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 umol/h/gHb
Interferences	A high haemoglobin level may interfere with the test and cause false positive results. A false negative result may occur of 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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