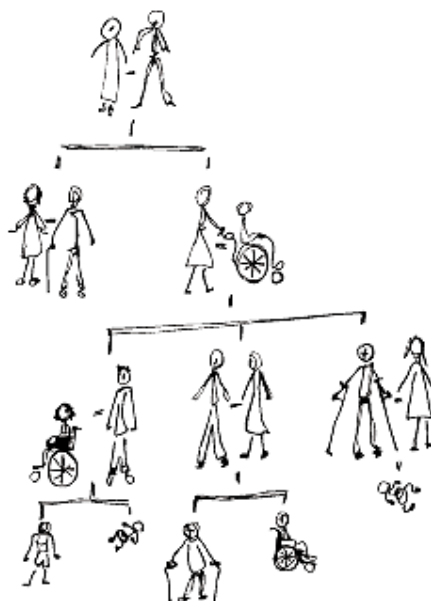


**CROSS-CURRICULAR MATERIALS FOR KEY STAGE 4  
(SCOTTISH CERTIFICATE OF EDUCATION - STANDARD GRADE)**



## **Genes and You**

**Teaching about genetics from a human perspective**

by Gill Mullinar

**GENETIC INTEREST GROUP**

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**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:



**EPIDERMOLYSIS BULLOSA (EB)**

## Epidermolysis Bullosa (EB)

### WHAT IS EB?

EB is a rare genetic skin condition which causes the skin and mucous linings within the body to blister very easily. The skin is made up of several layers held together by fibres. In a person with EB, these fibres do not work properly, so the slightest knock or friction can cause a gap to form between the layers which then fills with fluid and a blister is formed.

### EPIDERMOLYSIS BULLOSA (EB) HOW IS EB PASSED ON?

The pattern of inheritance for some types of EB is autosomal dominant and in others autosomal recessive. In the case of autosomal dominant inheritance, each and every child of an affected parent has a 50% (1 in 2) chance of inheriting the faulty gene and having EB.

In the case of autosomal recessive inheritance, neither parent is affected, but they both carry one copy of the faulty gene for EB and one working copy. Each and every child of theirs has:

- a 25% (1 in 4) chance of inheriting both faulty genes and having EB;
- a 50% (1 in 2) chance of inheriting one faulty copy and one working copy of the gene and being an unaffected carrier like their parents;
- a 25% (1 in 4) chance of neither having EB nor being a carrier.

Some people will have no family history of the condition and are probably affected because of a spontaneous genetic mutation which took place in the egg or sperm before fertilisation.

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

In some types of EB, the blister forms below the surface of the skin where there are blood vessels and nerves, so the blisters are deep, blood-tinged and painful. Treatment is usually to open and flatten each blister. This leaves exposed wounds which may become infected or leave scar tissue when they heal.

Hands and feet are subject to constant stress and injury and may become malformed overtime. Repeated blisters and scarring can cause the fingers to join together, joints to contract and the nails to peel off. Eating and swallowing can be difficult for people who get blisters in the mucous membranes which line the mouth and the gullet (oesophagus). They may need to eat very soft food with a drink to help them swallow, or to have a near-liquid diet while the blisters are healing. Gentle stretching, arm and trunk exercises, disco dancing (with the body moving and the feet still) and swimming are all useful forms of physical activity for a person with EB. Rough play, games involving fast movement and activities like jumping, kneeling and handstands are likely to cause blisters and are best avoided. Children with EB are often small for their age and puberty can be delayed. Pain and itching can make sleeping difficult, and getting enough food can be a problem if the throat is blistered. As well as this, people with EB may be low in iron (anaemic) and be using energy to heal wounds and fight infection, so they can feel very tired and depressed at times.

### OTHER INFORMATION

There are several different forms of EB which range from mild to severe. In some, blistering is limited to the hands and/or feet, while in others the whole body can be affected.

The name 'epidermolysis bullosa' means a breakdown (lysis) in the skin (epidermis), which causes blistering (bullosa).

Treating blisters usually involves a morning and evening routine. The dressings used to cover the blisters are similar to those used for burns treatment (i.e. non-stick dressings held in place perhaps by a bandage, as sticking plaster or anything adhesive could cause severe damage to the skin when removed).

If you are interested in finding out more about EB, you can write to: DebRA, DebRA House, Wellington Business Park, Dukes Ride, Crowthorne, Berks. RG45 6LS. [www.debra.org.uk](http://www.debra.org.uk)