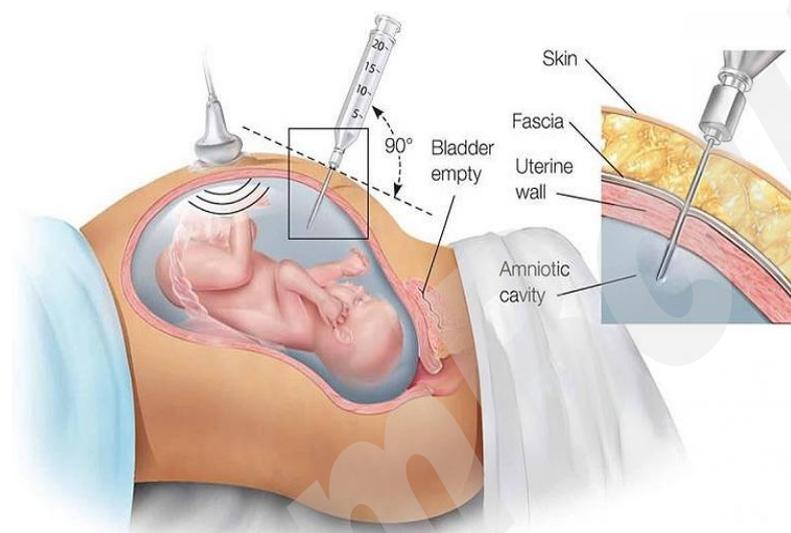
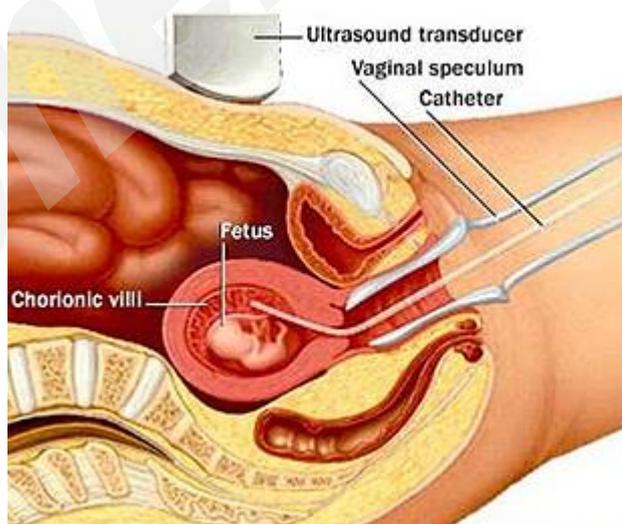


“Non-invasive Prenatal Testing: The Future is Now.”

Reviews in Obstetrics & Gynaecology 6(2): Prenatal detection of chromosome abnormalities has been offered for more than 40 years, first by **amniocentesis** in the early 1970s and additionally by **chorionic villus sampling (CVS)** in the early 1980s. This has drastically reduced the incidence of aneuploid children born to older mothers.



Amniocentesis



Chorionic Villus Sampling

Glossary

DNA: A molecule that carries the genetic instructions used in the growth, development, functioning and reproduction of all known living organisms.

Chromosome: A DNA molecule with genetic material of an organism.

Amniocentesis: a medical procedure used in prenatal diagnosis of chromosomal abnormalities and gender determination in which a small amount of amniotic fluid which contains foetal tissues is sampled from the amniotic sacs surrounding a developing foetus, and the foetal DNA is examined for genetic abnormalities.

Chorionic villus sampling: It entails sampling of the chorionic villus (placental tissue) and testing it for chromosomal abnormalities.

Aneuploid: The presence of an abnormal number of chromosomes in a cell, for example a human cell having 45 or 47 chromosomes instead of the usual 46.

Amniocentesis and CVS are invasive prenatal diagnosis; these procedures carry a small but finite risk and would ultimately cause miscarriages than they would detect aneuploidy. For this reason, a NEW, NON-INVASIVE test has been developed – **Cell/DNA Trafficking in Pregnancy.**

What is Cell/DNA Trafficking in Pregnancy?

Throughout pregnancy, foetal cells are constantly trafficking across the placenta into the maternal circulation and vice versa. It is estimated that, after first trimester, there is approximately 1 foetal cell in the maternal circulation for every 10,000 to 1 million maternal cells → 20 foetal cells in 20 ml of maternal blood. Given their scarcity, effort to isolate and purify these foetal cells for subsequent analysis has been largely unsuccessful, for this reason, attention has turned to cell-free DNA.

What is cell-free DNA (cfDNA)?

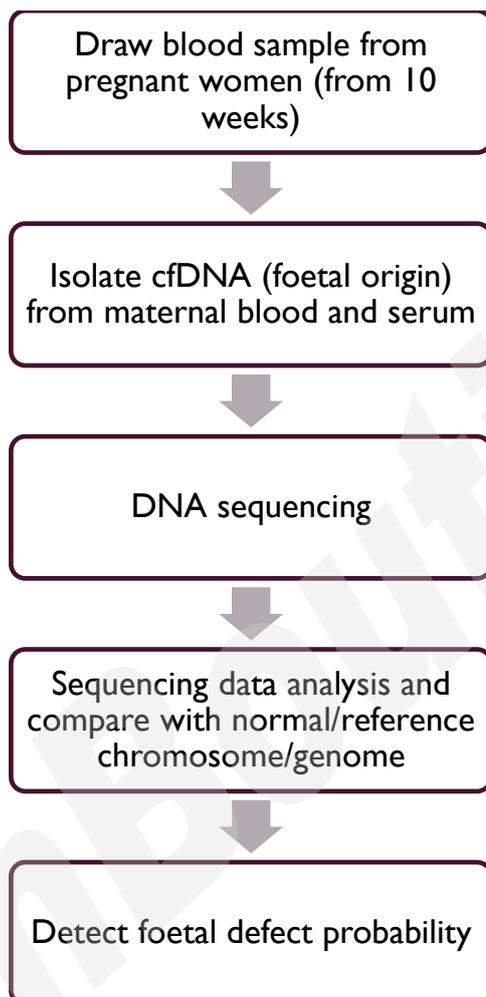
cfDNA in maternal blood is comprised of extracellular DNA fragments that can be found in the maternal plasma and serum. The majority of cfDNA in maternal circulation is of maternal origin and around 10% is of foetal origin. cfDNA is released into the maternal circulation from cells of the placenta. It can be detected very early in pregnancy and is cleared a few hours after birth.

What is genomic sequencing?

Genomic sequencing: high throughput DNA sequencing technology that can sequence millions of DNA molecules in parallel. For prenatal testing, both cfDNA of maternal and foetal origin present in maternal blood are sequence and these fragments are mapped to a reference/normal chromosome/genome.

What are the genetic inherited diseases that can be detected with this method?

- **Down syndrome**
Symptoms: flat face, short neck, intellectual disability
- **Edwards syndrome**
Symptoms: low birth weight, a small, abnormally shaped head, a small jaw and mouth, low sets ears, severe learning disabilities, short life span, etc.
- **Patau syndrome**
Symptoms: severe heart defects, brain doesn't divide into two halves, absence of one or both eyes, smaller than normal head size, deafness, etc.
- **Turner syndrome**: affects development in **females**
Symptoms: short stature, early loss of ovarian function, infertile, webbed neck, swelling of the hands and feet, skeletal abnormalities
- **Klinefelter syndrome**: affect male physical and cognitive development
Symptoms: small testes, do not produce much testosterone, infertility

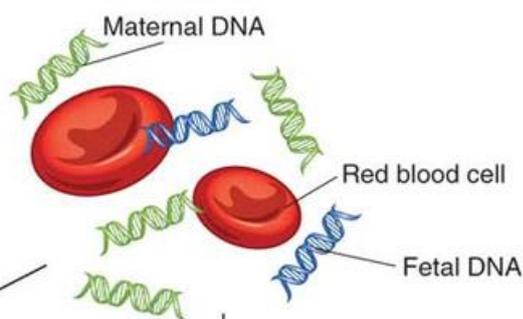


Cell-free DNA in placenta



Monogenic disorders

Cell-free DNA in maternal plasma



Aneuploidies

If you found this article interesting, you are interested in Genetics!

Genetics involves the application of fundamental genetics, genomics, evolutionary population and ecological genetics to all areas of biology, biomedical sciences and biotechnology. You will develop knowledge and skills in the theory of genetics and molecular biology, population genetics and evolution and in experimental design, data recording and analysis and scientific writing, which are essential preparation for roles in universities, research institutes, government departments, hospitals and in the biotechnology industry.

What are the entry requirements for Genetics degrees?

Minimum of AAB/ABB/ABC in A-Level:

- Chemistry
- Biology
- Physics
- Mathematics

What are the course structure and assessment methods?

Genetics course is broad in scope, with topics including: evidence for evolution, heritability and population growth, population genetics, molecular evolution, adaptive behaviours and sexual selection speciation and phylogenetics and applied evolution. Assessment is based on tutorials, practical, written exam, oral and written presentation and research projects.

What careers are possible with a Genetics Degree?

Graduates from Genetics go into a range of careers:

Academic Genetics: Teaching, work in scientific research and development (may require significant postgraduate study, usually a PhD)

Industrial Genetics: Research and development scientist (agriculture, biotechnology, ecology and conservation, medical genetics companies)

Top Ten Universities (2017) that specialise in Genetics:

1. Harvard University
2. Massachusetts Institute of Technology (MIT)
3. University of California, San Francisco (UCSF)
4. University of Cambridge
5. Stanford University
6. University of Washington
7. University of Oxford
8. Johns Hopkins University
9. University of California, San Diego (UCSD)
10. University of Toronto



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