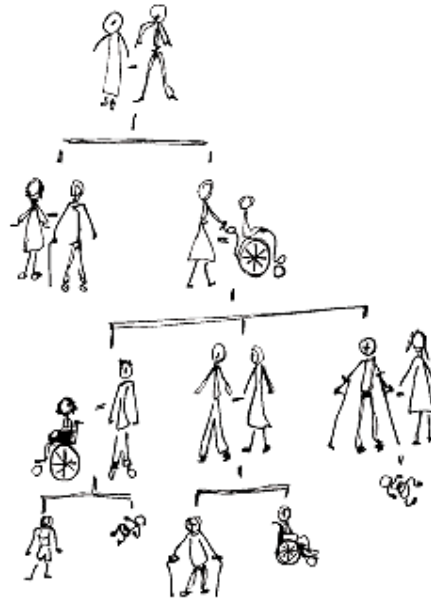


CROSS-CURRICULAR MATERIALS FOR KEY STAGE 4
(SCOTTISH CERTIFICATE OF EDUCATION - STANDARD GRADE)



Genes and You

Teaching about genetics from a human perspective

by Gill Mullinar

GENETIC INTEREST GROUP

*

THE WELCOME TRUST

Helping medical science to flourish

*

Published by the Wellcome Trust

The Genetic Interest Group is a registered charity, no.803424

The Wellcome Trust is a registered charity, no. 2107S3

GENETIC CHROMOSOMAL CONDITION CARDS

GENETIC CHROMOSOMAL CONDITION CARDS ON THE FOLLOWING:



HAEMOPHILIA

Haemophilia

WHAT IS HAEMOPHILIA?

Haemophilia is an inherited and lifelong blood disorder where an essential blood clotting factor is either partly or completely missing. There are two types of Haemophilia, Haemophilia A in which factor VIII is affected and the rarer Haemophilia B, also known as Christmas Disease in which factor IX is affected. The symptoms for both are very similar. It occurs in all racial groups and affects about 1 in 10,000 of the male population. A woman may have severe haemophilia if her father has the condition and is married to a carrier.

HOW IS HAEMOPHILIA PASSED ON? The gene for Haemophilia is carried on the X sex chromosome (females are XX; males are XY). A man with Haemophilia passes the faulty gene to his daughters who then carry this gene and may pass the condition onto their sons. A man with Haemophilia does not pass the faulty gene onto his sons because they receive a copy of his Y chromosome (their X chromosome comes from the mother).

Men are affected because the faulty gene on their X chromosome lacks the necessary instructions to produce the clotting factor which causes blood to clot. Women are usually unaffected because they have two X chromosomes and the working copy of the gene tends to override the faulty copy. However, about a third of female carriers have low clotting factor levels and may be mildly affected.

If a carrier mother decides to have children, all her daughters have a 50% (1 in 2) chance of being a carrier. This depends on whether they receive the X chromosome containing the faulty or the working gene. Sons also have a 50% (1 in 2) chance of inheriting the faulty or the working gene. If they inherit the faulty gene, they will have Haemophilia as they do not have a second X chromosome to override the faulty copy.

About 30% of people with the condition have no family history of Haemophilia. They are probably affected because of a spontaneous genetic mutation which took place in the egg or sperm before fertilisation.

A PERSON WITH HAEMOPHILIA MAY BE AFFECTED IN SOME, OR ALL, OF THE FOLLOWING WAYS:

- As their blood clotting mechanism does not work properly, people with Haemophilia tend to bleed for longer than normal. Cuts and grazes are no great problem as a little pressure and plaster are usually enough to stop the bleeding. The main problem is internal bleeding into joints, muscles and soft tissues.
- People with Haemophilia can be mildly, moderately or severely affected, and will stay that way throughout their lives. A mildly affected person might only have problems after a severe injury or operation, while a moderately affected person might have problems as a result of a knock or deep cut. But no injury is necessary for a severely affected person to have regular internal bleeding into joints, muscles and tissues.
- Untreated internal bleeding can cause pain and can eventually lead to severe damage to joints and to restricted mobility. Treatment for severely affected people is usually by injecting the missing clotting factor. Injections can be given either when a bleed occurs, or regularly (perhaps two or three times a week) as a preventative measure. Most people learn to do their own injections at home and are encouraged to do so by their treatment centres.

OTHER INFORMATION

Children tend to have more bleeds than adults because they are more active and less aware of their condition. Until 1986, some people with Haemophilia were treated with contaminated blood products. This led to about 1,200 people with Haemophilia being infected with HIV and over 3,000 with the Hepatitis C virus. No new infections have occurred since 1986 when heat treatment of blood products reduced the risk of viral contamination, but tragically more than half of the people who were infected with HIV through treatment for Haemophilia have died of AIDS. A synthetically produced recombinant clotting factor is now widely used and avoids the risk of contamination from viruses found in human blood.

If you are interested in finding out more about Haemophilia, you can write (enclosing an A5 stamped addressed envelope) to: The Haemophilia Society, First Floor, Petersham House, 57a Hatton Garden, London, EC1N 8JG.

Genes And You

Teaching about genetics from a human perspective

