Comparison of two types of Genetic Tests: Gene and Chromosome Tests

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	Gene Test	Chromosome Test
Function	Detect changes in the **genetic material** (including DNA and RNA) of a **gene**.	Detect changes in the **number and structural** appearance of the **chromosomes**
Techniques or Methods	• **Mutation Detection**: Identifying a **known fault** (mutation) in the gene. • **DNA Sequencing**: Identify an **unknown** fault (mutation) in a gene.	• **Karyotype:** Using a microscope to view and analyse **all the chromosomes** in a cell • **qPCR:** Identify the presence of **certain chromosomes**. • **FISH:** Identify **regions of certain chromosomes**.
Sample type and products used for testing	• Blood sample • DNA extracted directly from the blood	• Blood sample, amniotic fluid (amniocentesis), placenta cells (chorionic villus sampling) • Cells are grown and chromosomes viewed
Timing*	• Known mutation (6-8 weeks) • Gene sequencing (Months)	• Cell cultures for karyotype and FISH (4 weeks) • Quick test for certain chromosomes(72 hours)
Examples and Uses	• Determine the presence of a genetic condition, such as cystic fibrosis. • Diagnostic, Prenatal, Carrier, Predictive and PGD testing.	• Determine the presence of a chromosomal abnormality such as Down syndrome • Diagnostic, Prenatal and Carrier testing.