

GAL 1-PUT

Synonyms

Galactose 1-phosphate uridyl transferase

Clinical Indication

GAL 1-PUT deficiency (classical galactosaemia) characteristically presents in the first week of life with failure to thrive, jaundice and hepatomegaly. The incidence in the UK is approximately 1 in 45,000.

Untreated babies are susceptible to E.coli sepsis and galactosaemia should be considered when jaundice, hepatomegaly and sepsis occur in a neonate.

Altered metabolism of galactose caused by deficiency activity of one of three enzymes results in elevated blood galactose concentration. Classical galactosaemia, caused by complete deficiency of galactose-1-phosphate uridyl transferase is the most common and severe type. The demonstration of nearly complete absence of galactose-1-phosphate uridyl transferase activity in red blood cells (RBCs) is the gold standard for diagnosis. Quantitative assay of RBC GAL-1-PUT activity may be affected for as long as three months by the transfusion of RBCs from a normal donor. In such cases, DNA testing may be necessary. Other tests that may be helpful in transfused infants include measurement of GAL-1-PUT activity in the RBCs of both parents to determine whether they are carriers and measurement of RBC galactose-1-P in the infant. If it is necessary to test the parents, please discuss this with a Biochemist on ext 3539/3025 prior to taking the samples.

Performed if requested by Consultant Paediatrician or agreed with Consultant Biochemist.

Part of Profile / See Also

Request Form

Combined Pathology manual blood request form or ICE request

Availability / Frequency of Analysis

Analysed by Chemical Pathology, Great Ormond Street Hospital if specific criteria met. [8692](#)

Turnaround Time

2-3 weeks

Patient Preparation

Sample Requirements

Specimen Type

Lithium heparin whole blood – DO NOT CENTRIFUGE OR SEPARATE

Volume

2 ml

Container



Paediatric green top (lithium-heparin) tube



Paediatric lithium heparin (orange top Sarstedt)



Or Green top (lithium-heparin) for adults when their child has had a recent transfusion and unable to provide a sample.

Please ensure that a separate sample is collected if other Biochemistry tests are required. Recommended to only be taken Monday to Thursday to allow sufficient time to reach the referral laboratory.

Reference Range & Units

15 – 35 $\mu\text{mol/h/gHb}$

Interferences

A high haemoglobin level may interfere with the test and cause false positive results. A false negative result may occur if the baby has recently had a blood transfusion and the test is not valid within 6 weeks of a major transfusion.

Interpretation & Clinical

Decision Value (if applicable)

Values below the reference range indicate partial or complete absence of galactose-1-uridyl transferase activity.

References

Galactosaemia: Clinical features and diagnosis – Up to Date – Searched Jan 2019.

Test code

GPUT

Lab Handling

DO NOT SEPARATE SAMPLE. Store as whole blood at 4°C in the referrals rack. Sent daily by courier to Great Ormond Street, London. Sample must reach the referral laboratory within 72 hours.



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