[redacted] Regional Genetics Laboratory

Our reference: [redacted]

Referring Clinician:

[redacted]

Copies to:

[redacted]

MOLECULAR ANALYSIS - Tissue

Patient's Name: [redacted]

Lab No **D23.23028**
Hosp No [redacted]
CG No
Date of Birth [redacted]

NHS No [redacted]
Date of Sample: **28/03/2023**
Date of Receipt: **30/03/2023**
Date of Report: **11/05/2023**

Referral: **Inherited metabolic disorders**
Reason: **Diagnosis**
Other Reasons:

REPORT

REPORT SUMMARY:

A genetic cause for the patient's disorder has not been identified.

Report interpretation:

[redacted] is clinically well but has previously had a liver transplant. A request has been received for sequencing analysis of genes associated with hyperammonaemia on DNA extracted from a biopsy of the donated liver to rule out a diagnosis of ornithine carbamoyltransferase (OTC) deficiency in the donor.

RESULT
Sequence analysis has not detected a pathogenic variant in the 42 genes screened.

INTERPRETATION OF THIS RESULT
A genetic cause of the clinical features seen in the donor has not been identified. However, this result does not exclude a genetic diagnosis.

RECOMMENDED ACTIONS
Differential diagnoses should be considered. These results are dependent upon the information supplied being correct and complete. If you wish to discuss this result further please contact this laboratory.

Basis of test:

Genes tested: R98.1 Slice 2 Hyperammonaemia. ACADM, ACADVL, ALDH18A1, ARG1, ASL, ASS1, AUH, BCKDHA, BCKDHB, CASA, CPS1, CPT1A, CPT2, DBT, ETFA, ETFB, ETFDH, GLUD1, HADHA, HADHB, HLCS, HMGCL, IVD, MLYCD, MMAA, MMAB, MUT, NAGS, OAT, OTC, PC, PCCA, PCCB, POLG, PYGM, SERAC1, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, TMEM70.

An Illumina NGS platform has been used to sequence coding and splice regions of a panel of genes captured by the Nonacus Cell3Target ExomeCG enrichment system. Data processing, variant calling and analysis using Congenica against reference human genome GRCh37 (hg19).

Consultant Clinical Scientists: [redacted]
[redacted] Regional Genetics Laboratory, [redacted]

Variants with a maximum population allele frequency >0.01 were excluded. The remaining variants were filtered on the basis of their Variant Effect Predictor (VEP) consequence. Further information is available on request.

100% of the target region has been covered to a minimum depth of 20X and regions below this minimum threshold have not been Sanger sequenced. Sequence nomenclature is according to HGVS guidelines. Variant interpretation and reporting is according to ACMG/ACGS guidelines (Richards et al., 2015 Genet. Med. 17:405-24, Ellard et al., 2020 www.ACGS.uk.com). DNA has been stored.

LIMITATIONS OF THE TEST:
This method is not capable of detecting exonic or whole gene deletion/duplications. Variants outside of exonic or splice site regions (+/-5 bp) may not be detected by this analysis.

[redacted] Regional Genetics Laboratory is a UKAS accredited medical laboratory, No[redacted] However, this assay using the Nonacus Cell3Target ExomeCG enrichment system and the Congenica analysis pipeline is not currently accredited.

Signed By: [redacted] [redacted] Registered Clinical Scientist