STAT Whole Genome Sequencing in the NICU

CASE STUDY

think genetic

The Case

Jacob* is a two-month-old male presenting with Shone's complex, short limbs, hypospadias, wide-spaced eyes, retrognathia, low-set ears, and dysplastic nails. Jacob's physician ordered rapid genome sequencing of Jacob and both of his parents.

The Outcome

The STAT Whole Genome Sequencing showed that Jacob has a *de novo* variant in *MAP3K7* which was classified as likely pathogenic. *MAP3K7* variants are associated with cardiospondylocarpofacial syndrome, and despite failure to thrive and cardiac defects, patients with this disorder typically have good outcomes. Jacob required a heart transplant, and the diagnosis helped guide post-transplant treatment.

*Fictional name

When standard testing fails to reveal a diagnosis, **STAT WGS** is available. By integrating our expertise in genomics with clinical decision support, the **HudsonAlpha Clinical Services Lab, LLC.**, works with NICUs across the nation to provide clinicians with the information they need to make a fast and informed diagnosis, saving precious time when it matters most.

STAT Whole Genome Sequencing can provide:

- Rapid Diagnosis
- Early intervention
- Treatment indications

HudsonAlpha CLINICAL SERVICES LAB

Offering high-quality rapid genomic testing, analysis and interpretation in a CAP accredited, CLIA certified laboratory with the technical experts you need at your service.

For more information, contact info@clinicallab.org

(HudsonAlpha Clinical Services Lab does not endorse or prescribe drugs, diagnose patients or recommend therapy.)

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The Case

David* is a three-week-old male presenting with low APGAR scores and no heartbeat at birth, subsequent ventilator dependency, hypotonia, areflexia, decreased bone density with multiple fractures, and mild dysmorphisms. David's physician ordered rapid genome sequencing of David and both of his parents.

The Outcome

The STAT Whole Genome Sequencing showed that David has compound heterozygous pathogenic *RYR1* variants. Previous reports of similar *RYR1* variants suggested severe myopathy with negative outcomes were likely. No treatment is currently available, but the information from the genome sequencing allowed David's physician to change treatment strategies to palliative care for David.

*Fictional name

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The Case

Blake* is a one-month-old male with a history of respiratory distress, severe hypotonia, club foot, arthrogryposis, long bone fractures and dysmorphic features. Blake's physician ordered rapid genome sequencing of Blake and both of his parents.

The Outcome

The STAT Whole Genome Sequencing showed that Blake has a homozygous variant in *KLHL40*. The gene is associated with Nemaline myopathy which has a high mortality rate in infancy, indicating likely poor outcomes for Blake. The diagnosis allowed Blake's care team to decide between a permanent G-tube and palliative care for Blake.

*Fictional name

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