- Down's syndrome: non-disjunction happens in 21st pair of chromosomes; 3 instead of 2
- Consequences:
  - malformations of the digestive system
  - learning difficulties
- Most common chromosomal anomaly: affects 1 birth in 800

Karyotypes

- Karyotype = photograph of the chromosomes arranged according to a standard format
- Chromosomes placed in order according to their size and shape
- How a karyotype is made:
  - Cells stained and prepared on glass slide to see cells under light microscope
  - Photomicrograph images obtained from the chromosomes during mitotic metaphase
  - Images cut out and separated
  - Images of each pair placed in order by size and the position of their centromeres

Obtaining cells for karyotyping

- Unborn baby's cells can be obtained by amniocentesis or by removing them from chorionic villi
- Amniocentesis = extraction of amniotic fluid using a hypodermic needle
- Foetal cells are then grown in laboratory
- Used to determine chromosomal anomalies before birth

4.3. Theoretical genetics

Gregor Mendel

- Austrian monk
- 1865: publication of results of his experiment on garden peas
- Study of genetics of garden peas

Key terminology

- Genotype: Symbolic representation of a pair of alleles possessed by an organism, typically represented by two letters
- Phenotype: Characteristics or traits of an organism
- Dominant allele: Allele that has the same effect on the phenotype whether it is paired with the same allele or a different one. Dominant alleles are always expressed in a phenotype
- Recessive allele: Only has an effect on the if in homozygous state
- Codominant alleles: Pairs of alleles that both affect the phenotype when present in a heterozygote
- Locus: Particular position on homologous chromosomes of a gene
- Homozygous: Having two identical alleles of a gene
- Heterozygous: Having two different alleles of a gene
- Carrier: An individual who has a recessive allele of a gene that does not have an effect on their phenotype