Genetic Abnormalities
What IS the difference?

- “SYNDROME”:

- “DISORDER”:

- DISEASE”:
  → A condition of
Sex Chromosome Abnormalities

- do not cause as many problems as abnormalities with the autosomes

→ genes from the other 22 pairs of chromosomes are still present in affected individuals
How does this occur?

- **Error in Meiosis** – **NONDISJUNCTION**
  - when homologous chromosomes / chromatids **do not** separate properly during meiosis

(see text p. 178)
**Figure 4.20** Non-disjunction results in gametes with too many or too few chromosomes. Non-disjunction may take place during anaphase I (A) or anaphase II (B).
Karyotype from affected individual
Examples of Sex Chromosome Abnormalities

Turner Syndrome: XO
- has only one X chromosome
- genetically female but **no secondary sex characteristics** develop
  (e.g. breast development, menstruation)

Metafemale: XXX
- has three X chromosomes
- limited fertility
- often have learning disabilities
More Examples

**Klinefelter Syndrome: XXY**
- has extra X chromosome
- limited fertility
- excessive breast development
- sparse facial/body hair

**Jacob Syndrome: XYY**
- has extra Y chromosome
- taller than average
- more physically active
Autosome Abnormalities

Genetic defects may be transmitted as:

a) **A dominant allele**

- Examples:
  - Huntington’s disease (progressive nervous system degeneration)
  - Retinoblastoma (tumour in retina)
  - Achondroplasia (short-limbed dwarfism)
  - Hypercholesterolemi (high blood cholesterol levels leading to heart disease)
Autosome Abnormalities

b) A recessive allele

- **Examples:**
  - **Cystic Fibrosis** (affects function of mucous and sweat glands)
  - **Phenylketonuria – PKU** (unable to breakdown phenylalanine leads to development delays)
  - **Tay Sachs** (lack enzyme needed to breakdown fatty accumulation in brain and nerve cells)
  - **Adenosine deaminase (ADA) deficiency** (minimal immune response any susceptible to disease)
Other Genetic Abnormalities

Genetic defects may be transmitted as:

c) A sex–linked (on X or Y chromosome) allele

Examples:
- **Red–Green Colourblindness** (can not distinguish between red and green)
- **Duchenne Muscular Dystrophy** (progressive wasting of muscles)
- **Hemophilia** (blood clotting disorder)
Other Genetic Abnormalities

d) Interaction of many genes and/or with environmental factors

- **Examples:**
  - *Spina bifida* (neural tube does not close completely leading to paralysis of lower body)
  - *Asperger syndrome* (impaired communication and social interactions)
Other Genetic Abnormalities

e) **Presence of Extra or Missing Autosomes**

- **Examples:**
  - *Down syndrome* (flattened face, short stature in appearance and intellectual disabilities)
Example: Down’s Syndrome (Trisomy 21)
Other Genetic Abnormalities

e) **Errors in Chromosome Structure**

- **Examples:**
  - *Cri du chat* (high-pitched cry, developmental delays)
  - *Charcot Marie–Tooth Disease* (muscle weakness leads to loss of sensation in lower legs/feet and hands)
**NOTE:**

- Not all genetic defects are evident at birth:
  - **Cystic Fibrosis** – birth to 4 years
  - **Tay Sachs** – 6 months to 1 year
  - **Huntington’s Disease** – late 30’s and on

- Q: How do we determine AND possibly prevent these abnormalities from being passed on?
GENETIC SCREENING
Ways to do Genetic Screening:

- Screen potential carriers
- Pedigree Analysis
- Amniocentesis
- Chorionic villus sampling
- Ultrasound
- Fetoscopy
Chorionic Villus Sampling

- Procedure is completed **early in the pregnancy** at 9–11 weeks (1st trimester)

- Samples **tissue from the placenta**. (invasive) → cells do not need to be cultured therefore results take only a few days!

- Early in the pregnancy therefore a **higher risk** of miscarriage than amniocentesis.
Amniocentesis

- Procedure is completed later in pregnancy at 15–18 weeks (2nd trimester).

- A sample of the amniotic fluid is taken. → cells of the fetus are cultured (grown in the lab) therefore takes 2–3 weeks for results!

- Later in the pregnancy therefore risk of miscarriage is lower than C.V.S. but there is still a risk.
Comparison

Amniocentesis & Chorionic Villus Sampling

- Ultrasound scanner
- Placenta
- Amniotic fluid
- Fetus
- Uterus

(a) Amniocentesis

(b) Chorionic villus sampling
Ultrasound

- uses high frequency sound waves to reflect off fetus
  → produces black and white 3D image

- no known risks to humans
- limited by position of baby and visibility
Examples of Ultrasound
Fetoscopy

- direct view of fetus
  - uses tube with camera inserted into the uterus
- risk is greater than ultrasound
- image can be clearer and samples can be taken at same time for analysis
Understand the difference between CVS and amniocentesis:
→ procedure
→ advantages/disadvantages