Rady Children's Institute Genomic Medicine

Clinical Genome Center
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CLIA ID# 05D2129627
CAP ID# 9487427

RCIGM Genome Report

PATIENT NAME	Baby Boy	ORDERING PHYSICIAN	Dr. Doctor	RCIGM CASE ID	CSXXXX_INXXXX
SEX	Male	ORDERING PROVIDER	John Doe, CGC	MOTHER ID	CSXXXX_INXXXX
DATE OF BIRTH	01/07/2023	HOSPITAL	Hospital for Children	FATHER ID	CSXXXX_INXXXX
INDICATION FOR TESTING	Suspected Genetic Condition	SPECIMEN	Blood	PRELIMINARY REPORT DATE	01/26/2023
TEST TYPE	Trio Whole Genome Sequencing	COLLECTED	01/22/2023	REPORT DATE	02/09/2023
		RECEIVED	01/24/2023		

PATIENT PHENOTYPE

Hepatomegaly; Hepatic steatosis; Fever; Anemia; Leukocytosis; Thrombocytosis; Neutrophilia; Elevated circulating C-reactive protein concentration; Patent foramen ovale; Atelectasis; Hydronephrosis; Hypotonia; Persistent head lag; Pinguecula; Hypoglycemia; Large for gestational age; Subglottic laryngitis; Feeding difficulties; High palate; Narrow forehead; Pallor; Hypoplastic spleen

TEST RESULT: PRIMARY FINDING IDENTIFIED, AND ADDITIONAL FINDING REPORTED

Sequence Variants

REPORT CATEGORY	GENE	VARIANT	CONDITION	ZYGOSITY (INHERITANCE)	VARIANT CLASSIFICATION
VARIANTS RELATED TO PATIENT PHENOTYPE	XIAP	c.653C>G p.Ser218Ter	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2	Hemizygous (Maternal)	Likely pathogenic
VARIANTS POSSIBLY RELATED TO PATIENT PHENOTYPE	F2	c.*97G>A	F2-RELATED DISORDERS	Heterozygous (Maternal)	Pathogenic

*Details on the variant(s) and gene(s) are located in the subsequent sections of the report

VARIANTS RELATED TO PATIENT PHENOTYPE

CONFIRMATION STATUS	GENE (TRANSCRIPT)	CONDITION	GENOMIC COORDINATES	VARIANT	ZYGOSITY (INHERITANCE)	CLASSIFICATION
Confirmed	XIAP (ENST00000371199)	LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2	X:123020165	c.653C>G p.Ser218Ter	Hemizygous (Maternal)	Likely pathogenic

Variant Information (XIAP)

A hemizygous c.653C>G (p.Ser218Ter) variant in the XIAP gene was detected in this individual. This nonsense variant found in exon 2 of 7 is predicted to result in loss of normal protein function through either protein truncation or nonsense-mediated mRNA decay. The XIAP gene is constrained against loss-of-function variation (pLI = 0.92) and loss of function variation has been reported in individuals with XIAP-related syndrome (PMID: 20301580). This variant has not been previously reported or functionally characterized in the literature to our knowledge. The c.653C>G (p.Ser218Ter) variant is absent from the gnomAD population database and thus is presumed to be rare. This result was confirmed by orthogonal testing. Analysis of the parental samples showed the mother is heterozygous and the father is negative for this variant. Based on the available evidence, c.653C>G (p.Ser218Ter) is classified as Likely Pathogenic.

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Gene Information (XIAP)

The XIAP gene is located on chromosome Xq25 and encodes X-Linked Inhibitor of Apoptosis, which plays a role in blocking apoptosis and is involved in a variety of signaling pathways and cellular responses which play an innate role in immunity (PMID: 25666262). Pathogenic variations in the XIAP gene are associated with X-linked recessive lymphoproliferative syndrome-2 (XLP2) (MIM: #300635), a primary immune deficiency syndrome characterized by variable clinical features including hemophagocytic lymphohistiocytosis (often associated with Epstein-Barr virus), recurrent infections, splenomegaly, fever, colitis or inflammatory bowel disease (IBD) (PMID: 22228567, 25666262). Laboratory findings include dysgammaglobulinemia, cytopenias, and low levels of invariant natural killer T (iNKT) cells. XLP2 has a high degree of clinical variability and severity, even within families, which can range from asymptomatic to life-threatening (PMID: 20301580, 25666262). Heterozygous females are typically asymptomatic; however, an affected female with skewed X-chromosome inactivation has been previously reported (PMID: 25801017). Additional clinical information and management recommendations for X-linked lymphoproliferative disease are available on GeneReviews (PMID: 20301580).

VARIANTS POSSIBLY RELATED TO PATIENT PHENOTYPE

CONFIRMATION STATUS	GENE (TRANSCRIPT)	CONDITION	GENOMIC COORDINATES	VARIANT	ZYGOSITY (INHERITANCE)	CLASSIFICATION
Not Required	F2 (ENST00000311907)	F2-RELATED DISORDERS	11:46761055	c.*97G>A	Heterozygous (Maternal)	Pathogenic

Variant Information (F2)

A heterozygous noncoding c.*97G>A variant in the F2 gene was detected in this individual. This variant is also referred to as prothrombin 20210G>A in the literature. This is a known Pathogenic variant that has been previously reported as a heterozygous and homozygous change in patients with prothrombin thrombophilia (PMID: 20301327, 8916933, 25693916, 28707429, 34570182, 33258288, 31472339, 31064749, 30005273, 26732783). This is a recurrent risk factor variant associated with elevated prothrombin plasma levels and an increased risk of venous thromboembolism (VTE) which is highest in homozygotes (PMID: 9569177, 20301327, 26732783, 8916933, 20301327, 19531787, 30297698). This variant has been reported in the ClinVar database (Variation ID: 13310). In-vitro functional studies indicate this variant acts as gain of function alteration leading to increase protein synthesis (PMID: 15059842). The c.*97G>A variant is present in the gnomAD population database at a frequency of 0.8% (265/31396) in the heterozygous state and a frequency of 0.003% (1/31396) in the homozygous state. Analysis of the parental samples showed the mother is heterozygous and the father is negative for this variant. Based on the available evidence, c.*97G>A is classified as a Pathogenic.

Gene Information (F2)

The F2 gene is located on chromosome 11p11.2 and encodes Coagulation Factor II Thrombin, which plays an important role in hemostasis and thrombosis (PMID: 19598065). Pathogenic variations in the F2 gene are associated with autosomal recessive congenital prothrombin deficiency (MIM: #613679) and autosomal dominant prothrombin thrombophilia (MIM: #188050). Congenital prothrombin deficiency is characterized by low levels of circulating prothrombin and is associated with two main phenotypes: type I deficiency, known as hypoprothrombinemia, characterized by severe bleeding from birth and type II deficiency, known as dysprothrombinemia, characterized by normal or low-normal synthesis of a dysfunctional protein where bleeding is more variable. Individuals heterozygous for variants in F2 associated with congenital prothrombin deficiency are usually asymptomatic but can show bleeding after tooth extraction or surgical procedures (PMID: 19598065). Prothrombin thrombophilia is associated with a specific pathogenic variant in F2 (20210G>A variant, also known as c.*97G>A) and is characterized by venous thromboembolism, which typically manifests in adults as deep-vein thrombosis in the legs or pulmonary embolism (PMID: 11309638, 21707594). The clinical expression of prothrombin thrombophilia is variable, and many individuals never develop thrombosis. Typically, individuals heterozygous for the 20210G>A variant remain asymptomatic until adulthood;

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however, some have recurrent thromboembolism before age 30 years (PMID: 20301327). Additional clinical information and management recommendations for prothrombin thrombophilia are available on GeneReviews (PMID: 20301327).

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Adam MP, Mirzaa GM, Pagon RA, Wallace SE, et al. None. 1993. Prothrombin Thrombophilia (PMID: 20301327)

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RECOMMENDATIONS

- Clinical correlation is recommended.
- Clinical molecular testing should be interpreted in the context of the patient's clinical presentation and the prior probability of the clinical signs and symptoms being associated with known single gene disorders (i.e. defects in the identified gene).
- Genetic counseling is recommended to assess the specific implications of these results relative to an individual's clinical context.
- Additional testing may be appropriate to evaluate for other types of variants not evaluated by this test.
- As knowledge increases, periodic re-evaluation of the clinical implications of variants is appropriate. Please contact RCIGM_rWGS@rchsd.org for questions about the RCIGM re-analysis policy.
- Mitochondrial DNA disorders can be sporadic or maternally inherited. If the reported mtDNA variant is found in the mother, the testing of appropriate matrilineal relatives is recommended.
- If there is a strong clinical suspicion of mitochondrial disease, additional testing of different tissue types may be warranted.

METHODOLOGY

Sequence via next generation sequencing (NGS) technology is generated from genomic DNA. PCR-free library preparation is performed prior to whole genome sequencing (WGS). An average genomic coverage of at least 35x, and/or at least 90% of OMIM genes will achieve 100% of coding base coverage of >10x for each proband. This ensures robust and uniform genome coverage. The current version of this test assesses single nucleotide variants (SNVs), small deletions and insertions, larger deletions and duplications, the mitochondrial genome, SMN1 and SMN2 copy number analysis, and repeat expansions in PHOX2B and DMPK. Alignment and variant calling are performed using the Illumina DRAGEN pipeline using the official reference build 37.1 (hg19). Copy number variation (CNV) calling is performed using a combination of CNV callers. Interpretation of CNVs is focused on variants that overlap or have a boundary that lies within 1 kb of an exon in all coding genes. Repeat expansion calling for PHOX2B and DMPK only are performed using

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Expansion Hunter (v.4.0.2) in DRAGEN 3.10.4. Repeat expansions in PHOX2B may also be detected by the DRAGEN 3.10.4 SNVs & Indels analysis pipeline.

Orthogonal Confirmation Policy

Reported sequencing variants are confirmed by Sanger sequencing, but may not be confirmed using orthogonal technologies if the following criteria are met: 1) the coverage at the variant's position is ≥ 20x; 2) the allelic balance for heterozygous calls is between 0.3-0.7; 3) the allelic balance is 0 (wild type allele as reference) for homozygous and hemizygous calls; 4) no systematic sequencing errors or local alignment problems are observed; 5) the call is not located in difficult sequence context (highly homologous and repetitive regions); 6) the call is not a complex insertion/deletion call resulting from nearby variants that may be difficult to align. If specific protocols require orthogonal confirmations of all reported variants, confirmations will be performed. If the case is ordered as proband-only and parental samples are available, targeted inheritance studies will be conducted for selected variants of interest.

Reported copy number variants are confirmed using orthogonal technologies including Multiplex ligation-dependent probe amplification (MLPA), but may not be confirmed using orthogonal technologies if the following criteria are met: 1) the deletion or duplication event contains robust coverage and/or NGS read support; 2) no systematic sequencing errors or local alignment problems are observed; 3) the call is not located in difficult sequence context (highly homologous, repetitive regions, or segmental duplication regions. If specific protocols require orthogonal confirmations of all reported variants, confirmations will be performed. If the case is ordered as proband-only and parental samples are available, targeted inheritance studies will be conducted for selected variants of interest.

Orthogonal confirmation of the PHOX2B and DMPK expansion variants is performed at an external CAP/CLIA laboratory. Final test results reporting may be affected by turnaround times of CAP/CLIA laboratory.

Reporting Categories

Variants related to patient's phenotype – findings with strong variant pathogenicity evidence and strong evidence that the reported gene-disease association overlaps with the patient's phenotype

Variants possibly related to patient's phenotype - findings that are suggestive of a diagnosis but lacks either conclusive variant pathogenicity evidence or lack conclusive gene-disease association evidence

Variants in genes of uncertain significance – findings in genes that lack strong or supporting evidence for association with human disease

Variants in the mitochondrial genome – findings located within the mitochondrial genome

Incidental findings - findings in genes that do not overlap with the patient's reported phenotype, but may be medically actionable for the patient or tested family members

Test Specifications

The sensitivity and specificity for SNVs (single nucleotide variants) and small insertions and deletions up to 50 base pairs is greater than 99%. The analytical validity of SNVs was assessed using reference samples provided through the Genome in a Bottle (Zook et al. 2019, PMID: 30936564).

The sensitivity for larger deletions and duplications from WGS is estimated to be greater than 85%, although reliable reference data for these types of events are not well established. Deletions and duplications from 1 Kb to whole chromosomal abnormalities are detected with this test.

The sensitivity for detecting GCN trinucleotide repeat expansions within the PHOX2B carboxy terminal polyalanine repeat region by Expansion Hunter on short-read NGS is > 90% for expansions greater than 20 repeats. The sensitivity for detecting 3' UTR DMPK trinucleotide repeat expansion (CTG repeats) is >90% for expansions greater than 50 repeats. However, due to short-read NGS technology's limitations, repeat expansion size cannot be precisely determined and

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reported (see Test Limitation section of the report).This test is validated for copy number analysis of exons 7 and 8 of the SMN1 and SMN2 genes. Over 95% of pathogenic variation for SMA involves biallelic loss of exon 7 of the SMN1 gene (Prior et al. 2010, PMID: 20057317). Other variation within the SMN1 gene is not detected with this assay. Results will only be reported in the proband if 0 copies of SMN1 are detected. Parental carrier status will be reported for affected patients if samples are available. Results are orthogonally confirmed by Multiplex Ligation-dependent Probe Amplification (MLPA). Whole genome sequencing is unable to determine the phase of SMN1 variants in the absence of parental testing. Therefore, in the absence of phasing, this assay does not exclude the possibility than an individual harbors two copies of SMN1 on the same allele and no copies on the other allele (silent carriers), two pathogenic sequence variants on the same SMN1 allele, two pathogenic sequence variants on opposite SMN1 alleles, and one pathogenic sequence variant and the loss of exon 8 on the opposite SMN1 allele.

Non-PCR amplified whole-genome sequencing (WGS) provides a stable, at least ~1,000-fold average, coverage across the entire mitochondrial genome (mtDNA). This test can detect SNVs, small insertions and deletions, as well as large deletions in the mtDNA. For mtDNA SNVs, variants with a heteroplasmy lower than 1% may not be detected. Variants that are classified as pathogenic or likely pathogenic that overlap with the patient's phenotype, with levels of heteroplasmy of >5% will be reported. However, suspicious variants of uncertain clinical significance will only be reported if heteroplasmy levels are >20%. If a patient is identified in having a SNV with heteroplasmy of >20%, Sanger sequencing will be performed for sequence confirmation. Variants with heteroplasmy levels between 1% - 20% will not be confirmed. Variants are considered to be rare if present in asymptomatic adults in fewer than 5 families in mtDB and the RCIGM internal database, combined. The revised Cambridge Reference sequence is used as a reference (rCRS NC_012920). Interpretations are made with the assumption that any information provided on family relationship is accurate.

Both phenotype-informed and phenotype-agnostic analyses are performed. Likely pathogenic and pathogenic variants that may explain the patient's phenotype will be reported as related/possibly related to the patient phenotype. Selected variants of uncertain significance may be reported as well. Should an incidental finding be revealed during genomic analysis for proband, and proband and parents have opted-in to receive incidental findings, it will be included on the proband's report. Parents do not have the option to opt-in for incidental findings if proband has opted-out. Reported variants are curated and classified in accordance with the American College of Medical Genetics and Genomics Guidelines (PMID: 25741868, 21681106, 24071793, 31690835).

Incidental findings may be reported if the patient and/or patient's family do not opt-out of receiving these results. RCIGM's internal incidental finding policy includes the following: 1) variant must be classified as pathogenic per ACMG guidelines and in alignment with the known inheritance pattern of the genetic condition; 2) the variant is located in a gene with a well-established gene-disease relationship; 3) the gene and associated condition is shown to be medically actionable as established by RCIGM policy and in consultation with the RCIGM clinical team. Pathogenic variants included within the ACMG list of 59 secondary findings genes that are identified will be reported as well as other findings that meet the above criteria.

LIMITATIONS

Full coverage of the genome is not currently possible due to technically challenging repetitive elements and duplicated regions within the genome. Thus, not all regions of the genome are sequenced and/or uniquely aligned to the reference genome. Mosaic variant detection is limited using whole genome sequencing. Repeat expansion detection is limited to the DMPK and PHOX2B genes. The exact number of repeats cannot be determined by the current methodology and therefore orthogonal confirmation for precise sizing may be required. Non-diagnostic findings do not rule out the diagnosis of a genetic disorder since some genetic abnormalities may be undetectable with the current version of this test. False negative results may occur in the setting of bone marrow transplantation, recent blood transfusion, or suboptimal DNA quality. The chance of false positive or false negative result due to laboratory errors incurred during any phase of testing cannot be completely excluded. This test is set up to evaluate the potential contribution of rare

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disease-causing variants in known disease genes. It is not designed to evaluate for common variants in genes that might contribute to disease risk or for disorders that have a multigenic inheritance. Based on current knowledge, potential disease-causing variants may not always be recognized at the time of testing.

REGULATORY DISCLOSURES

This test was developed and its performance characteristics determined by the Rady Children's Hospital and the Rady Children's Institute for Genomic Medicine. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. RCIGM has established and verified the test's accuracy and precision as outlined in the requirements of CLIA '88. The test is used for clinical purposes. It should not be regarded as investigational or for research. The Rady Children's Institute for Genomic Medicine is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing.

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Test results reviewed and approved by:
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