Alberta Health Services

Laboratory Bulletin

Date: **March 17, 2016**
To: All Zones
From: Genetic Laboratory Services (GLS)
Re: **New Algorithm for Cytogenetic Investigation of Stillbirth or Fetal Loss**

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Key Messages:

- Effective **March 21, 2016**, rapid aneuploidy detection (RAD) and chromosomal microarray (CMA) will be offered by GLS Cytogenetic Laboratories in Edmonton and Calgary in a new algorithm for the genetic investigation of stillbirth or fetal loss.
- Fetal tissue will no longer routinely be cultured for cytogenetic investigation and any testing for cytogenetic anomalies will be via direct DNA-based testing (RAD or CMA see below).
- Tissues of fetal origin (eg. umbilical cord, skin, etc) are preferable to placenta for RAD and CMA testing.
- The inclusion criteria for cytogenetic testing on fetal tissue are:
    - Ultrasound anomaly or pathology suggestive of a chromosomal or contiguous gene disorder
    - Clinically significant ***unexplained*** growth abnormality
    - Unexplained stillbirth or neonatal death (≥ 20 weeks gestational age)
    - Family history of cytogenetic anomaly
    - A third and/or subsequent miscarriage(s)

Rapid aneuploidy detection (RAD):

- **Rapid aneuploidy detection (RAD) will be performed as the initial test on fetal tissue samples in lieu of karyotype.**
- RAD detects the most common aneuploidies (trisomy 13, trisomy 18, trisomy 21, and sex chromosome aneuploidies) and triploidy.
- If RAD result is abnormal and explains the clinical features, no further testing will occur.
- If RAD result is normal or does not account for the clinical features, residual material will be stored pending activation of CMA testing.
- Turnaround time is 3 weeks.

Chromosomal microarray (CMA):

- CMA detects chromosome imbalances (additional and/or missing information) at a greater resolution than standard karyotyping (chromosomal analysis).
- CMA may be ordered by a Clinical Geneticist, Obstetrician/Gynecologist, Pathologist
- A complete and signed CMA requisition must be received to initiate testing. The requisition may accompany the sample or be faxed to the lab at a later date. The clinical information on the requisition is critically important. Detailed clinical information allows for a more accurate interpretation of CMA findings. The mother's name should appear on the requisition accompanying the fetal tissue sample.
- **It is the responsibility of the health care provider to be aware of the limitations and unanticipated outcomes of CMA testing and where feasible to discuss these issues in detail with the family.**
  NOTE: Parental blood samples may be requested to help interpret fetal results.
- Turnaround time is 6 weeks after RAD results and activation of CMA.

Action Required:

- Refer to the Genetic Laboratory Services website (http://www.albertahealthservices.ca/lab/page8667.aspx) for requisitions, information sheets and site specific collection information.

Inquiries and feedback may be directed to Genetic Lab Services Genetic Counsellors:

- Edmonton: 780-407-1015
- Calgary: 403-955-3097

This bulletin has been reviewed and approved by:
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