

DATE:	28 November 2022
TO:	All Zones - Nephrologists, General Pediatricians and Laboratory Collection Staff
FROM:	Alberta Precision Laboratories, Genetics & Genomics – Biochemical Genetics
RE:	Test Menu Changes for Investigating and Monitoring Cystinuria

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Key Message

- Effective November 28, 2022, the following changes to the Biochemical Genetics Test Menu will occur regarding investigating patients for, as well as the ongoing monitoring of patients with, cystinuria:
 1. *Cystinuria Monitoring, Urine* testing, performed on either random or 24-hour urine collections, is now available. This test is for the specific monitoring of known patients with cystinuria, and reports only the cystine concentration in mmol/L, mmol/mol creatinine and mmol/d based on total volume collected (24-hour collections only).
 2. *Cystinuria/Homocystinuria Screen, Urine* testing is now available on either random or 24-hour collections. Abnormal screens will continue to trigger progressive testing of both Amino Acids Quantitation, Urine and Homocysteine, Total, Urine for further investigating the possibilities of cystinuria or homocystinuria using the existing patient specimen (as long as sufficient residual volume remains).
 3. The previous *Cystinuria Investigation* test on random collections has been discontinued.

Background

- Programmatic efforts to harmonize provincial testing practices, through consultation with several Alberta pediatric and adult nephrologists, resulted in test menu changes that align with published consensus recommendations for the diagnosis and monitoring of patients with cystinuria. The availability of the *Cystinuria/Homocystinuria Screen, Urine* and *Cystinuria Monitoring, Urine* tests on 24-hour collections will prevent the need to collect an additional random specimen from patients already undertaking a 24-hour collection for the investigation of stones, while testing on random collections still allow for simple patient screening or more frequent monitoring of known cystinuria patients, as desired.

How this will impact you

- These changes are reflected in Epic as well as on the new provincial Biochemical Genetics Requisition.
- Ordering physicians and laboratory staff performing collections need to be aware of these changes to ensure that the correct test is accurately ordered and properly collected.

Action Required

- For the initial investigation of cystinuria in patients under investigation for kidney stones, order *Cystinuria/Homocystinuria Screen, Urine* as part of the battery of testing performed. Abnormal screen results will trigger progressive testing as long as sufficient residual specimen is available.
- For monitoring known patients with cystinuria, order *Cystinuria Monitoring, Urine*.



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Effective November 28, 2022

Questions/Concerns

- Dr. Iveta Sosova, Laboratory Head, Biochemical Genetics Lab North
- Dr. David Sinasac, Laboratory Head, Biochemical Genetics Lab South

Approved by

- Dr. Dennis Bulman, Medical/Scientific Director, Provincial Genetics and Genomics Program, APL
- Dr. Carolyn O'Hara, Chief Medical Laboratory Officer (Interim), APL