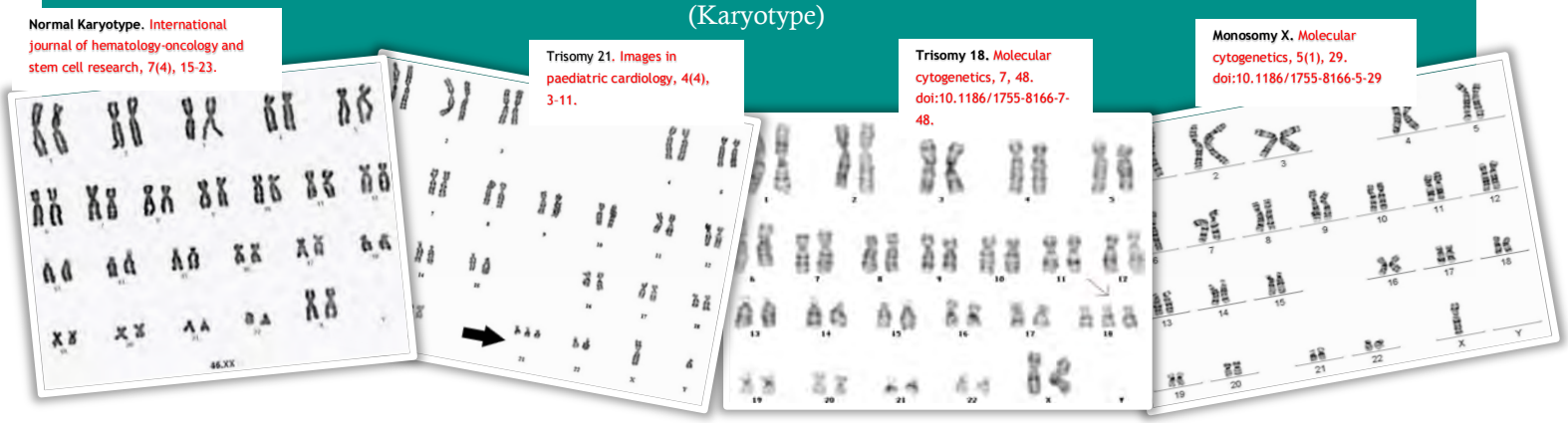
Metaphase Karyotyping

(Karyotype)



Detects:

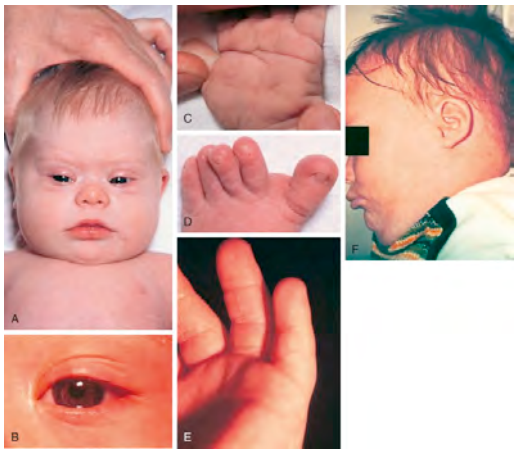
- Whole chromosome aneuploidy
- Deletions
- Duplications
- Chromosomal rearrangements ≥5-10 megabases

Limitations:

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

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



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

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