

Date: August 20, 2015
To: Central Zone, Edmonton Zone, North Zone
Pediatric Specialists and Subspecialists
From: AHS Laboratory Services, Genetic Laboratory Services - North
Re: Changes to Post-natal Testing for Turner Syndrome

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Key Message:

- As of June 25, 2015, karyotype analysis is the first line investigation for **Turner Syndrome** in the post-natal setting.

Background:

- Turner syndrome is a disorder that has distinct phenotypic and clinical features. It is caused by aberrations in karyotype, with loss of critical regions of the X chromosome.
- The genes associated with the physical features observed in Turner syndrome are mainly located on Xp (Xp11.2-p22); while loci contributing to ovarian function reside in Xq (Xq24).
- Rapid Aneuploidy Detection "RAD" (by QF-PCR) has replaced conventional cytogenetic analysis as a first line of investigation for certain aneuploidies: trisomy 21 (Down syndrome), trisomy 18 and trisomy 13. Additional markers on the sex chromosomes X and Y provide potential to detect the most common sex chromosome aneuploidies.
- RAD can detect "monosomy X" which is only one of the causes of Turner syndrome. The other karyotype aberrations that cause this condition are more complex.
- The utility of RAD as an investigation for Turner syndrome post-natally is limited.

Why this is important:

- RAD is not offered as an investigation for Turner syndrome in a post-natal setting because a normal RAD result cannot exclude a diagnosis of Turner Syndrome.

Action Required:

- The ordering physicians should complete a Cytogenetics Laboratory requisition requesting "karyotype analysis" for Turner syndrome. A blood sample in a sodium-heparin (green-top) tube is required.
- All EDTA samples received in the Edmonton Molecular Diagnostic Laboratory for Turner Syndrome testing will be discarded.

Inquiries and feedback may be directed to:

- Laboratory Genetic Counsellors at: 780-407-1015
- Dr. Stacey Hume, Molecular Geneticist, at: 780-407-1434
- Dr. Maisa Yoshimoto, Clinical Cytogeneticist, at: 780-407-1542

This bulletin has been reviewed and approved by:

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