Cystinosis: Genetics and inheritance

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Introduction to genes and chromosomes

Your body is made up of millions of cells, most of which contain a complete set of genes.

Genes act like a set of instructions for our bodies to follow, controlling our growth and how our bodies work. They are also responsible for characteristics such as eye colour, blood type and height.

We inherit two copies of each gene, one copy from our mother and one copy from our father. We all have thousands of genes in total.

Our genes are located on chromosomes. We have 46 chromosomes (23 pairs). We inherit 23 chromosomes from each of our parents.

Sometimes there is a change in one of the genes which stops the body from working properly.

How is cystinosis inherited?

Cystinosis is inherited in an autosomal recessive pattern. This means that a person must inherit two changed copies of the same gene in order to have cystinosis. If a person inherits one changed gene and one normal gene, then that person will be a healthy carrier. If both parents are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child.

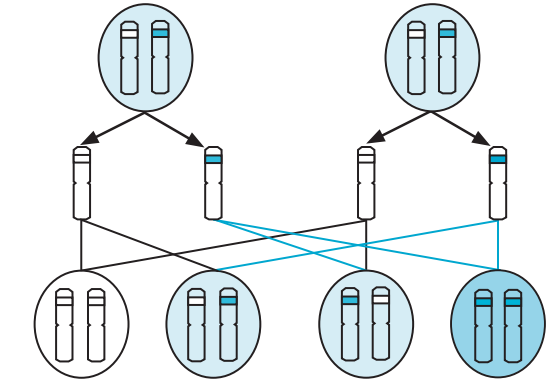
Each child of parents who both carry the cystinosis gene has a 25% chance of inheriting a changed gene from both parents and being affected by cystinosis.

There is a 50% chance that the child will inherit just one copy of the cystinosis gene. This means they will be healthy carriers like their parents.

There is a 25% chance that the child will inherit both normal copies of the gene and will not have cystinosis or be a carrier of cystinosis.

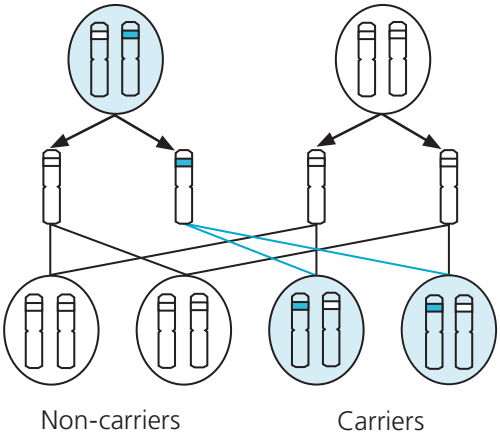
The chance remains the same in every pregnancy and is the same for boys and girls.

Autosomal recessive inheritance: both parents are carriers



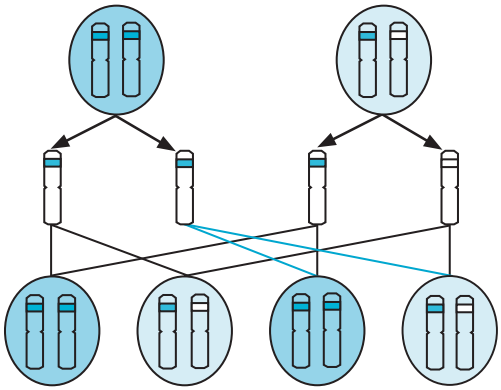
If only one parent is a carrier of the cystinosis gene, each child will have a 50% chance of being a carrier but will not be affected by the condition.

Autosomal recessive inheritance: one parent is a carrier



If one parent is affected and the other is a carrier, then there is a 50% chance the child will be affected by cystinosis. There is 50% chance the child will be a carrier of the cystinosis gene and be unaffected.

Autosomal recessive inheritance:
one parent affected one parent carrier



If one parent has cystinosis and the other is not a carrier there is 100% chance the child will be a carrier of the cystinosis gene but will not be affected by the condition.

Carrier testing in pregnancy

If both parents are known to be carriers of the cystinosis gene, and are considering having a baby, prenatal detection may be an option to see if the baby has inherited cystinosis.

Chronionic villus sampling involves taking a tiny sample of tissue from the developing placenta and is usually performed at 10-13 weeks.

Amniocentesis is when a sample of fluid is taken from the amniotic sac and can be performed after 15 weeks.

The discussion to have prenatal testing can be very difficult and you should only make a decision following consultation with your genetic councillor/genetic consultant and if you, and your partner, feel that it is important to know the information that the prenatal test will give.

www.geneticalliance.org.uk and **www.nhs.uk** has further information about prenatal genetic testing.

Contacts

Genetic Alliance UK
Telephone: 02077 043 141
Email: contactus@geneticalliance.org.uk
Website: www.geneticalliance.org.uk

A directory of regional genetic centres in the UK can be found at **www.geneticalliance.org.uk/services.htm**