Alberta Health Services

Laboratory Bulletin

Date: **December 3, 2013**

To: **Calgary Zone, South Zone and Central Zone (Three Hills, Hanna, Drumheller)**
Physicians, Laboratory Directors and Managers

From: **AHS Laboratory Services – Genetic Laboratory Services**

Re: **Change in Method for Prenatal Rapid Aneuploidy Detection**

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Key Messages:

- Effective immediately, Quantitative Fluorescence Polymerase Chain Reaction (QF-PCR) has replaced iFISH as the methodology used by the Calgary Cytogenetic Laboratory for prenatal Rapid Aneuploidy Detection (RAD).

Background:

- Rapid Aneuploidy Detection by Quantitative-Fluorescence PCR (QF-PCR RAD) is a prenatal testing option offered when a pregnancy is at increased risk for aneuploidy due to ultrasound findings and/or First Trimester Screening results (FTS).
- QF-PCR RAD provides the rapid detection of aneuploidies involving chromosomes 13, 18, 21, X and Y.

How this will impact you:

- This change applies to those individuals who qualify for prenatal diagnosis AND meet the criteria for RAD. Genetic Laboratory Services South at the Alberta Children's Hospital will accept prenatal samples for QF-PCR RAD in the following circumstances:
  - Risk of aneuploidy of 1/50 or greater or
  - Gestational age – 20 weeks or greater or
  - Documented ultrasound abnormalities (not isolated soft markers) consistent with a suspected chromosome aneuploidy where QF-PCR RAD results will influence decision making regarding management of the pregnancy.
  - All CVS samples received for cytogenetic testing.

- QF-PCR RAD is currently offered as an adjunct to standard cytogenetic analysis. QF-PCR RAD provides rapid results but has limitations:
  - QF-PCR RAD is limited to identifying aneuploidies of chromosomes 13, 18, 21, X & Y.
  - QF-PCR RAD cannot detect the presence of most structural abnormalities.
  - QF-PCR RAD cannot reliably detect mosaicism.
  - Maternal cell contamination above 20% can be detected by QF-PCR. If significant maternal cell contamination exists, the results will be reported as uninterpretable.
  - Approximately 2.7% of samples fail to produce an interpretable result using QF-PCR.

- Overall, the performance for the detection of aneuploidy of chromosomes 13, 18, 21, X and Y is equivalent to iFISH and will also allow for the detection of maternal cell contamination in both male and female fetuses.
- QF-PCR analysis is being performed in conjunction with the Molecular Diagnostic Laboratory.

Action required:

- To order QF-PCR RAD, please complete in full a Requisition for Constitutional Cytogenetics and FISH Studies.
- Indicate QF-PCR RAD studies on the requisition and specify the indication. This requisition must accompany the fetal sample. There is no change in the sample requirements.

Inquiries and feedback may be directed to:

- Contact Kim Gall or Allison Sluyters at: 403-955-3097

This bulletin has been reviewed and approved by:
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