Karyographs = photo of chromosomes which reveals karyotypes. They reveal:

- If the correct numbers of chromosomes are present (diploid).
- If there are any extra or missing chromosomes (aneuploidy).
- The sex of the individual
- If there are extra or missing pieces of chromosomes

They are most commonly prepared from the cells of unborn babies harvested through amniocentesis.

Down Syndrome

Extra chromosome 21 = Down syndrome.

- Named after John Langdon Down – 1866
- 95% of all cases have 3 copies of the 21st chromosome
    - Also known as trisomy 21.
- 5% have a fragment of a third chromosome at 21.
- Extra 21st chromosome alters the course of development.
- Occurs in 1 in 800 live births.
- Occurs in 1 in 500 live births in poorer countries.
- Increased risk in Down syndrome the older the mother is.
- Old-fashioned name: mongalism

Features of Down syndrome

- Simian line – transverse palmar crease
- Flattened features
- Often larger than normal gap between big and second toe
- Macroglossia (enlarged tongue) – could be surgically corrected.
- Epicanthal folds (eyes)
- Brushfields spots – accumulation of lipids in the eye.
- Septal defects (atrial) – opening between left and right atrium
    - Hole in the heart
    - Not life threatening (generally)
- Septal defects (ventricular) – opening between left and right ventricle
    - Generally requires immediate surgery – or early on.

Patau Syndrome

- Extra chromosome 13 – trisomy 13.
- Much more severe than Down syndrome.
- Old-fashioned name: Cyclops.
- Incomplete brain development
- Physical and mental abnormalities – serious.
- Missing eyes
- Severe cleft palate
- Occurs in 1 in 10,000 births
- Condition varies in severity.
- Most babies don't survive much longer than 3 days.
- Not normally treated – allowed to pass away – no quality of life at all.