exons 1-15)	1	<i>NEB</i>	p.Trp3603*	1	<i>SLC22A5</i>	p.Pro46Ser	1	<i>CYP21A2</i>	p.Gln319* and gene	2
<i>GLDC</i>	p.Arg739His	1	<i>NR2E3</i>	c.119-2A>C (SA)	1	<i>SLC25A15</i>	p.Glu180Lys	1		duplication (phase	
<i>GLDC</i>	p.Pro345Thr	1	<i>NTHL1</i>	p.Gln90*	1	<i>SLC26A4</i>	c.1001+1G>A (SD)	1		unknown)	
<i>GLE1</i>	p.Arg569His	1	<i>OAT</i>	p.Arg398*	1	<i>SLC26A4</i>	p.Glu384Gly	1		<b>Class IIc</b>	<b>1</b>
<i>GNPTAB</i>	p.Leu1168Glnfs*5	2	<i>PAH</i>	c.1315+1G>A	1	<i>SLC26A4</i>	p.Gly209Val	2	<i>SERPINA1</i>	p.Glu288Val (S allele)	1
<i>GRHPR</i>	p.Asp35Thrfs*11	2	<i>PAH</i>	p.Arg408Trp	2	<i>SLC37A4</i>	p.Ala113Glyfs*18	1		(homozygous) (LP)	
<i>HBA1</i>	CNV x4	1	<i>PAH</i>	p.Glu280Lys	1	<i>SLC37A4</i>	p.Leu116Glyfs*31	1		<b>Class IIIa</b>	<b>3</b>
<i>HBA1</i>	deleted gene	5	<i>PAH</i>	p.Pro281Leu	1	<i>SLC37A4</i>	p.Trp96*	1	<i>ASS1</i>	p.Val269Met and	1
<i>HBA2</i>	deleted gene	2	<i>PAH</i>	p.Val230Ile	1	<i>SLC7A7</i>	deletion (exons 4-5)	1		p.Gly390Arg (bi-allelic	
<i>HBB</i>	p.Gly70Ser	1	<i>PCDH15</i>	p.Arg1106*	1	<i>SMARCA11</i>	p.Glu848*	1		phase unknown)	
<i>HEXA</i>	c.1073+1G>A	1	<i>PCDH15</i>	p.Arg245*	1	<i>SMARCA11</i>	p.Phe279Ser	1	<i>CYP21A2</i>	p.Val282Leu (zygosity	1
<i>HEXA</i>	c.805+1G>A	1	<i>PEX1</i>	p.Arg949Gln	1	<i>SMN1</i>	c.*3+80T>G (g.27134T>G)	1		uncertain)	
<i>HFE</i>	p.Cys282Tyr	23	<i>PEX1</i>	p.Ile700Tyrfs*42	2		(ICR)			p.Leu308Phefs*6	
<i>HFE</i>	p.His63Asp (LP)	69	<i>PEX12</i>	p.Tyr74*	1	<i>SMN1</i>	deleted gene	8		p.Arg357Trp as well as	
<i>HGD</i>	p.Gly161Arg	1	<i>PEX6</i>	p.Leu614Argfs*5	1	<i>SMPD1</i>	p.Asp253His	1		p.Ile237Asn, p.Val238Glu,	
<i>HGD</i>	p.Met368Val	1	<i>PEX7</i>	p.Gly217Arg	1	<i>SMPD1</i>	p.Gly244Arg	1		p.Met240Lys (phase	
<i>HJV</i>	p.Gly302Val	1	<i>PEX7</i>	p.Leu292*	2	<i>TGM1</i>	p.Trp263	1		unknown)	
<i>HOGA1</i>	c.700+5G>T (I)	3	<i>PFKM</i>	c.237+1G>A (SD)	1	<i>TMEM216</i>	p.Asn121Glufs*15	1	<i>DNAH5</i>	c.2744-1G>T and	1
<i>HOGA1</i>	p.Ala36Val	1	<i>PKHD1</i>	c.390+1G>T (SD)	1	<i>TPP1</i>	p.Arg208*	1		p.Arg36Glyfs*8 (bi-allelic	
<i>HPS3</i>	p.Arg397Trp	1	<i>PKHD1</i>	p.Ala1254Glyfs*49	1	<i>TPP1</i>	p.Lys18*	1		phase unknown)	
<i>HPS3</i>	p.Gly146*	1	<i>PKHD1</i>	p.Arg494*	1	<i>TRMU</i>	p.Leu195Ilefs*12	1			
<i>IDUA</i>	c.386-2A>G	1	<i>PKHD1</i>	p.Ile2331Lys	2	<i>TYMP</i>	p.Glu289Ala	1		Abbreviations: SA – splice	
<i>LAMB3</i>	p.Val948Cysfs*82	1	<i>PKHD1</i>	p.Thr36Met	1	<i>TYMP</i>	p.Val208Met	1		acceptor, SD – splice donor, I –	
<i>LIPA</i>	p.Ser133*	1	<i>PMM2</i>	c.255+2T>C (SD)	1	<i>USH1C</i>	c.216G>A (Silent)	2		intronic, NC – non-coding, LP –	
<i>LPL</i>	p.Arg197Cys	1	<i>PMM2</i>	p.Arg141His	6	<i>USH1C</i>	c.748_759+5del (SD)	1		low penetrance, PD – partial	
<i>LRPPRC</i>	p.Ala354Val	1	<i>PMM2</i>	p.Val67Gly	1	<i>USH1C</i>	deletion (exon 9)	1		deficiency, ICR – increased	
<i>MAN2B1</i>	p.Trp714Arg	1	<i>POMGNT1</i>	c.1539+1G>A (SD)	1	<i>USH2A</i>	c.949 C>A	1		carrier risk	
<i>MCC2</i>	p.Gly475Arg	1	<i>PPT1</i>	p.Arg151*	2	<i>USH2A</i>	p.Arg205Glufs*11	1			
<i>MCOLN1</i>	p.Thr232Pro	1	<i>PYGM</i>	p.Arg50*	1	<i>USH2A</i>	p.Arg4192His	1			
<i>MED17</i>	p.Gln482*	1	<i>PYGM</i>	p.Lys754Asnfs*49	1	<i>USH2A</i>	p.Glu3562*	1			
<i>MKS1</i>	p.Cys492Trp	1	<i>PYGM</i>	p.Phe599Leufs*6	1	<i>USH2A</i>	p.Glu767Serfs*21	1			
<i>MKS1</i>	p.Gln408*	1	<i>RAPSN</i>	p.Asn88Lys	3	<i>USH2A</i>	p.Ile1183Phefs*19	1			
<i>MLC1</i>	deletion (exon 3)	1	<i>RMRP</i>	n.-17_-2dup (NC,	1	<i>USH2A</i>	p.Ile5166Val	1			
<i>MMAA</i>	p.Arg145*	1		promoter)		<i>USH2A</i>	p.Trp3955*	1			
<i>MMACHC</i>	p.Arg91Lysfs*14	1	<i>RMRP</i>	n.-25_-3dup (NC)	1	<i>VPS13B</i>	c.10942+1G>A (SD)	1			
<i>MPL</i>	p.Lys553Argfs*77	1	<i>RMRP</i>	n.71A>G (RNA change)	1	<i>XPA</i>	p.Gln185His	1			
<i>MPV17</i>	p.Pro98Leu	1	<i>RMRP</i>	NR_003051.3:n.239C>T	1						
<i>MSH3</i>	deletion (exon 16)	1		(RNA Change)							
<i>MTHFR</i>	p.Arg52Gln	1	<i>RPGRIP1L</i>	p.Arg1177*	1						
<i>MUT</i>	p.Ala676Thr	1	<i>SACS</i>	deleted gene	1						
							<b>Class IIb</b>	<b>3</b>			