

Self-Assessment

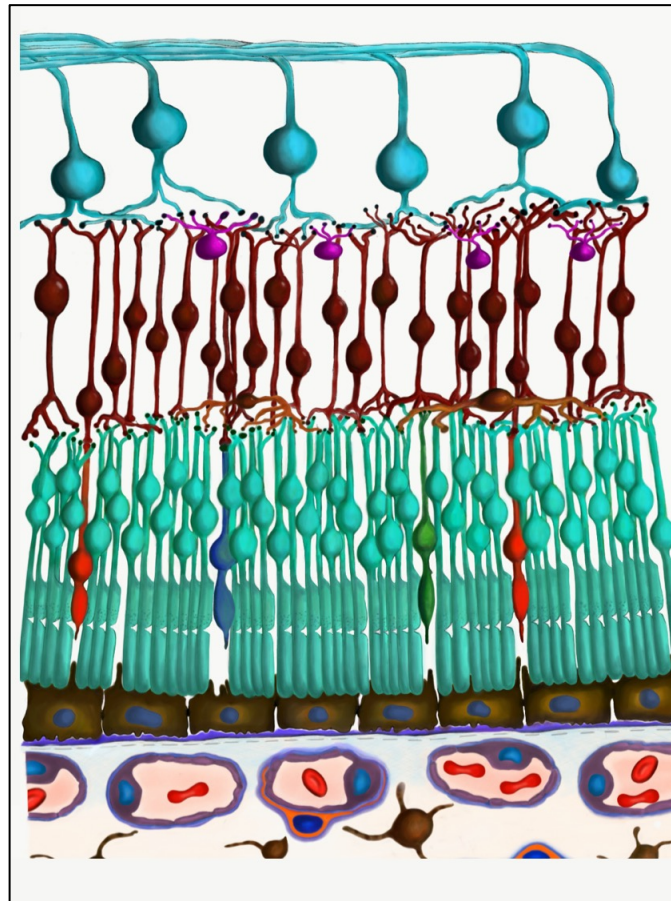
The Big 14 Inherited Retinal Diseases

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- 1) The relative frequencies of the Big 14 inherited retinal diseases that are summarized in this tutorial were determined by studying _____ families that were ascertained consecutively in a single clinic in the United States. These families traveled to the clinic from _____ different US states.
- 2) Nonsyndromic retinitis pigmentosa, is responsible for _____ percent of all inherited retinal disease and the second most common condition, autosomal recessive Stargardt disease represents another _____ percent.
- 3) Draw the relationships among the Big 14 here.

4) List five features of Mendelian retinal disease:

5) On the figure below, draw boxes around the anatomic zones that contain the initial pathologic changes associated with the following five categories of retinal disease: photoreceptor diseases, macular dystrophies, choroidopathies, retinoschisis and optic neuropathies. Label these five boxes on the right of the figure. On the left of the figure, label at least 10 anatomical features that are visible on a high quality OCT of a normal individual.



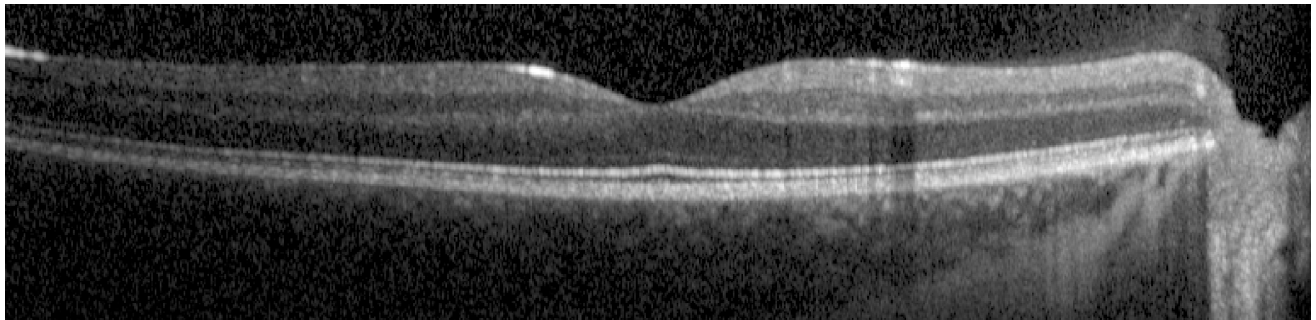
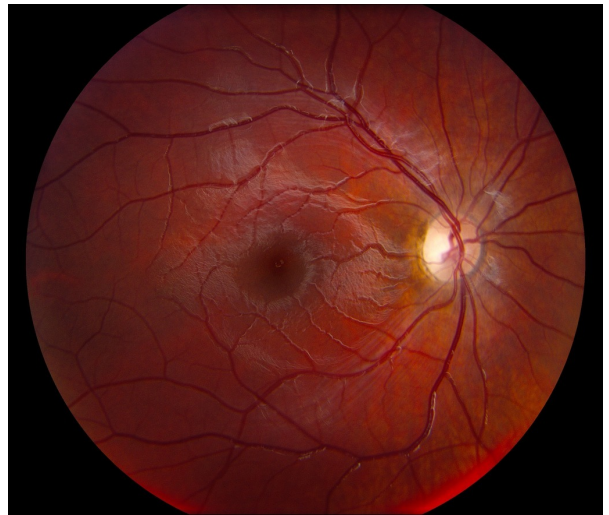
6) The function of the *ABCA4* protein in photoreceptors is to flip the visual cycle intermediate _____ from the inner to the outer leaflet of the disk membrane. Failure to do this at the normal rate results in the irreversible formation of a toxic molecule known as _____.

Case 1

This is an 11 year old male with 20/100 vision. He was first noted to have reduced acuity on a school vision screen at age 7.

Diagnosis? _____

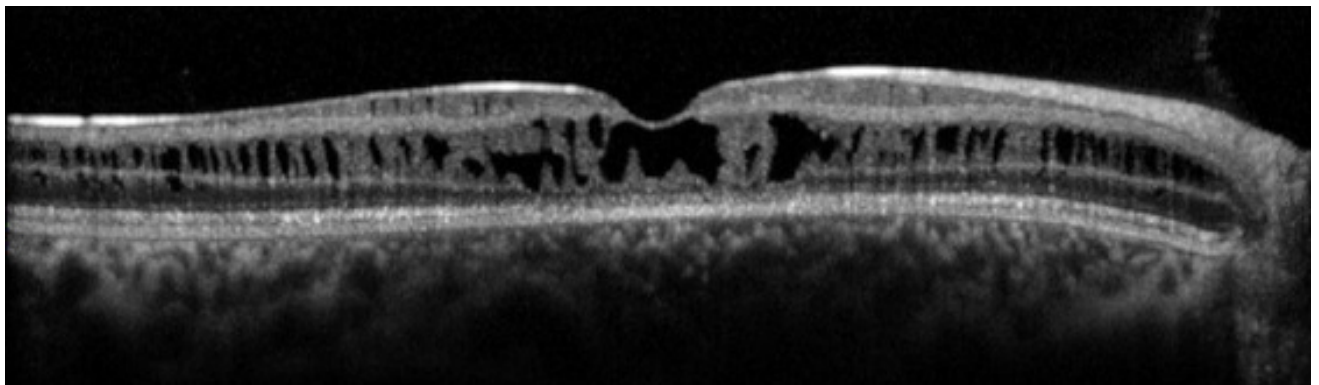
Most likely gene? _____

**Case 2**

This is the right eye of an 11 year old male with 20/60 vision. His parents have normal vision but he has a brother who also has reduced acuity.

Diagnosis? _____

Most likely gene? _____

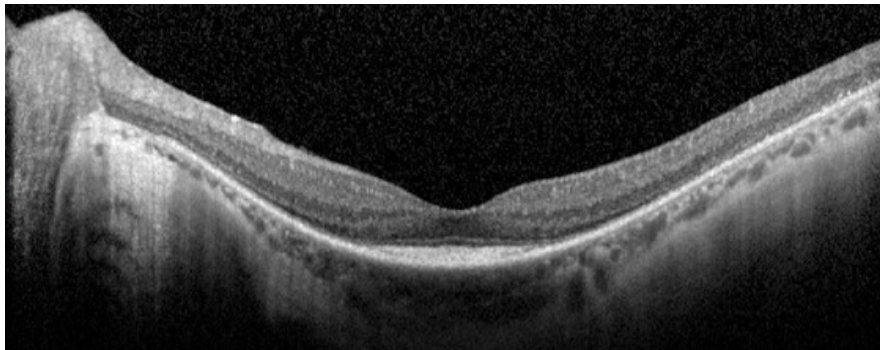
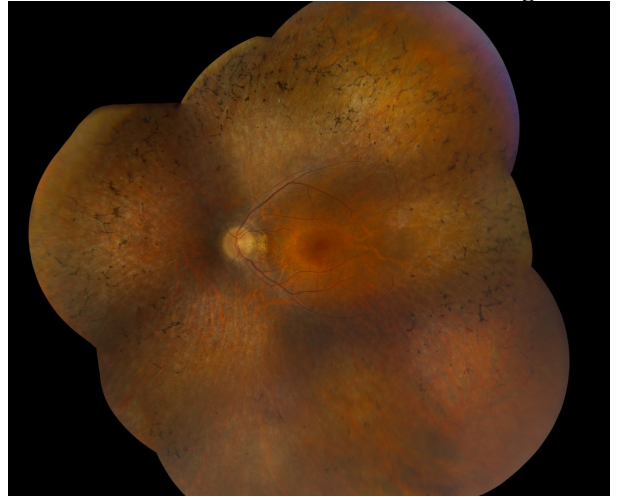


Case 3

This is a 49 year old woman with 20/20 acuity. In addition to her eye findings, her speech is abnormal and she has worn hearing aids in both ears since her hearing loss was first detected in early childhood.

Diagnosis? _____

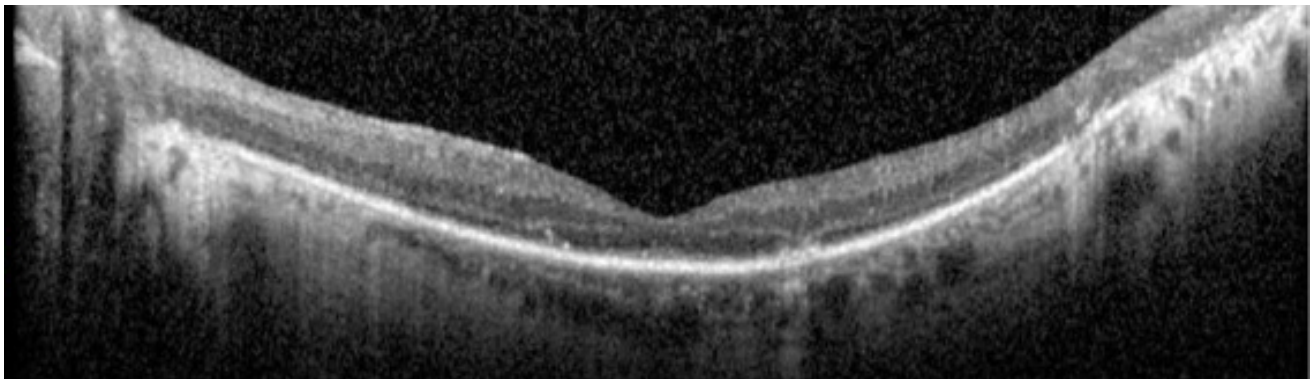
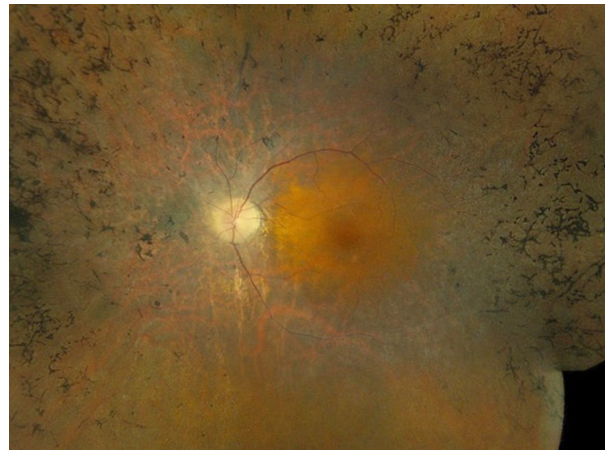
Most likely gene? _____

**Case 4**

Here is a color photograph and OCT of the left eye of a 65 year old man with 20/20 acuity. He has poor night vision and quite constricted peripheral vision. His parents had normal vision.

Diagnosis? _____

Most likely gene? _____

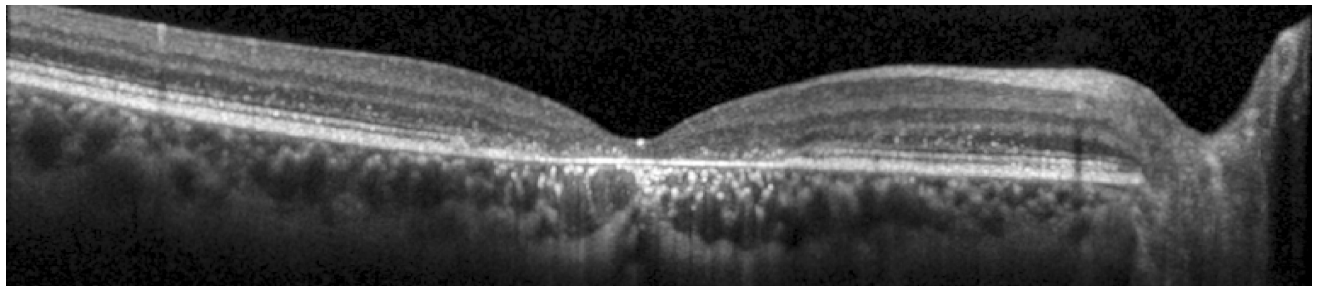


Case 5

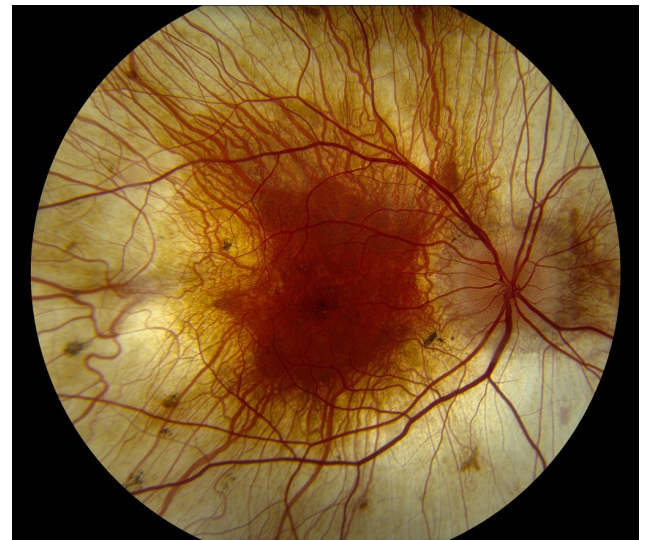
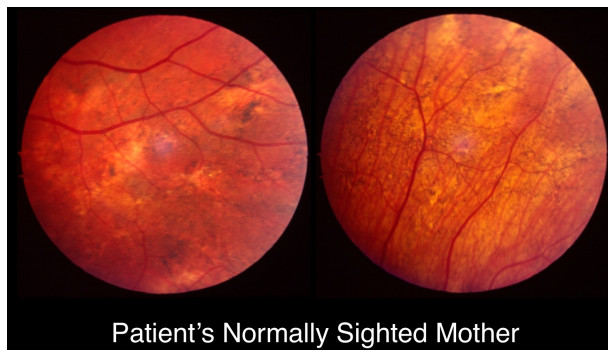
This is a 19 year old woman with 20/160 acuity. She had normal vision at age 5. Both of her parents have normal acuity.

Diagnosis? _____

Most likely gene? _____

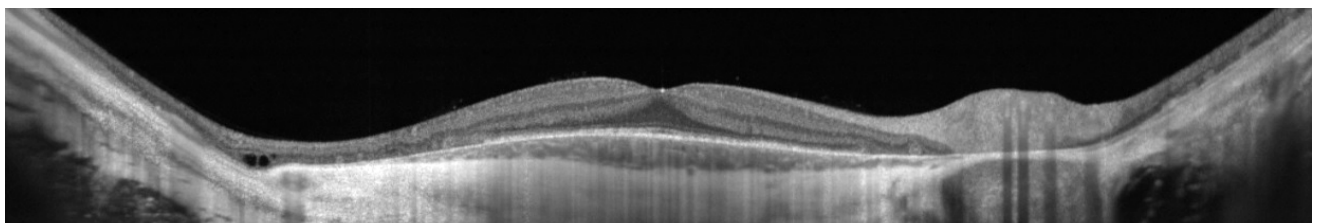
**Case 6**

This is a 19 year old man with 20/20 acuity. His mother and maternal grandmother have completely normal vision but both of them have an unusual appearance to their retinas.



Diagnosis? _____

Most likely gene? _____

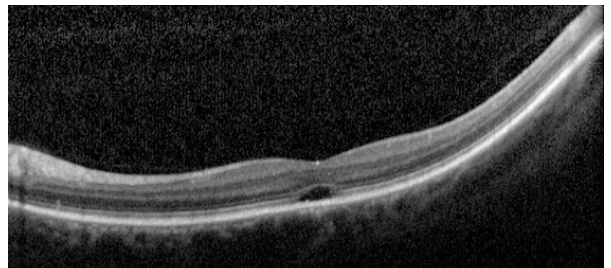


Case 7

This is a 47 year old woman who has 20/125 acuity. She has never had acuity better than 20/100. Her normally sighted parents noticed that she did not look at their faces normally or follow toys when she was just 12 weeks of age. Later she had an electroretinogram that revealed normal scotopic responses but extinguished photopic responses. Even as a child, she could not recognize any of the Ishihara plates or name the colors of common objects. She has been photophobic her whole life but could see as well as her friends in very dim environments like playing flashlight tag.

Diagnosis? _____

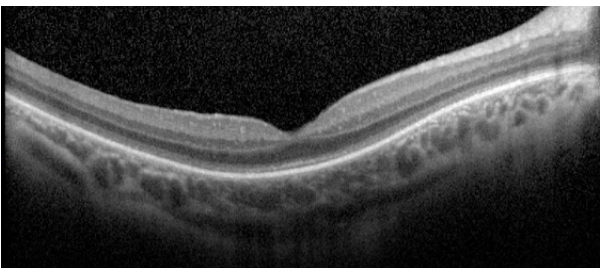
Most likely genes? _____



Case 8

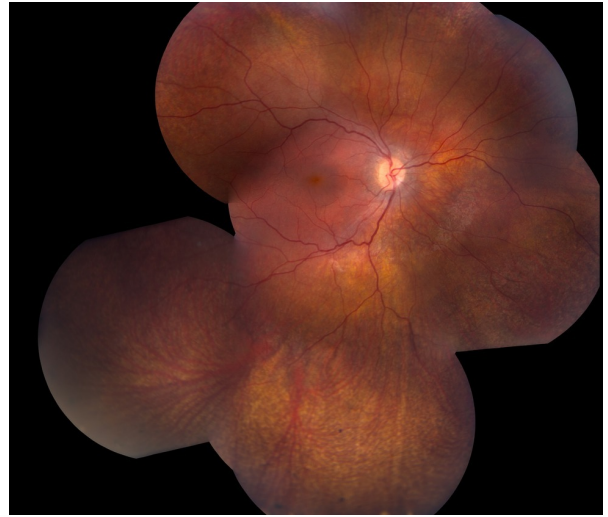
This patient is 5 years old and has 20/40 visual acuity. His normally sighted parents noticed that he inexplicably bumped into things shortly after he began to walk. They sought medical attention and he was given hyperopic spectacles of about 4 diopters which seemed to help with his central vision. However, his parents still suspected that his peripheral field was constricted because he would back up to find things. Electroretinography was then performed which revealed reduced scotopic responses and completely extinguished photopic responses. Goldmann perimetry revealed a V4e isopter about 50 degrees in diameter.

Diagnosis? _____

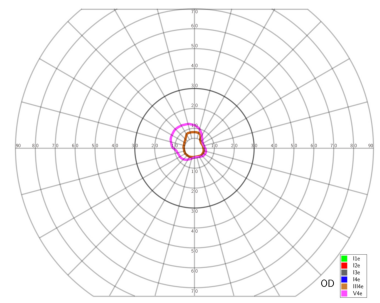
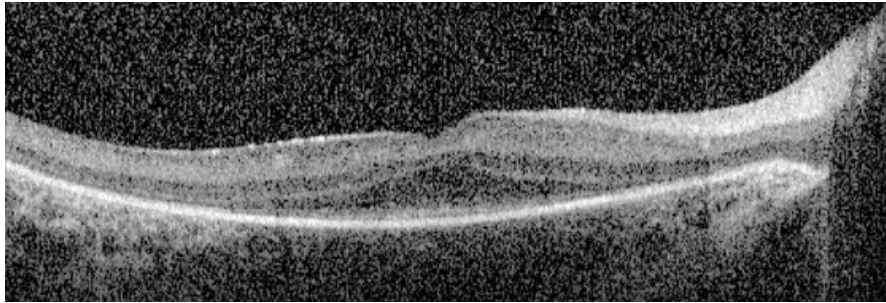


Case 9

This six year old girl was visually inattentive and had pronounced nystagmus at 4 months of age. An electroretinogram was performed at that time and no responses could be detected under any stimulus conditions. Her parents have normal vision. On our examination, her visual acuity was 20/80. Streak retinoscopy revealed about 5 diopters of hyperopia.



Diagnosis? _____ Most likely gene? _____

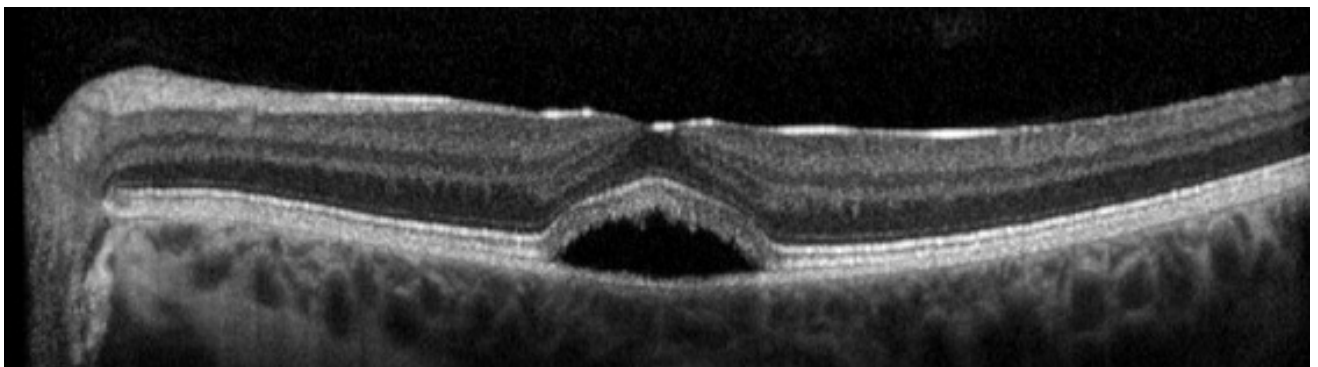
**Case 10**

This is a 17 year old male who has normal night vision, normal peripheral vision and 20/20 central acuity. He has a family history of adult onset loss of visual acuity in his mother and a number of his maternal relatives. Below is his optical coherence tomogram.



Diagnosis? _____

Most likely gene? _____

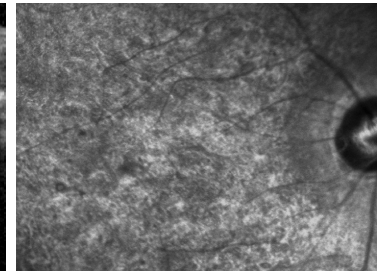
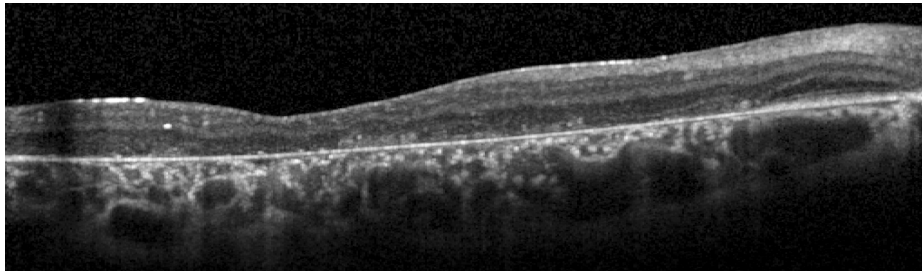
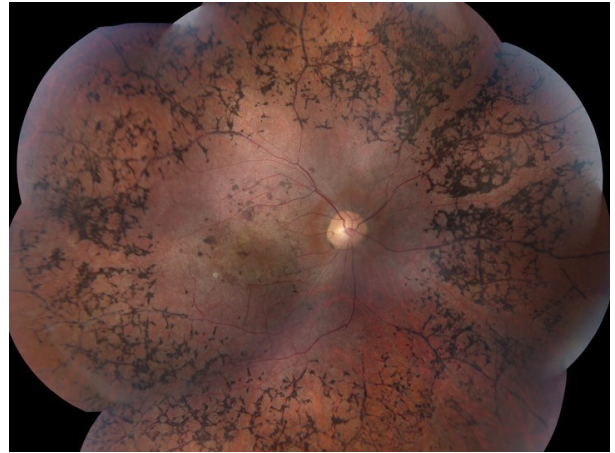


Case 11

This is a 17 year old woman with 20/200 acuity. She had completely normal vision until about age 9. Her parents have normal acuity.

Diagnosis? _____

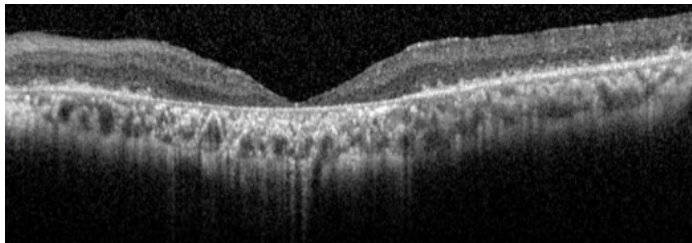
Most likely gene? _____

**Case 12**

This 47 year old woman has 20/500 visual acuity. Her parents have normal vision. She was born with an extra digit on the postaxial side of both hands and both feet and small scars can still be seen where these digits were removed. Her cognitive ability is a bit below normal and her body mass index is above normal. She has type 2 diabetes mellitus.

Diagnosis? _____

Most likely gene? _____

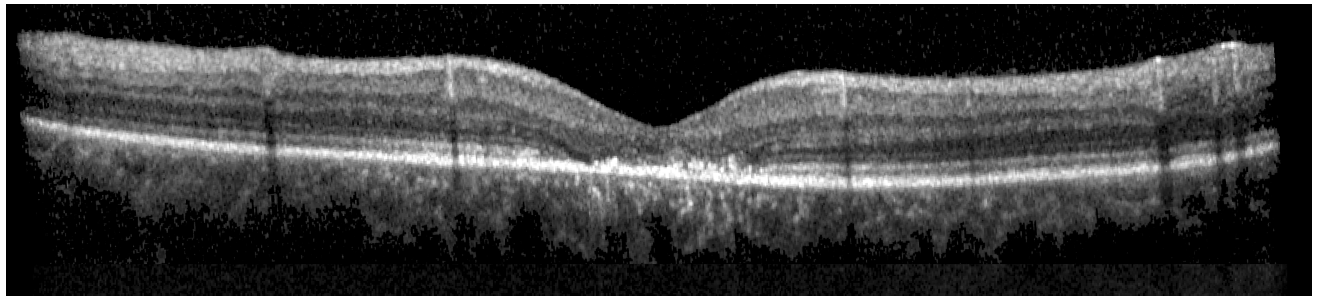
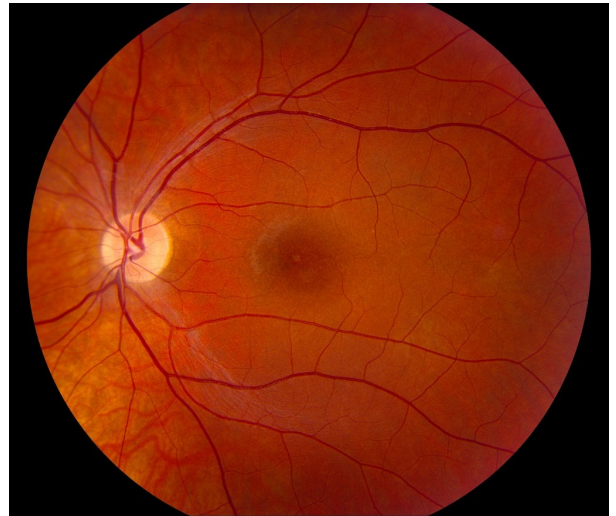


Case 13

This is a 32 year old man with 20/150 visual acuity. He had relatively normal acuity as a young child but gradually lost vision after age 10. He is photophobic and cannot recognize any of the Ishihara pseudo-isochromatic plates. Two of his siblings, his mother and his maternal grandfather are all similarly affected.

Diagnosis? _____

Most likely gene? _____

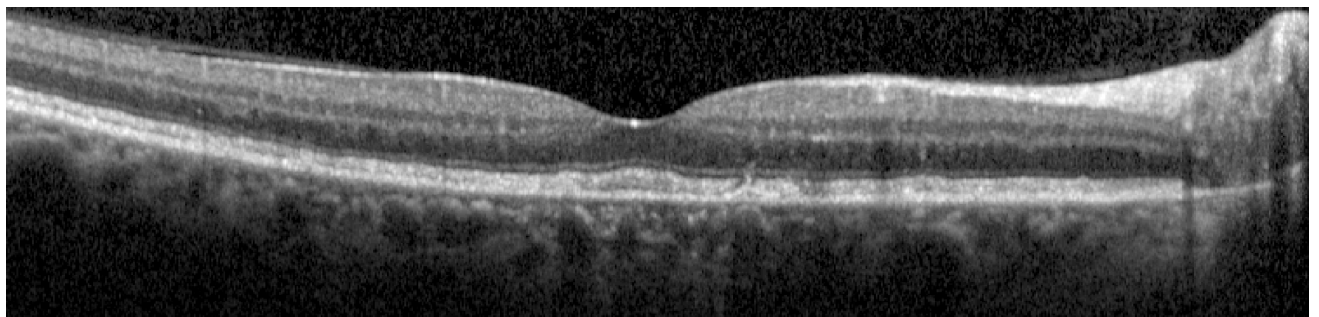
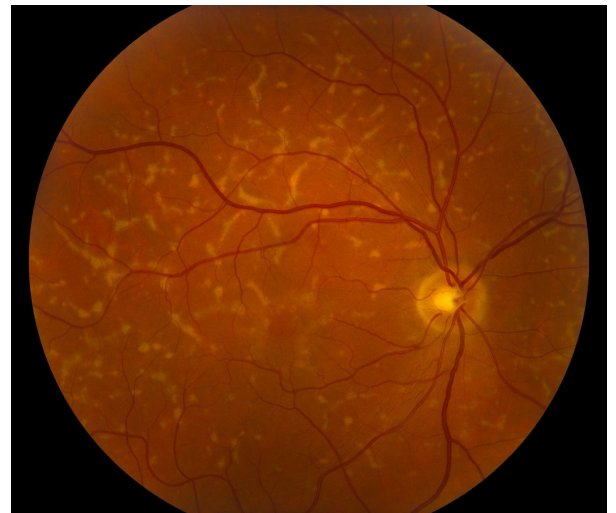
**Case 14**

This is a fundus photograph of the right eye of a 63 year old woman with 20/25 vision. She has many similarly affected relatives including one of her parents. Abnormalities were first observed in her retina in her 20's.

Below is her optical coherence tomogram.

Diagnosis? _____

Most likely gene? _____

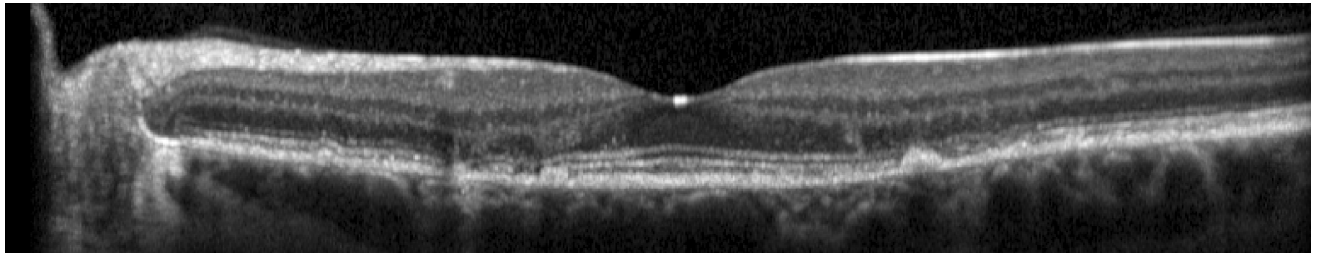


Case 15

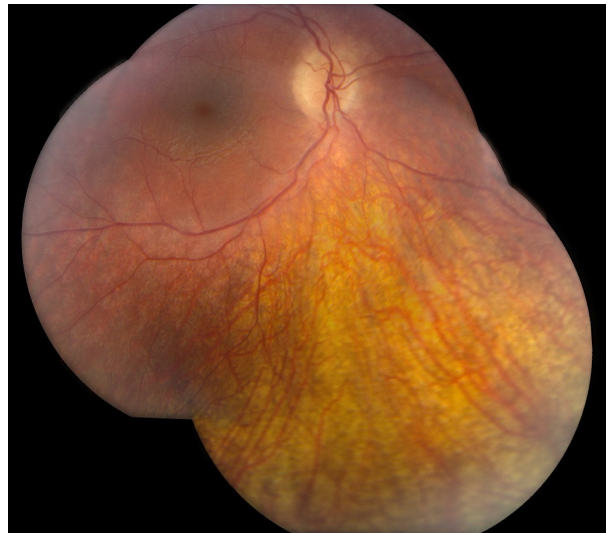
This is a 38 year old woman with 20/20 vision. Both of her parents have good vision. No one in her quite extensive family has any history of retinal disease.

Diagnosis? _____

Most likely gene? _____

**Case 16**

This 15 year old male was noted to have abnormal vision at 3 months of age. He had large amplitude nystagmus, did not respond to his parents faces and would not follow any type of toy or light. His pupils constricted when the room lights were turned off and dilated when turned on. Retinoscopy revealed over seven diopters of hyperopia and electro-retinography detected no responses under any stimulus conditions. The best acuity ever recorded was bare light perception. His parents have normal vision.



Diagnosis? _____ Most likely gene? _____

