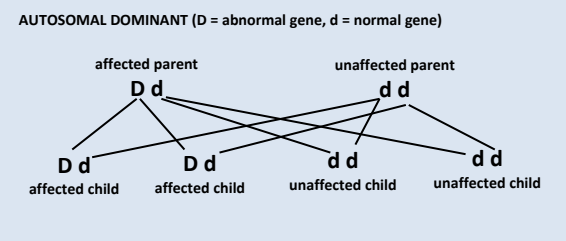
Basic Genetic Counselling

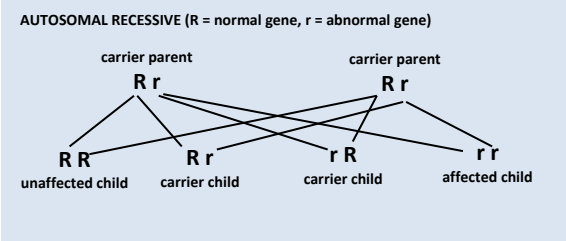
Possible stations

- Discuss antenatal screening for genetic abnormalities
- Explain to a newly pregnant mother about the tests for Down's syndrome
- Draw a pedigree for a family with an autosomal recessive condition (e.g. CF, sickle cell anaemia) or autosomal dominant condition (e.g. Huntington's, myotonic dystrophy) and discuss the risk of the patient having an affected child
- Explain genetic test results and the implications e.g. patient is a carrier for the CF gene

Inheritance risks

- **Autosomal dominant**: only need gene (from either parent) to have disease
  - If a parent is affected, there is a 1 in 2 chance of the child being affected
- **Autosomal recessive**: need two copies of gene (one from mother, one from father) to have disease and one copy to be a carrier
  - If one parent (only) is a carrier, there is a 1 in 2 chance of the child being a carrier
  - If one parent (only) is affected, the child will be a carrier
  - If one parent is affected and the other is a carrier, there is a 1 in 2 chance the child will be affected and a 1 in 2 chance the child will be a carrier
  - If both parents are carriers, there is a 1 in 4 chance of the child being affected and a 2 in 4 chance of the child being a carrier





Antenatal screening

<u>Conditions of interest</u>

- **Familial (inherited) genetic conditions** e.g. CF, haemophilia, muscular dystrophy, sickle cell, thalassemia
- **Developmental abnormalities** (not genetic) e.g. neural tube defects like spina bifida, other structural developmental defects
- **Chromosomal abnormalities** (caused by cell division error – 'genetic' but not usually inherited) e.g. Down's syndrome – risk increases with age

<u>Parental blood tests</u>

- Genetic tests of mother and father can be performed to determine exact risk of baby being affected by a familial (inherited) genetic condition
- If there is a significant risk to the baby, invasive testing is offered

<u>Down's syndrome risk screening</u>

- **'Combined test'** scan + blood test (11-14 weeks) – *in most cases (better)*
  - Nuchal translucency scan
  - Blood test: PaPP, ↑βHCG
- **Quadruple blood test** (15-17 weeks): ↓αFP, ↓unconjugated estradiol, ↑βHCG, ↑inhibin A
- **Integrated** (both of above) – *best*
- Tests give a <u>risk value</u> – if risk >1in150, invasive testing is offered
- NOTE very rarely, a parent can have a balanced translocation of chromosome 21 that can cause 'translocation Down's syndrome' – if they've had a baby with translocation Down's syndrome, the parents should be tested for the abnormality

<u>Neural tube defect screening</u>

- **Blood test** ↑αFP (15-17 weeks) – *gives a risk value. Fetal blood from amniocentesis can also be used and is more accurate*
- **Anomaly scan** (20 weeks) – *confirms*

<u>Invasive testing for genetic condition diagnosis</u>

- **Amniocentesis** (>15 weeks) – 1% miscarriage risk
- **Chorionic villus sampling** (10-15 weeks) – 1-2% miscarriage risk
- These tests give a definitive answer if child has a certain genetic condition – results take 1-2 weeks but rapid tests for chromosome abnormalities can be done in 3 days
- They can be performed for: high risk of Down's syndrome, familial genetic conditions above
- Termination can be performed at any time if there is confirmed genetic abnormality but is usually done at 18-20 weeks

Drawing a pedigree

Key

- Sex
  - Male □
  - Female ○
  - Sex undetermined ◇
  - Pregnancy Ⓟ
- Conditions
  - Affected ■●
  - Carrier for autosomal recessive condition ◨◑
  - Carrier for X-linked recessive condition ◉
- Matings
  - Mating □—○
  - Divorced/separated □-//-○
  - Two matings □-//-○—□
- Patient □↗
- Siblings
  - Siblings
  - Non-identical twins
  - Identical twins

EXAMPLE – mother wants to know the chance of her child being affected with Huntington's chorea (autosomal dominant) which runs in her husband's family