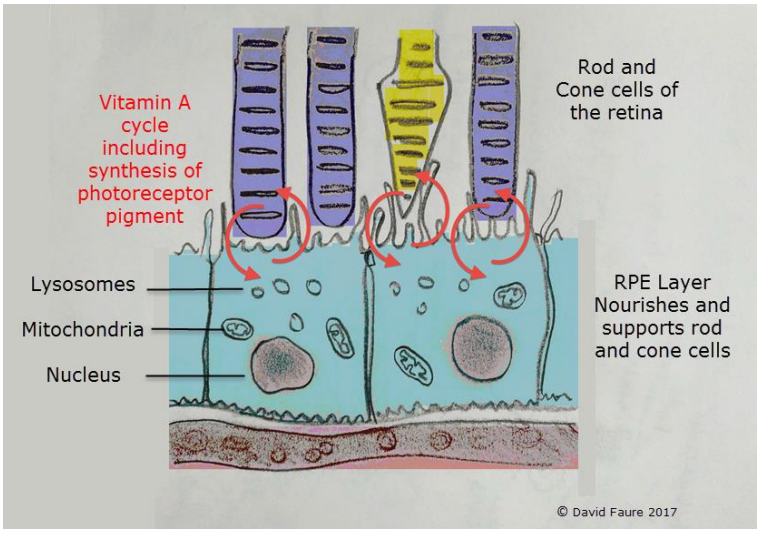
Stargardt disease IB style questions

Cells topic questions

The diagram below shows the relationship between light sensitive rod and cone cells and the RPE layer cells which support them.



1. State the roles of the mitochondria and the lysosomes in the RPE cells. (2)

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2. The Vitamin A cycle is part of the "metabolism" of the cells. Explain what is meant by the term "metabolism". (1)

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3. Describe how the shape of the PRE layer cells is adapted to their function (2)

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4. The exchange of materials between RPE cells and the rods and cone cells is very important. Suggest by which membrane transport methods ions, proteins and damaged fragments of rod cells would be taken into the RPE cells. (3)

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Stem cell questions

In a recent clinical trials, the London Project to Cure Blindness has used embryonic stem cells to produce an RPE cell line that can be cultured as a monolayer on a thin sheet of polymer (Carr et al., 2013).

1. Outline the properties of stem cells which will help to repair the RPE layer of a patients retina after implantatrion of the thin sheet of cells. (2)

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In a Japanese study (Hirami et al., 2009) the researchers have successfully reprogrammed adult cells taken from the patients' bone marrow. While the properties of these cells may not so closely match the RPE cells, there are some advantages in terms of tissue rejection and ethics.

2. Explain, in terms of ethics, why some people would prefer to use cells taken from adult bone marrow rather than embryonic stem cells.

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Protein synthesis questions

Mutations in a gene called ABCA4 are the most common cause of Stargardt disease. These mutations create recessive alleles of the ABCA4 gene. Some of the mutations are base substitutions or nonsense mutations resulting in a failure to produce the ABCA4 protein.

The ABCA4 protein, has 2273 amino-acids and its function in the retina is the removal of a lipid by-product from the manufacture of photosynthetic pigment from vitamin A.

Cells that lack the ABCA4 protein accumulate clumps of lipofuscin. Eventually, these fatty deposits lead to the death of photoreceptors and central vision becomes damaged.

Autosomal recessive mutations in the ABCA4 gene account for about 95 percent of Stargardt disease.

3. Suggest how many DNA base pairs would be found in the ABCA4 gene .(1)

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4. Outline how the presence of a base substitution mutation in the ABCA4 gene would result in the synthesis of a protein which cannot carry out its normal function. (4)

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Genetics topic questions

Stargardt disease is an inherited disorder of the retina – the layer of the eye that senses light. The dominant allele results in the production of the ABCA4 protein. Mutations produce recessive alleles which don't synthesise the ABCA4 proteins.

5. Suggest symbols for each of the alleles, dominant and recessive (1)

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6. Two parents are thinking of having children, but they think that they are both carriers of a recessive allele for Stargardt disease. The gene is known to be found on chromosome 1. Using a Punnett square, explain what the probability is of the children having Stargardt disease. (3)

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