Clinical and Genetic Spectrum of Glycogen Storage Disease in Iranian Population using Targeted Gene Sequencing

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Table S1. Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing

GSD Types	Genes	Transcript	GRCh37 location	Exon/Intron	cDNA location	Strand	NGS result	Sanger result
GSD Ib	*SLC37A4*	NM_001164279.2	chr11:118900056	Exon8	c.24	-	T>G	T>G
GSD Ib	*SLC37A4*	NM_001164279.2	chr11: 118898407	Exon6	c.337	-	C>T	C>T
GSD III	*AGL*	NM_000642.3	chr1:100336041	Exon6	c.753_756	+	delCAGA	delCAGA
GSD III	*AGL*	NM_000642.3	chr1:100336041	Exon6	c.753_756	+	delCAGA	delCAGA
GSD III	*AGL*	NM_000642.3	chr1:100342081	Exon11	c.1351_1355	+	delAAAGC	delAAAGC
GSD III	*AGL*	NM_000642.3	chr1:100379113	Exon30	c.3980	+	G>A	G>A
GSD IV	*GBE1*	NM_000158.4	chr3:81643169	Exon8	c.998	-	A>T	A>T
GSD IV	*GBE1*	NM_000158.4	chr3:81754616	Exon2	c.292	-	G>C	G>C
GSD VI	*PYGL*	NM_002863.5	chr14:51378453	Exon16	c.1964	-	A>G	A>G
GSD VI	*PYGL*	NM_002863.5	chr14:51410891	Exon1	c.229_231	-	delGAC	delGAC
GSD IXc	*PHKG2*	NM_000294.2	chr16:30762461	Exon3	c.130	+	C>T	C>T
GSD IXb	*PHKB*	NM_000293.3	chr16:47531367	Exon2	c.134	+	T>A	T>A
GSD IXb	*PHKB*	NM_000293.3	chr16:47628046	Exon11	c.1127-2	+	A>G	A>G
GSD IXb	*PHKB*	NM_000293.3	chr16:47727384	Exon28	c.2840	+	A>G	A>G
GSD X	*PGAM2*	NM_000290.4	chr7:44105115	Exon1	c.14	-	G>A	G>A
GSD heart, lethal congenital	*PRKAG2*	NM_001040633.1	chr7:151329185	Exon5	c.592	-	A>T	A>T