

Department of Clinical Biochemistry

Core Clinical Services Directorate

Pathology Sciences Laboratory (Blood Sciences and Bristol Genetics) Southmead Hospital Westbury on Trym Bristol BS10 5NB

Tel: 0117 414 8425 Fax: 0117 414 8413

Email: nbn-tr.ClinicalBiochemistryNBT@nhs.net

Website: www.severnpathology.com

8th March 2018

Dear Colleague

Withdrawal of Urine Reducing Substances / Sugar Chromatography analysis

We have taken the decision to withdraw urine reducing substances (sugar chromatography) from our test repertoire here at Southmead Hospital in Bristol.

This test has been used historically as a first line screen in infants with prolonged/conjugated jaundice to detect galactosaemia and to diagnose other disorders of carbohydrate metabolism. Unfortunately the test is known to have limited sensitivity, and these disorders should not be ruled out on the basis of a negative reducing substances test result.

Classical Galactosaemia

The following alternative tests for galactosaemia provided by our laboratory are more specific (and have a shorter turn-around time):

- Galactosaemia screen (Beutler test) if baby not transfused. 1 mL whole blood lithium heparin tube.
- Urine Galactitol (first line test if baby has had a red cell transfusion). 5 mL urine plain container

Hereditary Fructose intolerance:

Urine reducing substances also has limitations when screening for the presence of fructose. Under UKGTN testing criteria, samples for genetic testing of the Aldolase B gene (Addenbrookes Hospital, Cambridge) are accepted based on clinical /family history, without a prerequisite for biochemical testing.

Should you have any queries relating to the removal of this service these can be directed to the Head of Biochemical Genetics, Consultant Chemical Pathologist, Dr Kemp on Helena.Kemp@nbt.nhs.uk.

Yours sincerely

Leila Cornes

Principal Clinical Scientist

Biochemical Genetics & Newborn Screening