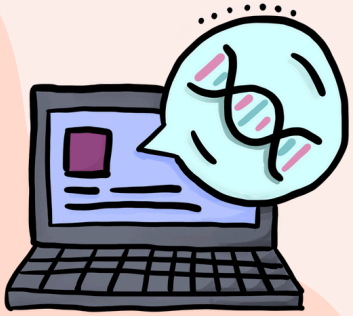
WHAT CAN BE DONE IF YOU HAVE AN AUTOSOMAL RECESSIVE CONDITION?

While many genetic conditions currently do not have a cure, genetic counselling is recommended, especially when planning for pregnancy. Genetic testing can provide valuable information about the risks and available options.

Depending on the genetic test results, the couples may consider:

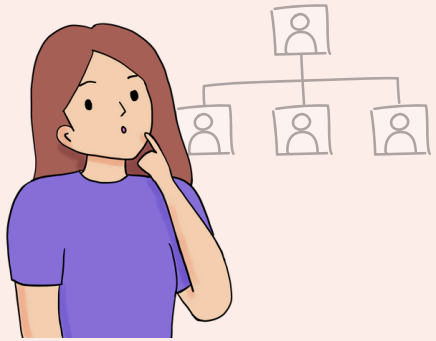
- Prenatal diagnosis by fetal genetic analysis through chorionic villus sampling or amniocentesis when the couples conceive again.

- In vitro fertilization with special genetic testing called preimplantation genetic diagnosis (PGD) to select unaffected embryos.



SHOULD I TELL MY FAMILY MEMBERS IF I HAVE AN INHERITED CONDITION?

If you have an inherited condition, it is important to inform your family members, as they may also have inherited the condition and could benefit from knowing about it. This is particularly crucial if they are planning to have children. It is recommended for them to seek advice from healthcare professionals to understand the implications and potential risks associated with the condition.



For further enquires, you may contact us.

Department of Clinical Genetics
Specialist Outpatient Clinic,
1/F, Tower B,
Hong Kong Children's Hospital,
1 Shing Cheong Road, Kowloon Bay,
Kowloon, Hong Kong

Office Tel: 5741 3186

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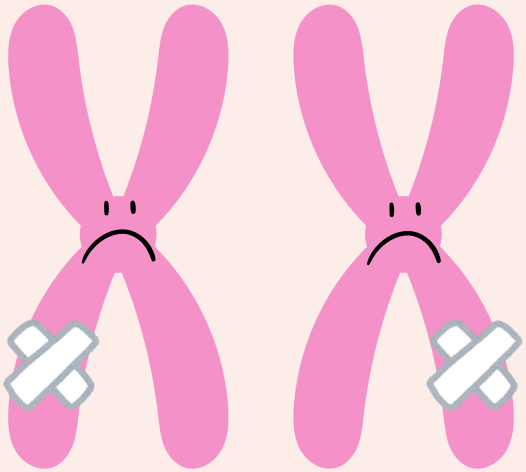
香港兒童醫院
Hong Kong Children's Hospital

GENETICS AND GENOMICS CLINIC
DEPARTMENT OF CLINICAL GENETICS



Autosomal Recessive Inheritance

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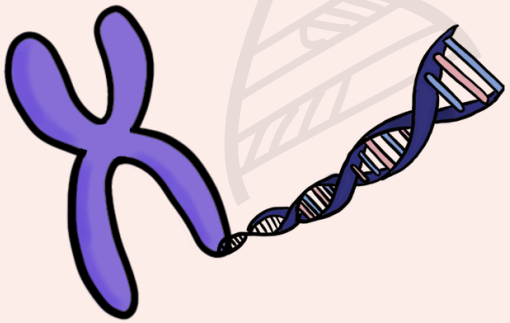
Autosomal Recessive Inheritance



WHAT ARE GENES AND CHROMOSOMES?

Genes act as instructions that guide the growth and functioning of the human body. They are located on structures known as chromosomes. In each cell, there are typically 46 chromosomes arranged in 23 pairs. The first 22 pairs, called autosomes, carry numerous genes responsible for determining human physical characteristics. Chromosomes are inherited from the father and mother, with one set received from each parent.

WHAT DOES RECESSIVE INHERITANCE MEAN?

Autosomal recessive inheritance is the way certain genetic conditions are passed down through the family in a recessive pattern.

In autosomal recessive inheritance, two copies of an altered gene are necessary for an individual to be affected. Typically, both parents are asymptomatic carriers of a single altered copy and one normal copy of the gene. Carriers of an altered recessive gene do not show any symptoms or signs of the condition because the normal copy of the gene compensates for the altered copy. However, they can pass the altered gene to their children without being affected by the condition themselves.

The occurrence of autosomal recessive conditions can be influenced by ethnic background and consanguinity (marriages between close relatives), as individuals who share a common ancestry are more likely to carry the same altered recessive gene. However, it can occasionally occur in non-consanguineous couples without any family history.

HAVING CHILDREN

If both parents are carriers of the same altered gene, each child (regardless of gender) has:

- 25% (1 out of 4) chance of inheriting the normal gene from both parents. The child would not be affected.
- 50% (1 out of 2) chance of inheriting an altered gene from one parent, and a normal gene from the other parent. The child would be a healthy carrier, just like the parents.
- 25% (1 out of 4) chance of inheriting an altered gene from both parents. The child would be affected by the condition.