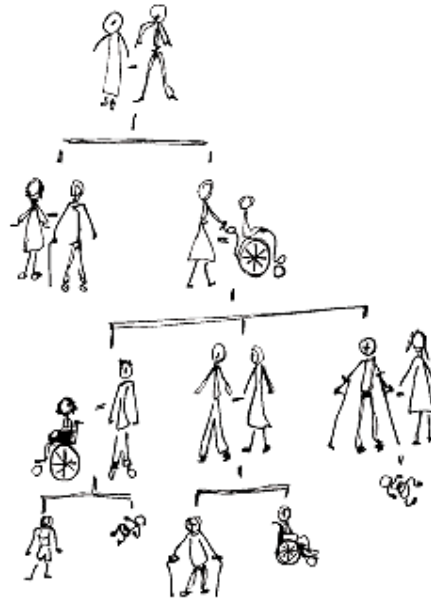


CROSS-CURRICULAR MATERIALS FOR KEY STAGE 4  
(SCOTTISH CERTIFICATE OF EDUCATION - STANDARD GRADE)



## 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:



**USHER SYNDROME**

## Usher syndrome

### WHAT IS USHER SYNDROME?

Usher syndrome is a genetic condition which causes a person to be born deaf and to lose their sight over a number of years. Loss of vision often begins in late childhood and is caused by an eye condition known as Retinitis Pigmentosa (pronounced rett-in-eye-tis pig-men-toe-ser) or RP.

### HOW IS USHER SYNDROME PASSED ON?

The pattern of inheritance for Usher syndrome is autosomal recessive. A person who inherits one faulty gene for Usher syndrome will be a carrier. Carriers are usually unaffected but can pass the faulty gene onto any children they may have. If one or both parents is a carrier, there is a 50% (1 in 2) chance that each child of theirs will also be a carrier. A child who inherits two copies of the faulty gene (one from each parent) will have Usher syndrome. If both parents are carriers, there is a 1 in 4 chance with each conception that a child will be born with Usher syndrome.

Three types of Usher have been described:

Type 1 is characterised by severe to profound deafness from birth, poor balance that can cause late walking and symptoms of RP before the age of 10.

Type 2 is characterised by mild to moderate hearing loss from birth, balance is normal and symptoms of RP may occur in the teens or twenties.

Type 3 the hearing may be normal or near normal in childhood and adolescence. Diagnosis of RP occurs usually around 30 to 40 years at the same time hearing starts to deteriorate. Vestibular (balance) problems also develop in midlife.

Currently, 11 genes have been located which cause the Usher syndromes and of these 7 have been identified and the protein involved described.

### USHER SYNDROME

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

- A person with Usher can be born deaf or partially hearing. As a child, he/she will probably learn to communicate through lip reading and sign language. Those who prefer to sign and become members of the Deaf community experience deaf culture.
- Diagnosis of RP often in their teens can be a terrible shock. It means that not just one, but two of the senses they use to communicate with others and help them learn about the outside world (hearing and vision) are now impaired. As their field of vision gets smaller, lip reading and sign language become more difficult and they can feel isolated not just from the hearing world but also from the deaf people who may be their closest friends.
- Balance is often affected in people with Usher syndrome. This can severely restrict their ability to move around freely and safely and make them appear clumsy or even drunk at times.
- As time goes on, and without special support, it becomes more difficult for a person with Usher to use public transport, hold down a job, and live independently.

### OTHER INFORMATION

Out of every 100 people who are born deaf, about 3-6% of them will have Usher syndrome.

**Retinitis Pigmentosa** or RP does not cause blindness straightaway but causes gradual loss of vision. In RP, the retina cannot take in low levels of light, so a person with Usher syndrome cannot see at night, in the dark or in poor lighting. Bright sunlight is a problem because the retina takes time to get used to changes in the level of light entering the eye. RP also leads to tunnel vision (when a person cannot see very well at the sides or up and down). This can mean that people with Usher syndrome trip over things, miss steps and stairs, bump into people and are unable to find things which they've dropped.

There is currently no treatment or cure for Usher syndrome, but training and support can help people with Usher to learn the skills they need to cope with life when their sight starts to fail.

If you are interested in finding out more about Usher syndrome, you can write (enclosing an A4 stamped addressed envelope) to: Usher Information, SENSE, 11-13 Clifton Terrace, Finsbury Park, London N4 3SR. Telephone: 020 7272 7774  
Text: 020 7272 9648 Website: [www.sense.org.uk/usher](http://www.sense.org.uk/usher)

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