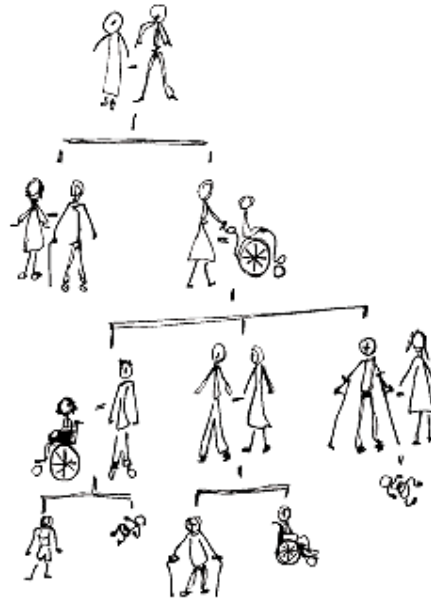


CROSS-CURRICULAR MATERIALS FOR KEY STAGE 4
(SCOTTISH CERTIFICATE OF EDUCATION - STANDARD GRADE)



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:



TAY SACHS DISEASE

Tay Sachs Disease

WHAT IS TAY SACHS DISEASE?

Tay Sachs Disease is rare, neurodegenerative metabolic disorder caused by the absence of an enzyme called hexosaminidase A (hex-A). Without this enzyme, a fatty substance called GM(2) ganglioside builds up in the body, particularly in brain cells, and destroys the central nervous system of affected children do not normally live beyond the age of five. There are different forms of Tay Sachs – infantile, juvenile, late onset and variant forms.

HOW IS TAY SACHS DISEASE PASSED ON?

The pattern of inheritance for Tay Sachs Disease is autosomal recessive.

A person who inherits one faulty gene for Tay Sachs 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 Tay Sachs Disease. If both parents are carriers, there is a 25% (1 in 4) chance of this happening.

TAY SACHS DISEASE

A CHILD WITH TAY SACHS DISEASE MAY BE AFFECTED IN SOME, OR ALL, OF THE FOLLOWING :

- A baby with Tay Sachs seems to develop normally until about six months old although infantile form will start to show during the first three to five months. Development may be seen to slow down, the baby begins to lose his/her sight, and becomes hypersensitive to sound so that any sudden noise or movement makes him/her jump.
- At about a year, the baby starts to have convulsions and fits and begins to lose the skills (s)he has developed (e.g. turning over, sitting, reaching out and crawling). Muscles become progressively weaker, swallowing becomes difficult and the lungs do not function well.
- Children with Tay Sachs Disease usually don't live past the age of five because their nervous system has been severely damaged. Symptoms of the disease will include blindness, deafness, loss of skills, paralysis and seizures.
- Treatment is symptomatic, anti-convulsants may be used for seizures. Treatment for individuals with this disease aims to provide relief for symptoms and support in the care of the individual. The symptoms, severity and rate of progression of this condition vary from one individual to another.

OTHER INFORMATION

- Tay Sachs Disease occurs most often in people of Central and Eastern European (Ashkenazi) Jewish descent. Approximately 1 in 25 Ashkenazi Jews are unaffected carriers, compared to 1 in 250 of the general population. But the disease also occurs amongst other groups, for example non-Jewish French Canadians.
- The disease is named after a British doctor called Warren Tay, who in 1881 described eye changes in a person with the disease, and an American neurologist called Bernard Sachs, who recognised that cell changes took place in affected children and also understood that the disease was inherited.
- If you are interested in finding out more about Tay Sachs Disease, you can write (enclosing an A5 stamped addressed envelope) to: Children Living with Metabolic Diseases (CLIMB) Climb Building, 176 Nantwich Road, Crewe, Cheshire CW2 6BG.
www.climb.org.uk

Genes And You

Teaching about genetics from a human perspective

