Genetics and Primary Immunodeficiency

Primary immunodeficiencies (PIDs) are rare genetic disorders that impair the immune system. Over 480 PIDs exist, with genetic mutations passed down in different inheritance patterns.

Genetic Inheritance Patterns

- **X-linked:** Mainly affects males. Females are usually carriers.
- **Autosomal Recessive:** Both parents must carry the gene for a child to be affected.
- **Autosomal Dominant:** One copy of the mutated gene causes the disorder.

Genetic Diagnosis and Testing

- Advances in genetic testing have made it possible to identify the underlying mutations in many immunodeficiencies, improving diagnosis, treatment options, and family planning.
- Prenatal testing is available for some PIDs, offering options such as DNA analysis, foetal blood sampling, and enzyme analysis to assess the likelihood of a child inheriting an immunodeficiency.
- Newborn screening for some PIDs is being investigated in the UK.

The Importance of Genetic Counselling

Understanding genetic risks and testing options is essential for families. Genetic counselling provides crucial support to help families make informed decisions about testing, treatment, and family planning.