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Providing knowledge

Guidance on the possibilities for genetic testing before fertility treatment

FERTILITY

What is a gene?

A gene is a physical and fundamental unit of the DNA strand.
The DNA strand is a molecule, which contains the genetic elements that are of vital importance to the growth, reproduction and general, correct functioning of our bodies.

Genes contain the information that determines your traits and the characteristics that you have inherited from your parents.
For example, if your mother has freckles, you may also have freckles, because you have inherited the trait for freckles.

A gene is a piece of the DNA strand.

Genes carry the information that determines which traits or characteristics are passed on to you, and which traits or characteristics you pass on to your children.

Why do genetic diseases occur?

When a cell divides and becomes two new cells, all the genes of that cell must be duplicated and distributed correctly in the two new cells. If an error occurs during this process, it may result in the destruction and mutation of a gene.

Most of the errors that occur are of little or no importance. Some errors, however, can significantly change the gene and prevent the gene from functioning properly. These changes are the pathogenic mutations that cause genetic disease.

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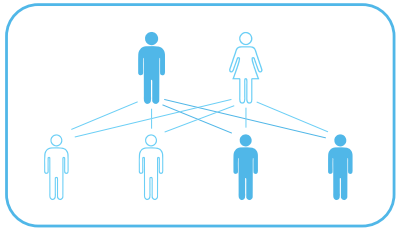
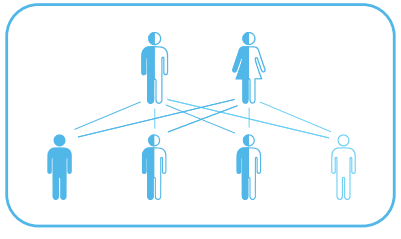
What is a genetic disease?

A genetic disease is a disorder which may originate from one single gene. Humans have around 25,000 genes, and all genes are important for our bodies to work properly.

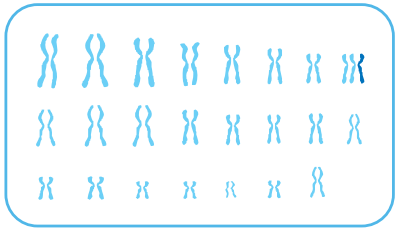
On the next three pages we review three types of genetic diseases:



Dominant



Recessive



Chromosomal

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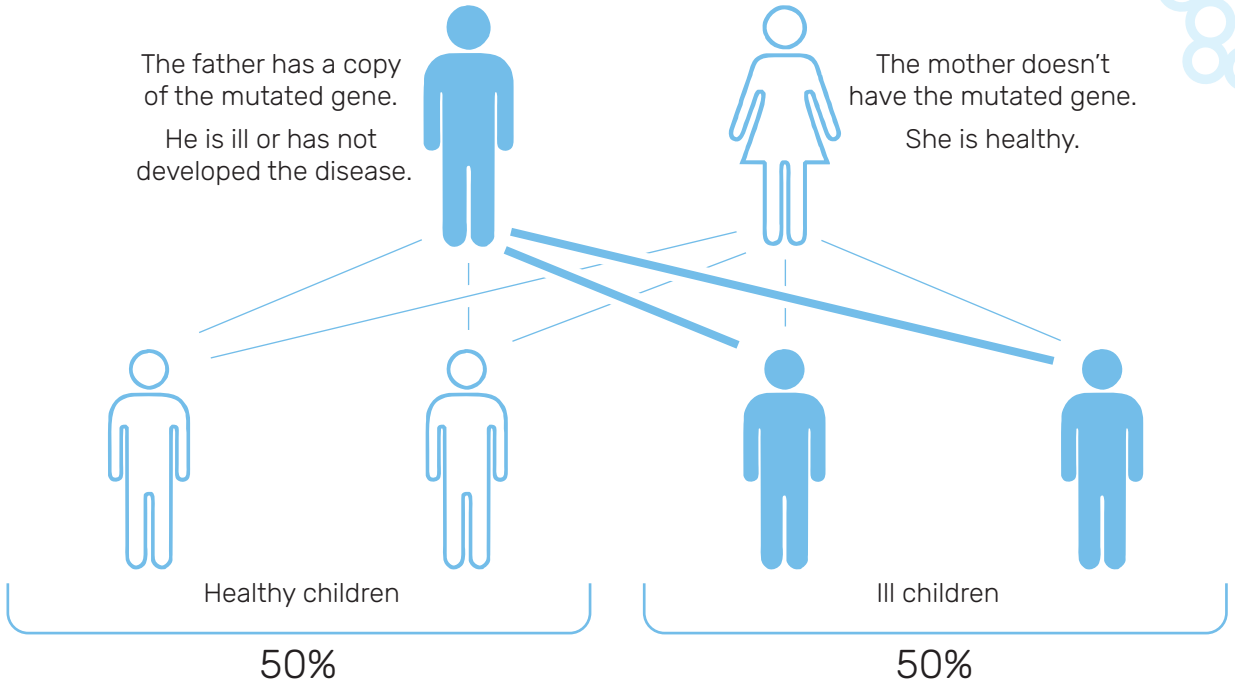
Type of genetic disease: **Dominant**

A single copy of the mutated gene in one parent causes the disease.



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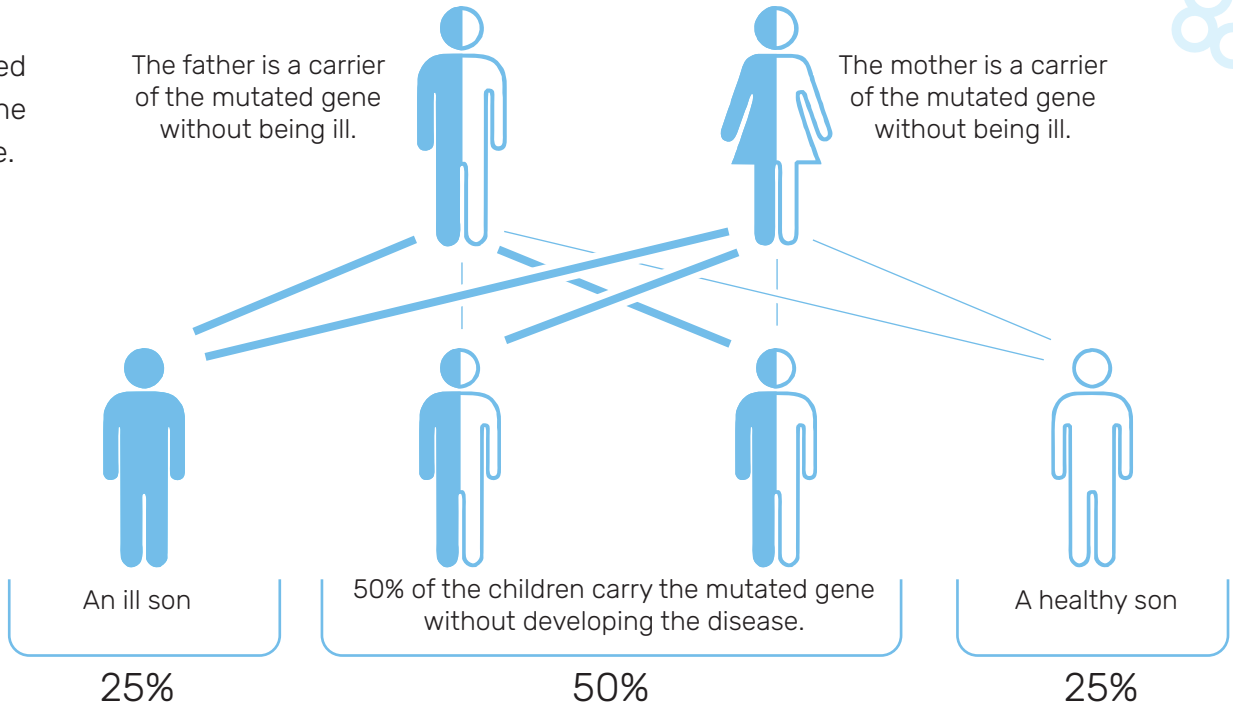
Type of genetic disease: **Recessive**

Two copies of the same mutated gene, one in the mother and one in the father, cause the disease.



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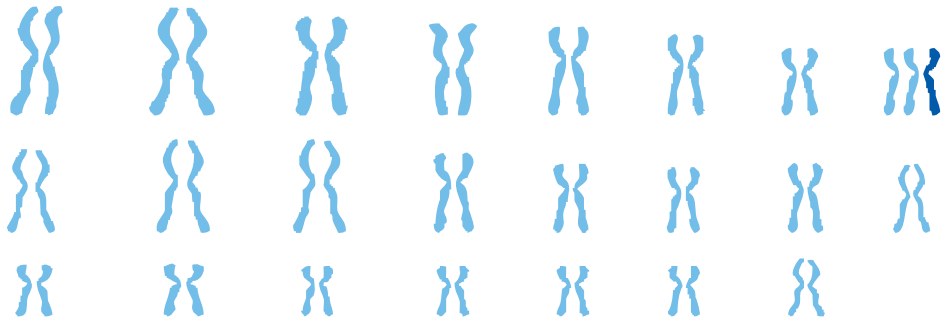
Type of genetic disease: **Chromosomal**

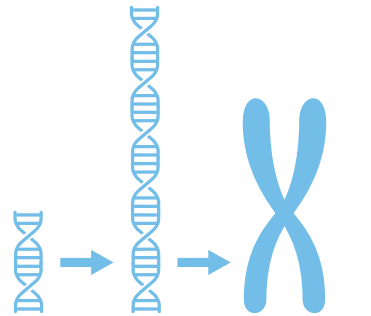
Normally we have 23 chromosome pairs, equal to 46 chromosomes in total. One half of the chromosomes come from the mother, and the other half from the father.

Genetic chromosomal diseases occur, when there are more or less than 46 chromosomes, illustrated here with an extra dark blue chromosome.



What is a chromosome?

A chromosome consists of tightly packed DNA.



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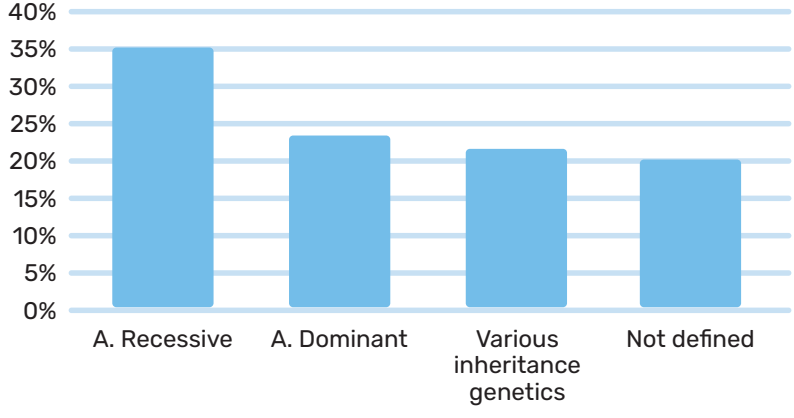
Can healthy people transmit a genetic disease?

You have inherited your genetic structure from your mother and father.
Parents can be healthy carriers of a mutation without knowing.
If a disease-causing mutation is passed on to the next generation
from both mother and father, there is a risk that the child will become ill.

35% of genetic diseases have a recessive genetic pattern.

Distribution of inheritance patterns of rare genetic diseases



Source: https://www.researchgate.net/publication/335846919_Estimating_cumulative_point_prevalence_of_rare_diseases_analysis_of_the_Orphanet_database

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Can failed fertility treatments be caused by a genetic problem?

One of the reasons why fertility treatments may fail is the presence of genetic mutations, that can lead to repeat abortions and unsuccessful treatments.

Why undergo a genetic test before fertility treatment?

Genetic testing is used to investigate whether there is a risk of having a child with an inherited genetic disease. Hereditary diseases are inherited through our genetic material, from one generation to the next.
In some cases, diseases can “skip” one generation and be inherited in the next; this happens in diseases with recessive inheritance.
Recessive inheritance means that a person can carry disease-related mutations without being aware of it.

Genetic testing plays an important role in fertility treatments. Both patient and donor can be healthy carriers of diseases with a genetic mutation in the same gene.

A genetic disease may occur in a fertility treatment when:

- Both egg donor and sperm donor have a genetic mutation in the same gene.
- The chosen sperm donor has a genetic mutation in the same gene as the woman.
- The chosen egg donor has a genetic mutation in the same gene as the man.

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Choose between two test packages:

Amplexa Genetics offers two test packages: Genes**2**Life and Genes**4**Life. The main difference between the packages is the number of genes investigated.

We need a blood or saliva sample for the test.

The test result is available within 20 working days.

	Genes**2**Life	Genes**4**Life
Test Type	NGS basic, recommended by ACOG*	NGS extended, includes Genes2Life
Range	96 genes	482 genes
Carrier probability	1 in 100 couples at risk	2 in 100 couples at risk
Diseases investigated	+ 100 diseases	+ 400 diseases

Both tests are genetic tests and are performed using NGS (Next Generation Sequencing). This technology enables us to investigate if there are mutations in genes associated with a known disease. The tests will show if you are a carrier of one of these mutations.

* (ACOG = American College of Obstetricians and Gynecologists)

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Which diseases will be investigated?

Types of genetic diseases investigated with Genes2Life and Genes4Life:

Genetic diseases	Statistics
Spinal muscular atrophy	1 in 6,000-10,000 children is born with the disease. In the United Kingdom, approx. 2,000-2,500 children and adults with SMA
Cystic fibrosis	Approx. 1 in 30 people is a healthy carrier of cystic fibrosis without knowing it. In Denmark 1-2 children are born with cystic fibrosis every month This represents approx. 150,000 people in Denmark.
Lysosomal diseases such as: Tay-Sachs, Sandhoffs, Niemann, Gauchers, Mannosidosis, Fucosidosis, Sialidosis, Hurler, Hunter, Sanfilippo, Morquios, I-cell, Sallas, Cystinuria Wolman	Approx. 70 different lysosomal diseases. Each of these is very rare, but as a group they affect about 1 in 5,000-8,000 newborns. Every year about 10 children are born with one of these diagnoses in Denmark.

Genetic testing plays an important role in fertility treatments

Both patient and donor can be healthy carriers
of diseases and have a genetic mutation in the same gene.

Get tested before:

- Trying to get pregnant.
- Starting an assisted fertilization treatment.
- Starting a treatment with donor eggs or sperm.

How to do it?

Sample collection
of blood or saliva.

Gene analysis
of the liquid.

Genetic report
for each sample.

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