

Autosomal recessive inheritance and carrier testing

Background information

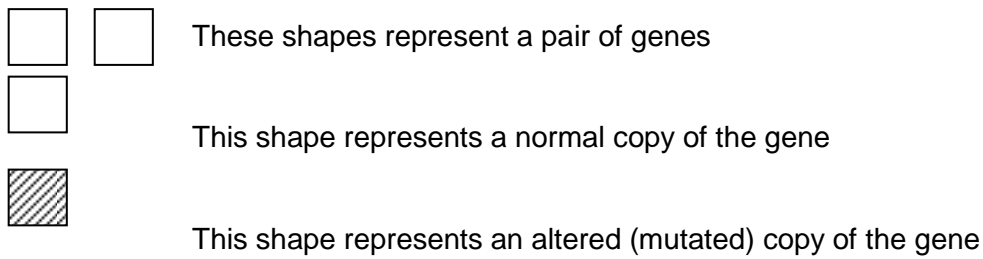
Genes determine how our bodies develop and function. We have about 20,000 pairs of genes in total. We have two copies of every gene, one inherited from each parent. We all have several genes where one copy is altered i.e. there is a gene mutation). This usually has no effect as the other copy of the gene is normal. Some genes tend to have mutations in the same place, but most genes can have mutations anywhere, making testing of the gene more difficult.

What is autosomal recessive inheritance?

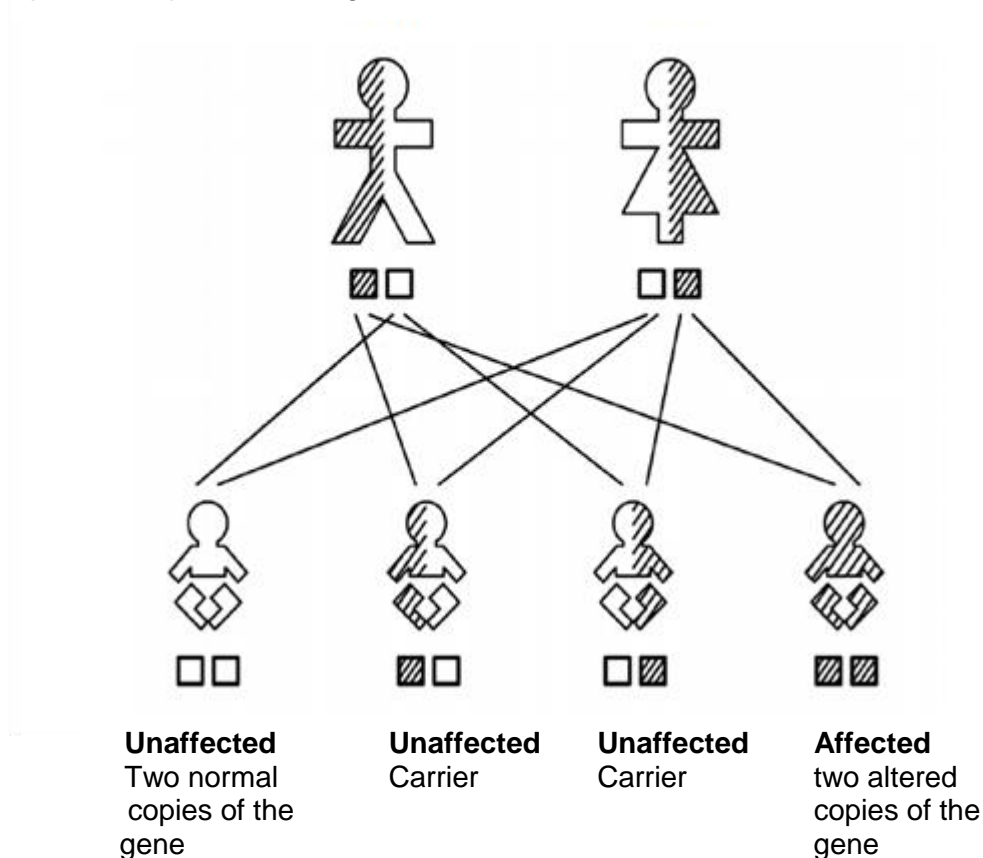
In autosomal recessive inheritance, both parents have a mutation in one copy of the same pair of genes and a child inherits an altered copy of the gene from both parents.

What is a carrier?

A carrier is a person with one normal and one altered copy of a gene. Being a carrier of an autosomal recessive condition does not cause medical problems.



Both parents are carriers of the same genetic condition. There are four possible ways for the parents to pass on their genes.



When should carrier testing be offered?

An individual with a family history of a known recessive disorder will have an increased chance of being a carrier for that disorder, compared to the general population. Accurate carrier testing may be possible for that individual; though will only be possible if the family mutation is known. However, an accurate test for their partner will only be possible in certain circumstances, usually if there is a common mutation in that particular gene or the couple are related to each other

In many situations, accurate carrier testing of a partner is not possible. In these situations there is little benefit in testing one of the couple since risks of having an affected child would be low and testing in pregnancy would not be possible.

Referrals should be made for:

- SMA (spinal muscular atrophy), CF (cystic fibrosis), CAH (congenital adrenal hyperplasia) and Friedrich's Ataxia as the interpretation of the results in these conditions can be difficult.
- Consanguineous couples with a family history of a recessive condition

Referrals for Tay Sachs screening should be sent to:

Tay Sachs Clinic
South Thames Regional Genetics Service (East)
Clinical Genetics
7th Floor, Borough Wing
Guys Hospital
London, SE1 9RT

We are happy to discuss any cases if you are unsure about whether a referral would benefit a couple. Contact details are:

The Clinical Genetics Unit
Great Ormond Street Hospital
Great Ormond Street
London WC1N 3JH

Tel: 020 7405 9200 Ext 6856
Fax: 020 7813 8141
Email: genetic.correspondence@gosh.nhs.uk

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