UKAS MEDICAL
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Rare & Inherited Disease Genomic Laboratory
Great Ormond Street Hospital for Children NHS Foundation Trust
Levels 4-6 Barclay House, 37 Queen Square, London WC1N 3BH

Operational Lead & Head of Service (Cytogenetics): Deborah Morrogh FRCPath
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Great Ormond Street Hospital for Children NHS Foundation Trust

External Requesting CLINICIAN Dr Robin Lachmann Neurology & Neurosurgery National Hospital for Neurology & Neurosurgery Queen Square London WC1N 3BG	Patient Name: **YOUNG, Gaia Inigo** Patient DOB: **4/3/1996** Patient Gender: **Female** NHS Number: **601 541 0655** GOSH MRN: **945192** Family Number: **Z2066527** External Pat ID: **21365083**

REFERRAL REASON:

Likely inborn error of metabolism

Clinical Information: Alkalosis, hyponatremia, cerebral edema, and encephalopathy. A post-mortem spleen sample was received for testing but there was only one reference number on the sample tube (FB05699831) and no patient name or DOB, and paperwork with the reference number ODT151438. It is assumed that these reference numbers are unique to this patient.
Genotype: No pathogenic or likely pathogenic variants identified.

Result Summary
A molecular diagnosis has not been confirmed or excluded in Gaia Inigo YOUNG.

Result

Whole genome sequencing (WGS) has been carried out followed by analysis of the likely inborn error of metabolism panel, see technical information below. No clinically relevant sequence variants, copy number variants or short tandem repeats were identified in Gaia Inigo YOUNG.

Further testing

No further testing has been requested.

Reported by: Clare Beesley, Clinical Scientist	Date: 01/02/25
Authorised by: Emma Ashton, Principal Clinical Scientist	Date: 4/2/2025

Technical Information

Variant details

Only variants which are considered likely to be relevant to this patient's clinical phenotype are reported. Variants classified as benign, likely benign or uncertain significance, where further testing would not alter classification, or monoallelic variants in recessive genes are not reported.

Panel and coverage information

Clinical indication:	R98.2 Likely inborn error of metabolism
Family Structure	Singleton
Penetrance	N/A
Panel App Panel(s) and version(s):	Likely inborn error of metabolism v7.0
Gene list	https://nhsgms-panelapp.genomicsengland.co.uk/panels/467/v7.0
Target bases >15x:	99.4%
Average depth across target:	142.6

For details of the sequence coverage for individual gene(s) please contact the laboratory.

Test Information
Whole genome sequencing (Illumina) and analysis of variants prioritised by the Genomics England bioinformatics pipeline for the Genomic Medicine Service gene panel(s) listed above. Please note that the sensitivity of this test is limited by the types of detectable variants, regions of low read depth coverage and incomplete ascertainment of disease-gene associations.
This analysis only includes interrogation of Tier 1 and Tier 2 variants (ie variants in 'green' genes - confirmed clinically relevant genes). If required, Tier 3 variant analysis is restricted to relevant *de novo* variants and prioritised variants identified by Exomiser (https://www.sanger.ac.uk/tool/exomiser/). Known benign polymorphisms and sequence variants that are considered unlikely to be pathogenic are not reported.
Numbering starts with c.1 at the A of the ATG start codon. All nomenclature described using the Human Genome Variation Society (HGVS) guidelines (http://www.hgvs.org). Variants are classified using the ACMG/AMP guidelines (Richards et al 2015 Genet Med) and ACGS Best Practice Guidelines for Variant Classification in Rare Disease 2024 (Durkie et al. 2024 https://www.acgs.uk.com/media/12533/uk-practice-guidelines-for-variant-classification-v12-2024.pdf).
Classification of variants identified during the course of genomic testing may change over time; where this change affects the clinical impact of a variant an amended report will be issued.
NB: Results depend on samples being labelled correctly and referral information being accurate.

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Tissue specimen ID 24RG-131G0081 from Spleen Collected 22/7/2021 00:00 Received 10/5/2024 10:50 Authorised 4/2/2025 17:26 by Emma ASHTON, CS Priority Routine

Lab Comments

Referred by:
Dr Robin Lachmann
Email: r.lachmann@nhs.net