

GENETIC DISEASES

1. Check the pronunciation of the words below

cell	/sɛl/	dominant	/ˈdɒmənənt/
chromosome	/ˈkrɒsɒməˈsoʊm/	recessive	/rɪˈsɛsɪv/
characteristic	/ˈkærɪktəˈrɪstɪk/	genotype	/ˈdʒɛnəˌtaɪp, ˈdʒɪnə-/
genetic	/dʒəˈnɛtɪk/	phenotype	/ˈfiːnəˌtaɪp/
gene	/dʒiːn/	hemophilia (AE) haemophilia (BE)	/ˌhiːməˈfɪliə, -ˈfɪlyə, ˌhɛmə-/
allele	/əˈliːl/	sickle-cell anemia(AE) anaemia (BE)	/əˈniːmiə/
homozygote	/ˌhoʊməˈzaɪɡoʊt, - ˈziɡoʊt, ˌhɒmə-/	cystic fibrosis	/ˈsɪstɪk/ /faɪˈbroʊ sɪs/
homozygous	/ˌhoʊməˈzaɪɡəs, ˈhɒmə-/	carrier	/ˈkæriər/
heterozygote	/ˌhɛtərəˈzaɪɡoʊt, - ˈziɡoʊt	heterozygous	/ˌhɛtərəˈzaɪɡəs/

2. Complete the text

..... are different versions of the same gene. An can be dominant or recessive. Individuals, meanwhile, can be homozygous or heterozygous:

- individuals who are for a certain gene carry two copies of the same
- individuals who are for a certain gene carry two different

A recessive characteristic will only be shown if an individual is homozygous for the recessive A dominant characteristic will be shown even if an individual is heterozygous for the dominant

Adapted from: http://www.bbc.co.uk/schools/gcsebitesize/science/ocr_gateway/ourselves/7_who_am_i4.shtml
(Accessed: April 20, 2009)

3. Listen and decide whether the statements are true or false. (timing 28:30)
Tay Sachs Disease

- a. Among Orthodox Jews, among Ashkenazi Jews between 2% and 3% of the population carries an allele for Tay-Sachs disease which is phenotypically silent in the heterozygous state but in the homozygous state is a devastating condition which leads to death in the first ten years of life.

- b. So now among the Orthodox Jews in New York before two young people will get married they will do a test to see whether they are heterozygous for the Tay-Sachs allele. It is limited to Orthodox Jews.

- c. Among those who don't live in such a closely structured society such people face the possibility of one of their four offspring on average coming out as a homozygote and having an incurable genetic disease which is going to lead to their early death.

Adapted from:

http://ocw.mit.edu/NR/rdonlyres/Biology/7-012Fall-2004/B800FEAA-439B-4510-93DE-AFD866458EA/0/7_0122004L35.pdf
<http://ocw.mit.edu/OcwWeb/Biology/7-012Fall-2004/VideoLectures/detail/embed35.htm>

4. Listen and fill in the gaps

Sickle Cell Anemia

There are villages in Greece where there are a substantial percentage of people who carry the 1..... sickle cell anemia which, as you may know, is not so serious phenotypically in heterozygous form, but in the 2..... form is actually devastating. And the reason they have sickle cell anemia is that those areas of Greece historically had high 3..... of malaria. And, as you may know, sickle cell anemia actually protects, in the heterozygous state actually 4 one from the ravages of the malarial 5.....

So about 20 years ago it became possible to do a simple genetic test to 6..... whether an individual was 7..... for sickle cell anemia. And what happened is that somehow what was supposed to be confidential medical genetic tests got out. They became public. And young individuals in the population became known as 8....., as heterozygotes for sickle cell trait, even though 9..... they were reasonably normal because the heterozygote condition is not so devastating.

So those individuals were soon ostracized, to use an old Greek word. They were soon put to the side. They were placed in the pool of the unmarriageable because nobody wanted to marry them. And so they then, as a 10....., began to marry amongst themselves. Remember what I told you about 11..... with the sickle cell trait. But that's only one example of that.

Adapted from:

http://ocw.mit.edu/NR/rdonlyres/Biology/7-012Fall-2004/B800FEAA-439B-4510-93DE-AFD866458EA/0/7_0122004L35.pdf

Cystic Fibrosis

- Cystic fibrosis (CF) is one of the most common serious genetic (inherited) diseases.
- CF gene is carried by 1/20 persons (in Caucasian populations) and 1 in 400 couples is at risk for having children with CF.
- CF is characterized by the production of abnormal secretions leading to mucous build-up.
- CF mucous build-up can impair the pancreas and, secondarily, the intestine.
- CF mucous build-up in lungs can impair respiration.
- Early diagnosis of CF is of great importance.
- Early and continuing treatment of CF is valuable.

<http://www.medicinenet.com> (Accessed: April 13, 2009)

Both parents are heterozygous for the faulty allele

f is the cystic fibrosis allele

		mother	
		F	f
father	F	FF	Ff
	f	Ff	ff

Cystic fibrosis is an inherited disorder caused by a recessive allele. This genetic diagram shows the possible outcomes when both parents are heterozygous for the faulty allele. There is a one in four chance of the offspring being homozygous for the faulty allele, and so having cystic fibrosis.

One parent is heterozygous for the faulty allele

f is the cystic fibrosis allele

		mother	
		F	F
father	F	FF	FF
	f	Ff	Ff

Tay-Sachs disease /teɪ'sæks/

A genetic disease in which the products of fat metabolism accumulate in the nervous system, causing retardation, paralysis, and death by preschool age. Individuals of eastern European Jewish descent have a higher risk of inheriting Tay-Sachs disease. The disease is named after its describers, British ophthalmologist Warren Tay (1843-1927) and American neurologist Bernard Sachs (1858-1944).

<http://dictionary.reference.com/browse/tay-sachs+disease> (Accessed: April 13, 2009)

Sickle Cell Anemia

- Sickle cell anemia is an inherited disorder of the hemoglobin in blood.
- Sickle cell anemia requires the inheritance of two sickle cell genes.
- Sickle cell trait, which is the inheritance of one sickle gene, almost never causes problems.
- Virtually all of the major symptoms of sickle cell anemia are the direct result of the abnormally shaped sickled red blood cells blocking the flow of blood.
- The current treatment of sickle cell anemia is directed primarily toward managing the individual features of the illness as they occur.

<http://www.medicinenet.com> (Accessed: April 13, 2009)

Hemophilia B:

Hemophilia due to deficiency of coagulation factor IX in the blood which results in prolonged oozing after minor and major injuries, tooth extractions, or surgery. There is renewed bleeding after the initial bleeding has stopped.

The gene for hemophilia B gene is on the X chromosome, so males are affected and females carry the gene. About 10% of carrier females are at risk for bleeding. Each son of a female carrier stands a 50 percent chance of receiving the gene and having hemophilia B.

Treatment is by blood products that introduce clotting factor IX and replace lost blood. Hemophilia B is also called Christmas disease (so-named for the first patient studied in detail with the disease).

<http://www.medterms.com> (Accessed: April 13, 2009)

Huntington's disease (Autosomal dominant)

The disease runs strongly in families: it is inherited dominantly so offspring from an affected person are 50% at risk of suffering it.^[1] The exact way HD affects an individual varies, even between family members, but there is a characteristic progression.^[10] The earliest symptoms are a general lack of coordination and an unsteady gait. As the disease advances uncoordinated, jerky body movements become more apparent, as does a decline in mental abilities and behavioral and psychiatric problems.^[1] Physical abilities are gradually lost until full-time care becomes necessary and the decline in mental abilities generally results in dementia.^[1] Although the disorder itself is not fatal, complications reduce life expectancy to around twenty years after diagnosis.^[1]

The mechanism of the disease is not fully understood, but a number of factors have been identified.^[1] A mutation in the Huntington gene, causes the production of the mutant protein huntington, which in turn produces cell and macroscopical changes in the brain.^[11] There is no cure for HD, although there are treatments to relieve some of its symptoms.^[1]

http://en.wikipedia.org/wiki/Huntington%27s_disease (Accessed: April 18, 2009)

Huntington's disorder

One parent is heterozygous for the Huntington's allele, while one is homozygous

H is the Huntington's allele		mother	
		H	h
father	h	Hh	hh
	h	Hh	hh