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Supplemental Table: Additional pathogenic variants and variant of unknown clinical significance identified with the HHA panel

Patient	Gene	Refseq and variant	Amino acid change	Zygosity	Interpretation	HGMD	ClinVar	gnomAD-All	in silico prediction
2	<i>KLF1</i>	NM_006563.3:c.604G>A	p.Gly202Arg	het	VUCS	not reported	not reported	0.0064%	pathogenic
	<i>PKLR</i>	NM_000298.5:c.1456C>T	p.Arg486Trp	het	likely pathogenic	DM	path/likely path	0.2900%	pathogenic
	<i>SPTA1</i>	NM_003126.3:c.803G>A	p.Arg268Gln	het	VUCS	not reported	not reported	0.0007%	conflicted
8	<i>G6PD</i>	NM_001042351.2:c.844G>C	p.Asp282His	hemi	Pathogenic (class III G6PD variant)		pathogenic		
10	<i>PIEZO1</i>	NM_001142864.2:c.4674C>T	p.Gly1558Gly	HET		not reported	not reported	0.0330%	not predicted to create or disrupt a splice site
11	<i>ABCG5</i>	NM_022436.2:c.1535T>C	p.Ile512Thr	HET	VUCS	not reported	not reported	0.0028%	conflicted
	<i>ABCG5</i>	NM_022436.2:c.969G>T	p.Gln323His	HET	VUCS	not reported	not reported	0.0004%	benign
	<i>EPB42</i>	NM_000119.2:c.520+12C>T		HET	VUCS-LB	not reported	VUS	0.1400%	not predicted to create or disrupt a splice site
	<i>PIEZO1</i>	NM_001142864.2:c.1356C>G	p.Leu452Leu	HET	VUCS	not reported	not reported	NA	not predicted to create or disrupt a splice site
	<i>SEC23B</i>	NM_006363.4:c.2202T>C	p.Tyr734Tyr	HET	VUCS	not reported	not reported	NA	may disrupt a splice donor

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