Rapid Aneuploidy Detection (RAD) for Newborns: Edmonton and Northern Alberta

The Molecular Diagnostic Laboratory (MDL) offers Rapid Aneuploidy Detection (RAD) to look for aneuploidies involving chromosomes 13, 18, 21 and sex chromosomes, X and Y. RAD is now available as a replacement to having a preliminary karyotype performed. Routine karyotype will continue to be completed for these cases.

Aneuploidy: a variation in chromosome number that involves whole individual chromosomes rather than pieces of chromosomes

RAD should be considered a first line test in the following situations:

- Newborn with suspected Down syndrome
- Newborn with suspected trisomy 13 or 18
- Newborn with ambiguous genitalia

Testing by RAD will provide molecular results on aneuploidy involving chromosomes 13, 18, 21, X and Y
RAD will not detect aneuploidy of other chromosomes or structural rearrangements
RAD may not detect low level mosaicism (<15%)

Turner Syndrome:

- RAD will detect monosomy X which is only 1 cause of Turner syndrome
- A normal RAD result cannot exclude a diagnosis of Turner syndrome
- Karyotype remains the most appropriate investigation for Turner syndrome and RAD will not be performed

Turnaround Time:
RAD results will be available **2-3 business** days after sample receipt in the lab Routine karyotype results will be available **1-2 weeks** following sample receipt in the lab.

To order RAD:

- Submit **3 mL (min 1 mL) blood in an EDTA (mauve top) tube**
- Complete a Genetics Laboratories Requisition
- Order karyotype concurrently (**2 mL (min 1 mL) blood in sodium heparin (green top) tube** with a Genetics Laboratories Requisition)
- If sample volume is insufficient to send both EDTA and sodium heparin specimens at the same time, the EDTA specimen collection should be prioritized and sodium heparin specimen sent when available (exception is Turner syndrome – see above)

Requisition forms, contact information and other resources can be found at:
http://www.albertahealthservices.ca/8667.asp