

Rapid Aneuploidy Detection (RAD) in Calgary and Southern Alberta: Information for Ordering Physicians

This information sheet applies when patients are having invasive prenatal testing due to:

- An increased risk of aneuploidy,
- An increased risk of a single gene disorder or family history of a chromosome rearrangement
- Other (please contact the Cytogenetic Lab to discuss)

Testing on amniotic fluid and chorionic villus sampling (CVS) for patients having prenatal testing for the above indications may include:

- Rapid Aneuploidy Detection (RAD)
- Karyotype (for CVS samples to mitigate the risk of confined placental mosaicism **OR** amniocentesis samples where RAD has failed / is inconclusive **OR** for patients with a confirmed family history of an chromosome rearrangement)
- Single gene molecular testing, if indicated
- Maternal cell contamination study (MCC)

The majority of patients having amniocentesis will no longer have a fetal karyotype performed. RAD replaces standard karyotype for the majority of these patients.

NB: If fetal anomalies are present, refer the patient to Maternal Fetal Medicine or Prenatal Genetics to review options including prenatal chromosomal microarray (see below).

RAD is used for the rapid detection of triploidy, chromosomes 13, 18, 21 and sex chromosome aneuploidies.

Completing the Cytogenetic Requisition

To order RAD, send the sample with a completed Requisition for Constitutional Cytogenetic and FISH services. Request RAD in the 'other' section. Do NOT request karyotype unless indicated (for example, if the fetus is at risk of an unbalanced chromosome rearrangement).

Include detailed information on the requisition about the patient's risk factors (pregnancy history, family history, screening results, ultrasound findings). The information provided on the requisition is vital as it determines what testing the patient will receive.

If the fetus is at risk for a single gene disorder, consult with Prenatal Genetics.

RAD is the first line test on all prenatal samples. If RAD is abnormal, a karyotype will be completed.

Normal RAD
Results

- No karyotype unless indicated (see above)
- Single gene testing if indicated and requested
- Cell pellet stored for the duration of the pregnancy

NB: A cell pellet will be stored for the duration of the pregnancy for all prenatal samples received for the above indications to facilitate potential chromosomal microarray or single gene testing if indicated. If long term DNA banking is required, this must be specifically requested by completing a Molecular Diagnostic Laboratory (MDL) requisition along with the indication for DNA banking. Fax the completed MDL requisition to the Cytogenetic Laboratory

Maternal Cell Contamination (MCC)

Prenatal samples may be contaminated with maternal cells which prevents the interpretation of the results (nil result).

A maternal sample (EDTA) is required for when molecular testing is being performed.

How long will the results take?

- RAD results may take up to 2-3 days business days.
- Karyotype (if applicable) may take up to 2-3 weeks
- Single gene / molecular results (if applicable) may take up to 5 weeks or longer if sent out of province

Limitations of RAD

- RAD will not detect aneuploidy of chromosomes other than 13, 18, 21 and X and Y
- RAD will not detect structural rearrangements
- RAD may not detect low level mosaicism (<15%)

I am a health care provider and have questions. Who do I talk to?

Contact the laboratory genetic counsellors in Calgary at 403-955-3097

How do I refer to the Maternal Fetal Medicine Centre?

Call 403-289-9269 or fax 403-210-8381

How do I refer to the Prenatal Genetics Clinic?

Call 403-943-8375 or fax 403-943-8376

It is the ordering physician's responsibility to obtain the appropriate consent and discuss the points in this information sheet with their patient

**Requisition forms, contact information and other resources can be found at:
<http://www.albertahealthservices.ca/lab/Page8667.aspx>**