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Prenatal and Neonatal Testing

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Chapter summary

- The principles of pre- and neo-natal testing are outlined, together with a description of the direct (invasive) methods that are used for obtaining samples from the foetus and newborn. These include amniocentesis, CVS, PUBS, and heel-prick blood sampling.
- Non-invasive sampling techniques are explained: where information can be gained about a foetus from maternal serum sampling, without increased risk to the pregnancy. The presence and implications of certain protein markers, foetal cells, and cffDNA in maternal serum are discussed.
- Links are made with several other chapters to highlight the molecular techniques that are used to analyse and process pre- and neo-natal samples in the laboratory.
- A wide range of applications for pre- and neo-natal testing are discussed, including single-gene disorders, aneuploidies, disorders arising from structural damage to chromosomes, structural defects of the foetus, and metabolic diseases.
- The ethical concerns that surround prenatal testing are also discussed.