



**Aneuploidy** is a variation in chromosome number that involves whole individual chromosomes rather than pieces of chromosomes. The most common aneuploidies in infants and young children involve chromosomes 13, 18, 21 and the sex chromosomes, X and Y.

**Rapid Aneuploidy Detection (RAD)** provides molecular results for aneuploidy involving chromosomes 13, 18, 21, X and Y.

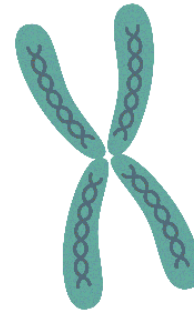
**Karyotype** is a cytogenetic test that characterizes chromosomal content including numerical and structural abnormalities and mosaicism.

*Indications for RAD and karyotype (order tests concurrently):*

- Newborns with suspected Down syndrome (trisomy 21)
- Newborns with suspected trisomy 13 or 18
- Newborns with ambiguous genitalia

*Indications for karyotype only (RAD will not be performed):*

- Newborns with suspected Turner syndrome



*Limitations:*

RAD will not detect aneuploidy of other chromosomes, structural abnormalities or mosaicism. Order karyotype concurrently with RAD for all indications except Turner syndrome.

*Turnaround times:*

- RAD 2-3 business days
- Karyotype STAT 1 week, Routine 4 weeks

### **Ordering Instructions:**

Please see [APL Test Directory](http://ahsweb.ca/lab/apl-td-lab-test-directory) (<http://ahsweb.ca/lab/apl-td-lab-test-directory>) for ordering instructions, requisitions and specimen requirements.

In situations where sample collection is difficult, prioritize the EDTA blood specimen and send the NaHep blood specimen when available.

If NIPS was performed during the pregnancy please include this information and the mother's name and healthcare number on the requisition. If prenatal RAD or karyotype was performed, aneuploidy testing in the newborn is not indicated unless there is a suspicion of mosaicism or phenotype is discordant with prenatal test results.

### **Contact Information:**

Genetic Counsellors, Genetics & Genomics, North (Edmonton) 780-407-1015

Genetic Counsellors, Genetics & Genomics, South (Calgary) 403-955-3097

Requisitions and other resources can be found at:

[Genetics and Genomics](http://ahsweb.ca/lab/if-lab-genetics-and-genomics) (<http://ahsweb.ca/lab/if-lab-genetics-and-genomics>)

[APL Test Directory](http://ahsweb.ca/lab/apl-td-lab-test-directory) (<http://ahsweb.ca/lab/apl-td-lab-test-directory>)