

THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES UPDATE

Neonatal Dried Blood Spot (DBS) Total Galactose

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INTRODUCTION:

The quantitative determination of the total galactose is a primary screening test for galactosemia, an autosomal recessive disorder. This disorder is due to deficiency of enzymes involved in galactose metabolism pathway, including galactose-1-phosphate uridylyltransferase, galactokinase, uridine diphosphate galactose-4-epimerase, and galactose mutarotase. The complete or near-complete deficiency of these enzyme can be life-threatening if newborn is not managed timely, leading to failure to thrive, liver failure, sepsis, and death.

PRINCIPLE:

Fluorometric Enzyme Immunoassay.

SPECIMEN COLLECTION:

- Blood sample is collected on dried blood spot filter paper (DBS) by trained phlebotomist / nurses.
- For newborn screening blood samples is collected from the pricked heel of the baby 3-5 days after birth.
- The blood spot should be air-dried for at least 3 hours. Once dry, sample should be mailed to the laboratory within 24 hours in a separate paper envelope, along with humidity indicator card, desiccant pouch to protect against moisture.

SCHEDULE:

Performed on: First Thursday of the month (Cut-off: Thursday morning 8:00 am)

Reported on: Following Monday

PLEASE FILE FOR QUICK REFERENCE



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