Result- Heterozygous for a single PNKP mutation; Heterozygous for a single PRRT2 mutation. No other reportable variants detected by sequencing and deletion/duplication analysis of the 75 genes included on this panel.

Interpretation: This individual is heterozygous for a novel disease-causing mutation in the PNKP gene. This gene is associated with an autosomal recessive disorder. A second mutation may exist that is undetectable by this test or this patient may incidentally be a heterozygous carrier of the PNKP mutation. The finding of a single mutation in PNKP is not sufficient to establish a diagnosis in this patient. This individual is heterozygous for a published missense variant in the PRRT2 gene. This gene is associated with autosomal dominant disorder. With the clinical and molecular information available at this time, the clinical significance of this variant is unknown.

Method â□□Using genomic DNA from the submitted specimen, the coding regions and splice junctions of 51 genes (all genes listed above except for CHRNA7 and MAGI2, since only large deletions have been reported in these genes) were sequenced with pair-end reads. Capillary sequencing was used to confirm all potentially pathogenic variants. Concurrent deletion/ duplication testing was performed for the genes in the panel using exon-level oligo array CGH, except for FOXG1. Confirmation of copy number changes was performed by MLPA, qPCR, or repeat array CGH analysis.

Additional Information â□□The test also found likely benign variants in genes KANSL1 and PNKP.