Laboratory Medicine Bulletin

[Saskatoon Genomics Laboratory]

Date: February 8, 2021

To: Medical Genetics, Neonatal Intensive Care Unit (JPCH), Genetics Resource Center and all other related physicians and healthcare providers

From: Genomics Laboratory, Royal University Hospital, Saskatoon

Re: Rapid Aneuploidy Detection (RAD) for Newborns now available at Saskatoon Genomics Laboratory

PLEASE POST AND DISTRIBUTE AS WIDELY AS APPROPRIATE

Key Messages:

- Effective **February 16, 2021**, the RUH Genomics Laboratory at Saskatoon will offer Rapid Aneuploidy Detection (RAD) for newborns.
- RAD should be considered in the following situations:
  - Newborn with suspected Down syndrome
  - Newborn with suspected trisomy 13 or 18
  - Newborn with ambiguous genitalia
- For Turner syndrome, karyotype remains the most appropriate investigation and RAD will not be performed.
- RAD detects the copy number of chromosomes 13, 18, 21 and sex chromosomes, X and Y. RAD does not detect structural abnormalities, nor low level mosaicism. It does not detect aneuploidies of chromosomes other than 13, 18, 21, X and Y.
- Chromosome studies will be recommended for abnormal RAD results to determine the etiology of the aneuploidy. Cases with normal RAD results being follow-up by Microarray may require additional studies based on those results.
- Specimen requirement:
  - 1-3 mL of blood in **EDTA** tube.

Why this change is important:

- RAD testing will provide a result in 2-3 business days, thus providing the opportunity should additional investigation and tests be required for timely patient management and care.

Inquiries and feedback may be directed to:

- RUH Genomics Laboratory, (306) 655-1706, Cytogenetics@saskhealthauthority.ca

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