Amplexa Genetics®

Providing knowledge

What you need to know:

Genetic testing before pregnancy

What is a genetic disease?

A genetic disease is a disorder which may originate from one single gene.

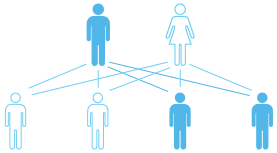
A gene is a piece of the DNA strand. Genes carry the information that determines which traits or characteristics are passed on to you, and which traits or characteristics you pass on to your children.

Humans have around 25,000 genes, and all genes are important for our bodies to work properly.

Types of genetic diseases

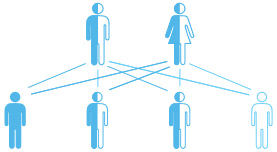
Dominant

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



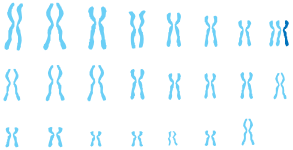
Recessive

Two copies of the same mutated gene, one from the woman and one from the man, cause the disease.



Chromosomal

Chromosomal diseases occur, when there are more or less than 46 chromosomes.

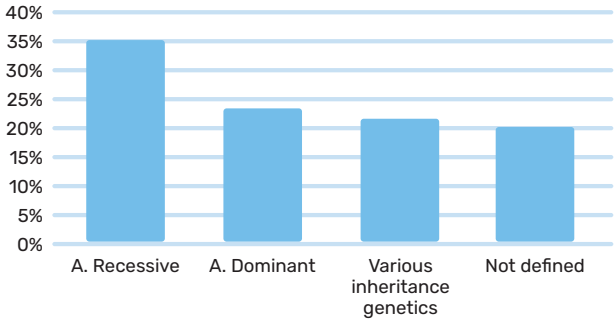


Can a healthy person transmit a genetic disease?

Anybody can be a healthy carrier of a mutation without knowing.
If the man and the woman are carrying a disease-causing mutation, there is a risk of passing it on to the next generation and then the child will be sick.

35% of genetic diseases have a recessive genetic pattern.

Distribution of inheritance patterns of genetic rare diseases



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

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 repeated abortions and unsuccessful treatments.

Genetic testing plays an important role in fertility treatments.

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 appear 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.

No matter what type of family you want to form, Amplexa Genetics supports responsible parenthood

Choose between three different genetic tests:

	Genes**2**Life	Genes**4**Life	Genes**4**Life **Plus**
Test level	NGS basic	NGS extended, includes Genes2Life	NGS extended Plus, includes Genes4Life
Range	93 genes	430 genes	+ 1400 genes
Genes tested	Recessive genes as recommended by ACOG*	Most common recessive genes	All relevant recessive genes
Diseases investigated	+ 90 diseases	+ 400 diseases	+ 750 diseases

We need a blood or saliva sample to perform the test.

The test result is available within 25 working days.

Genetic tests 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)

Which diseases will be investigated?

Types of genetic diseases investigated with **Genes2Life**, **Genes4Life** and **Genes4Life Plus**

Genetic diseases	Statistics
Juvenile amyotrophic laleral sclerosis	The prevalence and incidence of JALS are not known. A small number of cases have been reported to date. The disorder har been described in various ethnic groups.
Bloom syndrome	Bloom syndrome (BSyn) overall prevalence is unknown. In the Ashkenazi Jewish population it is estimated at approximately 1/48,000 births.
Triple A syndrome	Prevalence is unknown but less than 100 cases have been published since the first description in 1978.
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 live 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. Reference: https://www.orpha.net/consor/cgi-bin/index.php?lng=EN

When to undergo a genetic test?

No matter what type of family you want to form, the best decision is responsible parenthood.

Get tested before:

- Trying to get pregnant with your partner.
- Starting an assisted fertilization treatment.
- Trying to get pregnant with a sperm donor.
- Trying to get pregnant with an egg donor.
- Selecting an egg or sperm donor.

How to do it

Sample collection of blood or saliva.

Gene analysis of the liquid.

Genetic report for each sample.

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