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RECOMMENDATION FOR FURTHER TESTING: Please provide us parental samples to confirm the clinical significance of the identified variations.

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Please note that while mutation and/or linkage analysis often gives precise information about genotype, there are several sources of error. These include sample misidentification, erroneous paternity identification, and genotyping errors resulting from contamination of PCR reagents or contamination of fetal samples with maternal cells. Rare genetic variants may also interfere with accurate analysis. Families should understand that rare diagnostic errors might occur for these reasons. *This test was developed and its performance characteristics determined by the UCD DNA Diagnostic Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary* **REFERENCES:** Eur J Hum Genet 2005, 13:302-308.

Table 1. Heterozygous SNPs and mutations identified in the patient. Non-synonymous variations were predicted by PolyPhen (http://genetics.bwh.harvard.edu/pph/) and by SIFT (http://sift.jcvi.org/). Synonymous variations were analyzed by using RESCUE-ESE program (http://genes.mit.edu/burgelab/rescue-ese/). Intronic variations were analyzed by NetGene2 server (http://www.cbs.dtu.dk/services/NetGene2/).

Gene	Variations/mutations	SNP	pathogenicity
ASCC3L1	IVS40-20A>G	rs3214063	not likely (allele frequency)
BEST1	IVS4+44C>T	rs195162	not likely (allele frequency)
	IVS5insTCCTCCTCC	rs72141234	not rare
	p.T470T:c.1410G>A	rs149698	not likely (allele frequency)
FSCN2	5'UTR-39C>T	rs2075720	not likely (allele frequency)
GUCA1B	p.Y57Y:c.171T>C	rs3749921	not likely (allele frequency)
PRPF31	IVS5-31T>C(homozygous)	rs2303557	not likely (allele frequency)
	IVS10-9T>C(homozygous)	rs655240	not likely (allele frequency)
	p.N413N:c.1239C>T	no SNP data	polymorphism (mutation taster)