# Whole Exome Sequencing Test

| | | | |
|---|---|---|---|
| **Institution** | | Lab No. | |
| **Name** | | Age/Gender | |
| **Accession No.** | | Sample type | Whole Blood |
| Department/Doctor | / | Received /Reported | |

**Indication :** **Severe hemolytic anemia, cholestasis, liver failure**

**Test Result** (The variant related to patient's clinical phenotype)

**(This result is the final report.)**

| | |
|---|---|
| **Variant** | **The aberration was detected, related to patient's clinical phenotype.** |

**Type of variant**

| Gene | DNA change | Predicted AA change | Zygosity | Disease | Inherit | Class |
|---|---|---|---|---|---|---|
| PKLR | c.283+1914_1434del | p.(?) | Homo | PK deficiency Reference sequence: NG_011677.1(NM_000298.6) | AR | Pathogenic |

Abbreviations: Homo= Homozygous; PK= pyruvate kinase; AR= Autosomal recessive

**Interpretation**

Whole exome sequencing analysis identifed a homozygous pathogenic copy number aberration in the PKLR gene associated with red cell pyruvate kinase (PK) deficiency.

The c.283+1914_1434del is a deletion of 5,006 base pairs extending from intron 2 to almost the 3'end of exon 9 of the PKLR gene. The aberration has previously been reported in the pyruvate kinase deficiency patients (PMID: 15642665 described as PK Viet del 4-10; PMID: 15953013 described as del 5006bp IVS3--> nt 1431; PMID: 22183074 decribed as c.283+1914_c.1434del5006).

The PKLR is a causative gene of autosomal recessive red cell pyruvate kinase (PK) deficiency. Genetic counseling for the patient is recommended and further genetic analysis to check the carrier status of the parents would be helpful for family planning.

* NGS analysis found that no sequence read was aligned from exon 3 to exon 9 of the PKLR gene, which indicated a homozygougs deletion of that region. Gap-PCR followed by Sanger sequencing confirmed the finding and characterized the deletion margin. Primers described in the previous report were used(PMID: 22183074).



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## Whole Exome sequencing Methods

Genomic DNA was extracted from EDTA whole blood and we captured all the exons of human genes using SureSelectXT Human All Exone v5 (Agilent).For massively parallel sequencing, the post-capture library DNA was used to perform sequence analysis on Illumina NextSeq platform with 150 bp paired end reads. The DNA sequence was mapped to, and analyzed in comparison with,the public human genome build (UCSC hg19 reference). All reportable sequence variants were confirmed by Sanger sequence analysis.

| | |
|---|---|
| **Mean depth of coverage** | **115X** |
| **% of > 10x** | **97.2%** |

## Limitation of the test

In the case of not detecting pathogenic genes'reasonable variants on disease phenotypes by sequencing, this test could not exclude any genetic factors related with patient's health conditions. Some types of genetic abnormalities, such as copy number changes, may not be detectable with the technologies performed by this analysis.
It is possible that the genomic region where a disease causing mutation exists in the proband was not captured using the current technologies and therefore was not detected.



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