

# Metaphase Karyotyping

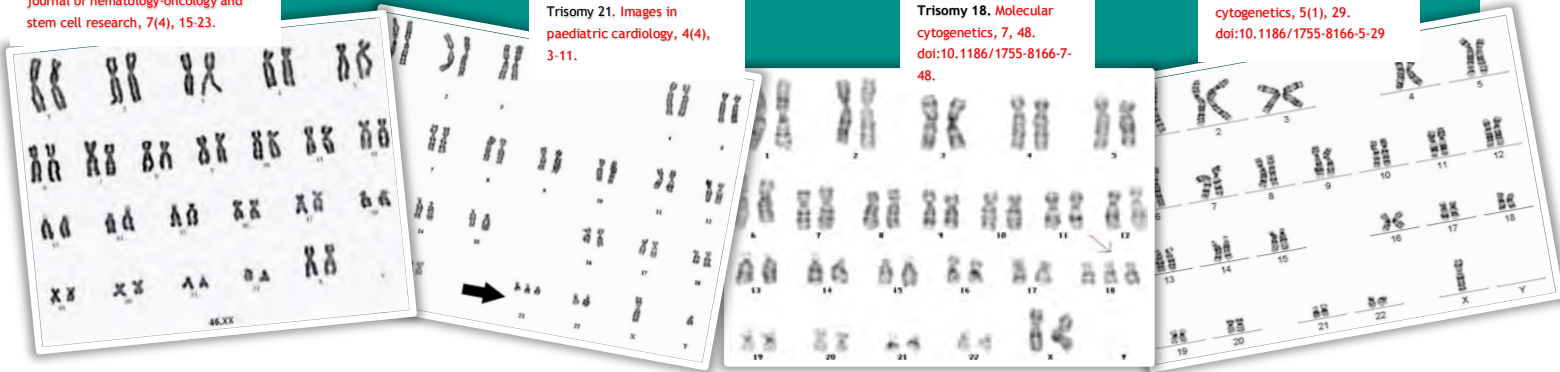
(Karyotype)

Normal Karyotype. *International journal of hematology-oncology and stem cell research*, 7(4), 15-23.

Trisomy 21. Images in *paediatric cardiology*, 4(4), 3-11.

Trisomy 18. *Molecular cytogenetics*, 7, 48. doi:10.1186/1755-8166-7-48.

Monosomy X. *Molecular cytogenetics*, 5(1), 29. doi:10.1186/1755-8166-5-29



## Detects:

- Whole chromosome aneuploidy
- Deletions
- Duplications
- Chromosomal rearrangements  $\geq 5$ -10 megabases

## Limitations:

- Turn-around time: ~2 weeks due to culturing, staining and scanning steps
- Unable to detect small Del/Dup and Single Nucleotide Variation



**Trisomy 21.** From Ferri F. Ferri's *Color Atlas and Text of Clinical Medicine*, 2nd ed. Philadelphia, PA: Elsevier; 2009

## Clinical Applications:

- Most Common Aneuploidies
  - Trisomy 13 (Patau Syndrome)
  - Trisomy 18 (Edward Syndrome)
  - Trisomy 21 (Down Syndrome)
  - Monosomy X (Turner Syndrome)
  - Klinefelter Syndrome (XXY)
  - XYY Syndrome



**Monosomy X.** Cura, D.O., Iscan, B., Gursoy, S., Guleryuz, H., Çankaya, T., Oztürk, T., & Bozkaya, O.G. (2017). A newborn with monosomy X in association with corpus callosum agenesis. *Pediatrics and neonatology*, 58 5, 455-457.