BAYLOR GENETICS

BAYLOR GENETICS
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PATIENT CASE

Rapid Whole Genome Sequencing (rWGS)

Rapid Trio Whole Genome Sequencing for your patients with large differential diagnoses or broad symptoms.

Initial Presentation:

- 3-day old infant with hypotonia, hypoglycemia, feeding difficulties, long fingers, retrognathia, and abnormality of the gingiva

Genetic Tests Performed:

- The patient had no prior genetic testing
- Initial differential diagnoses included spinal muscular atrophy, metabolic disorders, and Prader Willi/Angelman Syndrome (PWS/AS)

Rapid Trio WGS Test Findings:

- Rapid Trio WGS detected a heterozygous pathogenic copy number loss involving chromosome bands 15q11.2-q13.1, which is consistent with the common deletion in the PWS/AS critical region on chromosome 15
- Also, with the advantage of trio genome sequencing analysis, the copy number variant was determined to be on the paternally inherited chromosome 15, which is consistent with a diagnosis of Prader-Willi syndrome in this individual.

Impact on Medical Management:

- A rapid diagnosis allows the NICU team to quickly direct clinical care for the standard management of PWS patients

WGS is the most comprehensive test available for patients with large differential diagnoses or broad symptoms. By analyzing SNVs, CNVs, the mitochondrial genome, and repeat expansions quickly, in a single test, we can help end the diagnostic odyssey before it starts.

Rapid Trio WGS testing determined that the variant was on the paternal allele which indicated a diagnosis of Prader-Willi syndrome as opposed to Angelman syndrome. This finding also informed recurrence risk since the variant was *de novo*.

Whole Genome Sequencing: Rapid Trio

Proband Report

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DEMOGRAPHIC INFORMATION

PATIENT

NAME:
DATE OF BIRTH:
SEX:
MEDICAL RECORD #:
ACCESSION #:
LAB NUMBER:
FAMILY NUMBER:

TEST INFORMATION

TEST NAME:	Rapid Trio WGS
TEST CODE:	1822
SAMPLE TYPE:	BLOOD
DATE COLLECTED:	
DATE RECEIVED:	
DATE REPORTED:	

RECIPIENT

PHYSICIAN NAME:
FACILITY:
LOCATION:
PHONE:
FAX:
EMAIL:

ADDITIONAL RECIPIENT

NAME:
PHONE:
FAX:
EMAIL:

ADDITIONAL RECIPIENT

NAME:
PHONE:
FAX:
EMAIL:

CLINICAL INDICATION

Based on the submitted clinical information, the patient has hypotonia, hypoglycemia, feeding difficulties, long fingers, retrognathia, abnormality of the gingiva.

We have also received samples from the father (DNA# 123456) and the mother (DNA# 123456) of this individual.

RESULTS

POSITIVE FINDINGS

DISEASE	INHERITANCE PATTERN	GENE / VARIANT	VARIANT TYPE	GENOTYPE	INHERITED FROM	VARIANT CLASSIFICATION
15q11.2q13.1 Deletion	Autosomal Dominant	SNORD116-1,OCA2,UBE3A...: chr15:23304900-28293509DEL 4988.61(kb)	Copy Number Variant	Heterozygous	De Novo	Pathogenic

RESULTS SUMMARY

Heterozygous Pathogenic Copy Number Loss involving chromosome bands 15q11.2q13.1 detected. Genome sequencing analysis also showed this copy number loss variant is on paternally inherited chromosome 15. This finding maybe consistent with a diagnosis of Prader-Willi syndrome in this individual.

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Test Name

The clinical indication for testing was broad and the provider felt that comprehensive testing was indicated.

Key findings summary with disease and variant information. A *de novo* pathogenic deletion on the paternal allele was detected.