Genetic testing may include:
- Rapid Aneuploidy Detection (RAD)
- Chromosomal Microarray (CMA) when indicated and requested
- Maternal Cell Contamination (MCC)
- Single gene testing when indicated and requested

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

CMA allows for the detection of chromosome imbalances (gains or losses) that are smaller than can be detected by a standard karyotype.

Indications for genetic testing on fetal tissue:
- Ultrasound anomaly or pathology suggestive of a chromosomal or contiguous gene disorder
- Clinically significant unexplained growth abnormality
- Unexplained stillbirth or neonatal death (> 20 weeks gestational age)
- Family history of cytogenetic anomaly
- A third and/or subsequent miscarriage(s)

Sample requirements for CMA:
- Send directly to the Molecular Diagnostic Laboratory.
- Preferred tissue types include: direct fetal tissue (ex. thymus) or cord.
- Do NOT send placenta unless fetal tissue is unavailable
- Tissue must be fresh or fresh/frozen. Tissue cannot be fixed or paraffin embedded.
- A maternal blood sample (15mL EDTA) is required to assess for MCC (see box to right)

Maternal Cell Contamination (MCC)
Samples may be contaminated with maternal cells which prevents the interpretation of the results (nil result). A maternal sample may permit test interpretation in some contaminated samples.
A maternal blood sample must be collected (15mL EDTA) using a CMA requisition

Please note that culturing/karyotyping of perinatal tissue samples in no longer routinely performed.
How do I order RAD and CMA?
- To order RAD (without CMA) complete a Molecular Diagnostic Laboratory requisition
- To order RAD and CMA, submit a complete CMA requisition form

How long will the results take?
- RAD results will be available within 3 weeks
- CMA results will be available within 6 weeks of the test being initiated
- Please note: RAD reports are not available in Netcare

What are the benefits of CMA?
- Provides a detailed study of the chromosomes which may not have been previously possible
- May help to understand the cause of the pregnancy loss or stillbirth
- May help guide management and care of future pregnancies
- May identify couples at risk at increased risk of pregnancy loss or an abnormal liveborn with a chromosomal imbalance

What are the limitations and unanticipated outcomes of CMA?
- May find an imbalance unrelated to the pregnancy loss, but that indicates risk for other unanticipated health problems for the parents and family members (e.g., cancer, late onset neurological disease)
- Not all genetic conditions are detectable by CMA, as some are caused by mutations within a single gene or are multifactorial in nature
- Additional descriptions of result classifications can be found in the Chromosomal Microarray information sheet (http://www.albertahealthservices.ca/lab/page8667.aspx)
- A normal CMA result does not exclude all genetic causes of disease
- CMA testing may require testing of parents to help establish pattern of inheritance and clinical implications

I have CMA results, now what?
If an abnormal result is reported, it is appropriate to refer the family for genetic assessment and counseling. If the CMA result is normal, it does not mean that there is not a genetic cause for the pregnancy loss. Other genetic testing may be appropriate, in consultation with Clinical Genetics.

Referrals to Clinical Genetics can be sent to:
- Edmonton & North: Medical Genetics Clinic, Phone: 780-407-7333 Fax: 780-407-6845
- Calgary & South: R.B. Lowry Genetics Clinic, Phone: 403-955-7373 Fax: 403-955-2701

I have questions about array CMA. Who do I talk to?
Contact the laboratory genetic counselors:
- Edmonton at 780-407-1015
- Calgary at 403-955-3097

It is the ordering physician’s responsibility to obtain the appropriate consent and discuss the limitations and unanticipated outcomes of CMA with their patients where feasible.

Requisition forms, contact information and other resources can be found at:
http://www.albertahealthservices.ca/lab/page8667.aspx