BAYLOR GENETICS

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PATIENT CASE

Rapid Whole Genome Sequencing (rWGS)

Rapid Whole Genome Sequencing reveals genetic etiology in newborn with epilepsy.

Initial Presentation:

- 2-month-old with seizures, feeding difficulties, lethargy, concern for sepsis, and fever

Genetic Tests Performed:

- The patient had no prior genetic testing
- Initial differential diagnoses by the clinic included 22q11.2 deletion syndrome, 15q duplication syndrome, Angelman syndrome, GLUT1 deficiency, and Dravet syndrome

Rapid Trio WGS Test Findings:

- Rapid Trio WGS detected compound heterozygous variants in the *COQ5* gene, consistent with a diagnosis of a primary coenzyme Q10 (CoQ10) deficiency 9

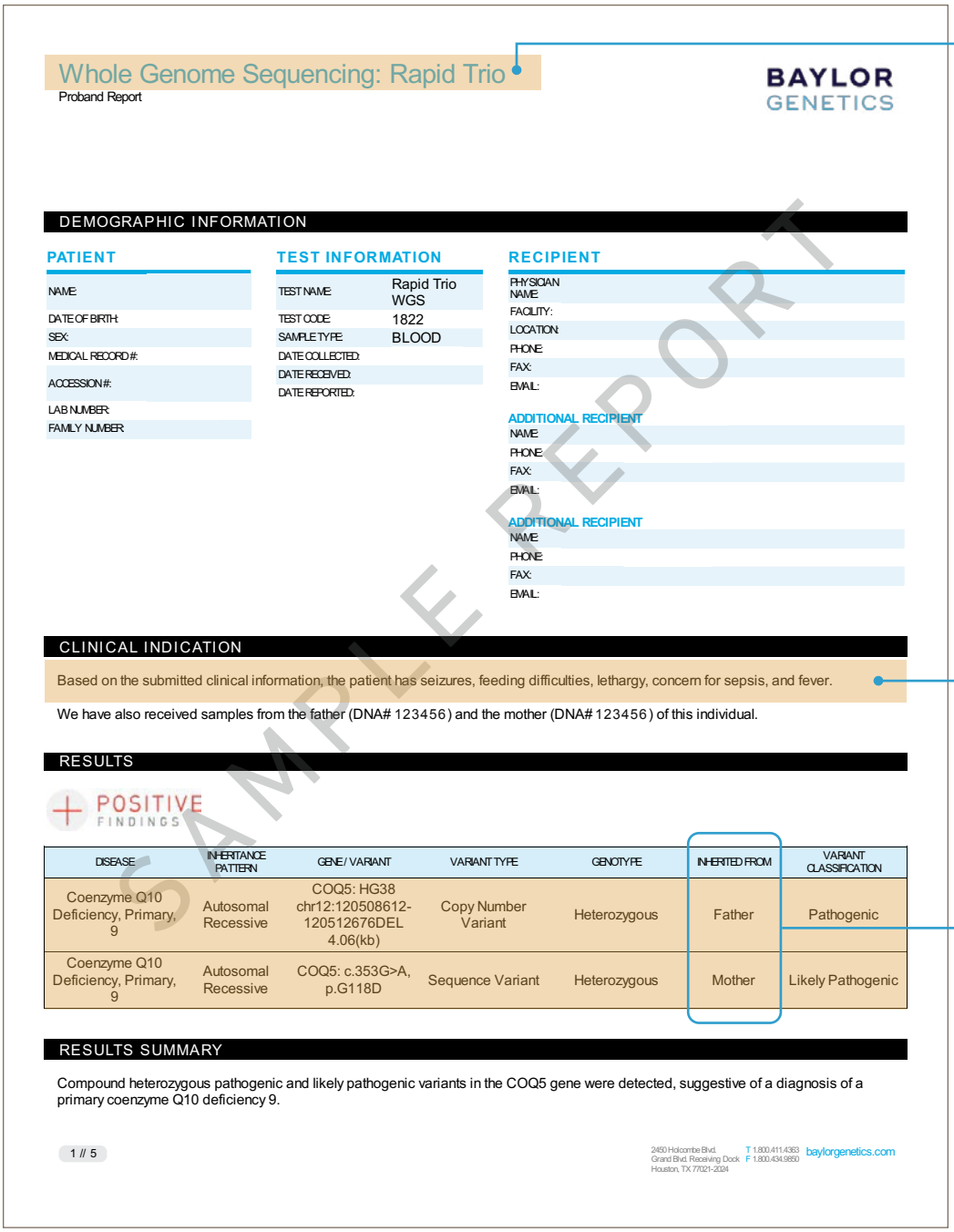
Impact on Medical Management:

- Patients with primary CoQ10 deficiency 9 may benefit from treatment with CoQ10 supplementation

This case demonstrates the ability of Rapid WGS to provide a timely diagnosis when a patient has a large differential, so that medical management may be quickly initiated.

Rapid Trio WGS revealed that both *COQ5* variants were inherited from either parent. These findings inform recurrence risk as both parents are carriers of this condition.

WGS being performed in a trio setting provided inheritance information. A rapid result also allowed for expedited actionable results that could directly impact the patients management.



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 seizures, feeding difficulties, lethargy, concern for sepsis, and fever.

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
Coenzyme Q10 Deficiency, Primary, 9	Autosomal Recessive	COQ5: HG38 chr12:120508612-120512676DEL 4.06(kb)	Copy Number Variant	Heterozygous	Father	Pathogenic
Coenzyme Q10 Deficiency, Primary, 9	Autosomal Recessive	COQ5: c.353G>A, p.G118D	Sequence Variant	Heterozygous	Mother	Likely Pathogenic

RESULTS SUMMARY

Compound heterozygous pathogenic and likely pathogenic variants in the COQ5 gene were detected, suggestive of a diagnosis of a primary coenzyme Q10 deficiency 9.

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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. One likely pathogenic and one pathogenic variant were identified.