Alberta Health Services

# Laboratory Bulletin

**Date:** August 12, 2014
**To:** All Zones
**From:** AHS Laboratory Services
**Re:** Changes to molecular testing for congenital adrenal hyperplasia

## PLEASE POST OR DISTRIBUTE AS WIDELY AS APPROPRIATE

**Key Messages:**
The Calgary Molecular Diagnostic Laboratory (MDL) currently performs molecular testing for congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency using a combination of allele-specific PCR and MLPA with reflex to sequencing of the CYP21A2 gene as appropriate. CAH due to 21-hydroxylase deficiency is thought to account for > 90% of cases of CAH.

Effective immediately, the Calgary MDL:

1. Will no longer perform allele-specific PCR for 21-hydroxylase deficiency as a first line test. Testing will consist of MLPA and CYP21A2 sequencing

AND

2. Will begin offering sequence analysis of CYP11B1 (encoding 11β-hydroxylase, the deficiency of which accounts for approximately 5-8% of cases of CAH).

Effective immediately, all samples received for confirmation of diagnosis of CAH with CYP21A2 sequencing and MLPA results ***not supportive*** of a diagnosis of CAH will be reflexed to 11β-hydroxylase testing. In addition, requests for 11β-hydroxylase deficiency testing as a first tier test will be considered if biochemical testing is suggestive of this diagnosis.

The analytical detection rate of sequencing and MLPA of the CYP21A2 gene is ~99%. The analytical detection rate of sequencing of the CYP11B1 gene is ~99%.

**Action Required:**
To initiate diagnostic testing for congenital adrenal hyperplasia, complete a Calgary Molecular Diagnostic Laboratory requisition in full and provide it to your patient to have their blood drawn (requisitions are available at www.albertahealthservices.ca/3310.asp)

For samples that previously tested negative for 21-hydroxylase mutations, the Calgary MDL will test samples for 11β-hydroxylase deficiency at the request of a health care provider if:

1. sufficient sample exists in the laboratory (complete a MDL requisition and fax it to the laboratory)
2. a repeat sample is received accompanied by a completed MDL requisition

**Inquiries and feedback may be directed to:**

- Please contact Allison Sluyters, Kim Gall or Charlotte Emmerson at 403-955-3097 if you have any questions regarding this change.

**This bulletin has been reviewed and approved by:**
**Dr. Martin Somerville, Medical / Scientific Director, Genetic Laboratory Services**