

SUMMARY

- ▶ **As of February 23, 2015, changes have been made to prenatal genetic testing offered by BCCH and BCWH Division of Laboratory Genetics, in order to align with standard of care in other Canadian provinces.**
- ▶ Rapid aneuploidy detection (RAD) by QF-PCR is performed on ALL patients who meet criteria for prenatal genetic testing (chorionic villus sampling (CVS) or amniocentesis) at BC Women's Hospital, regardless of indication.
- ▶ Chromosome microarray (CMA) is performed on those pregnancies with fetal structural anomalies consistent with a significant risk of chromosome anomalies other than aneuploidy of chromosomes 13, 18, 21, X and Y.
- ▶ Conventional chromosome analysis (karyotype) is only performed when indicated.
- ▶ **SOGC/CCMG Statement:** <http://sogc.org/wp-content/uploads/2013/01/gui265CPG1109E.pdf>

WHO IS IMPACTED

- ▶ All patients who meet criteria for prenatal genetic testing will receive Rapid Aneuploidy Detection.
 - Conventional cytogenetic analysis is no longer performed, unless indicated by RAD results or history.

DETAILS AND LIMITATIONS OF RAD

- ▶ RAD assesses only the specific chromosomes analyzed (13, 18, 21, X and Y);
 - Following a negative RAD result, the residual risk for a chromosomal abnormality of low, high, or unknown risk is estimated to be ~0.44% or ~1 in 227;
 - Remaining sample will be stored by the laboratory, in the event further genetic testing is indicated following detailed fetal ultrasound. In rare cases (insufficient sample, culture failure, etc.), a repeat procedure may be required.
- ▶ Maternal cell contamination may prevent the interpretation of results; karyotype will be performed in these cases.
- ▶ RAD does not provide a visual assessment of the chromosomes, thus, should aneuploidy be detected:
 - The interpretation provided is the *most likely* finding;
 - There may be other rare explanations for findings such as: Robertsonian translocation, marker chromosome, ring chromosome, unbalanced translocation, etc;
 - Karyotype analysis will be performed for assessment of recurrence risk;
- ▶ RAD cannot detect the presence of most structural abnormalities;
- ▶ RAD may not detect low level mosaicism.

DETAILS AND LIMITATIONS OF CMA

- ▶ The approach to analysis and reporting of prenatal CMA differs from postnatal CMA.
 - This approach reduces, but does not eliminate the detection of incidental findings and variants of unknown significance;
- ▶ This assay will not detect balanced chromosome rearrangements, regions not represented on the array, and may not detect low level mosaicism.
- ▶ This test may detect, but does not exclude uniparental disomy (UPD), and can detect identity by descent.

RESULTS REPORTING & PATIENT COUNSELLING

- ▶ Fetal gender will be provided on all reports; patients who do not wish to know the fetal gender should request that their physician(s) not disclose this information.
- ▶ Abnormal results will be reported to the Ordering Physician by phone.
 - Genetic counselling, by a recognized clinical genetics service, is recommended prior to any irreversible actions.
- ▶ All results (normal and abnormal) will be faxed and mailed to the Ordering Physician's office.