

**Date:** February 8, 2021

**To:** Medical Genetics, Maternal Fetal Medicine (SHR region), Genetics Resource Center and all other related physicians and healthcare providers

**From:** Genomics Laboratory, Royal University Hospital, Saskatoon

**Re: Rapid Aneuploidy Detection (RAD) and Chromosomal Microarray (CMA) for specimens from fetal demise or stillborn at Saskatoon Genomics Laboratory**

## PLEASE POST AND DISTRIBUTE AS WIDELY AS APPROPRIATE

### Key Messages:

- Currently conventional karyotyping is performed on Products-of-Conception (POC) specimens from fetal demise or stillbirth. However, the unsuccessful cell culture rate is high due to the possibility of degraded tissue. DNA-based molecular test such as RAD and possible follow up with CMA provide a higher rate of diagnosis. These tests have been recommended to replace karyotype as the first line of testing for POC specimens in the recent CCMG-SOGC practice guideline (Armour CM et al., J Med Genet. 2018 55(4):215-221).
- Effective **February 16, 2021**, all POC specimens will be tested using the RAD assay and then possible follow-up by CMA at the Genomics Laboratory at Royal University Hospital, Saskatoon.
- POC specimens will **not** be cultured for chromosome analysis.
- The new testing algorithm for POC specimens is attached. A maternal peripheral blood sample is required to rule out Maternal Cell Contamination (MCC).
- Please complete the SHA Cytogenetics General Requisition and request RAD. If the RAD result is normal, CMA will be recommended for follow-up. Please complete the CMA requisition to request CMA testing.
- Specimen requirements:
  - Umbilical cord (preferred) or direct fetal tissue (e.g. muscle or thymus).
  - Fetal tissue must be fresh or frozen.
  - **Every** POC specimen must be sent to Anatomic Pathology for fetal tissue identification prior to any genomic testing.
  - Maternal peripheral blood sample in EDTA tube (2-7 mL)

### Why this change is important:

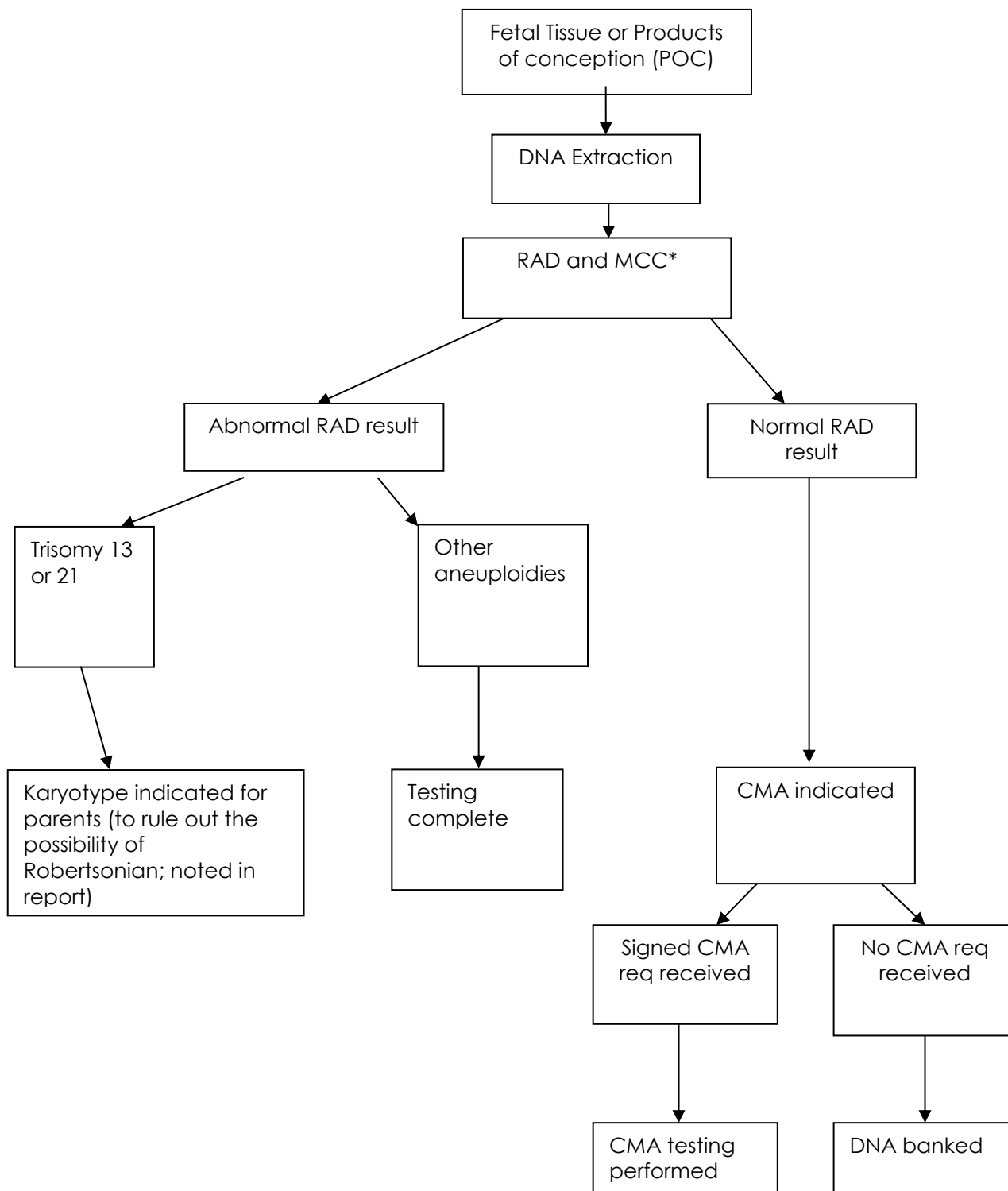
- In comparison with a karyotype, RAD in combination with CMA testing on POC specimens can provide detailed molecular genetics result and help to manage and guide future pregnancies.

### Inquiries and feedback may be directed to:

- RUH Genomics Laboratory, (306) 655-1706, [Cytogenetics@saskhealthauthority.ca](mailto:Cytogenetics@saskhealthauthority.ca)

For additional information regarding Laboratory Services in the former Saskatoon Health Region area, please refer to our website- [Laboratory Services Manual](#).

## Genetic Testing Algorithm for Fetal Tissue and Products of Conception (POC)



\* If MCC is detected, no additional molecular testing is available except CMA if MCC is low (< 20%).

\*\* If there is a suspected Mendelian or metabolic condition additional testing may be indicated. This should be noted on the requisition form to ensure culturing.

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