



Director

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Accredited Medical Laboratory
Reference No. 3098

Sheffield Children's
NHS Foundation Trust **NHS**

Sheffield Diagnostic Genetics Service
Sheffield Children's NHS Foundation Trust
Western Bank
SHEFFIELD
S10 2TH

www.sheffieldchildrens.nhs.uk/SDGS.htm

Expanded Newborn Screening (NBS) Pilot Referral information for genetic testing

CONDITION	ANALYSIS	PICK-UP RATE	REPORTING TIME
LCHADD (Long Chain Hydroxyacyl-CoA Dehydrogenase deficiency)	Common mutation c.1528G>C (G1528C)	60-87% mutation alleles (LCHADD + TFP deficiency)	3 working days
IVA (Isovaleric acidaemia)	Common 'benign' mutation c.941C>T (932C>T, A282V)	~47% mutation alleles in NBS presumptive positives	3 working days
GA1 (Glutaric Acidaemia Type 1)	Full gene sequencing	>95% mutation alleles	10 working days

Sample requirement

One of the following: -

- a) Dried Blood spot
- b) Blood – 0.5-1ml in K-EDTA
- c) DNA – 500ng

PLEASE INFORM THE LABORATORY PRIOR TO SENDING A SAMPLE TO ENSURE RAPID TESTING

For further details on any of the tests above, please do not hesitate to contact the laboratory

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Information correct until: 31st March 2014