Stargardt disease

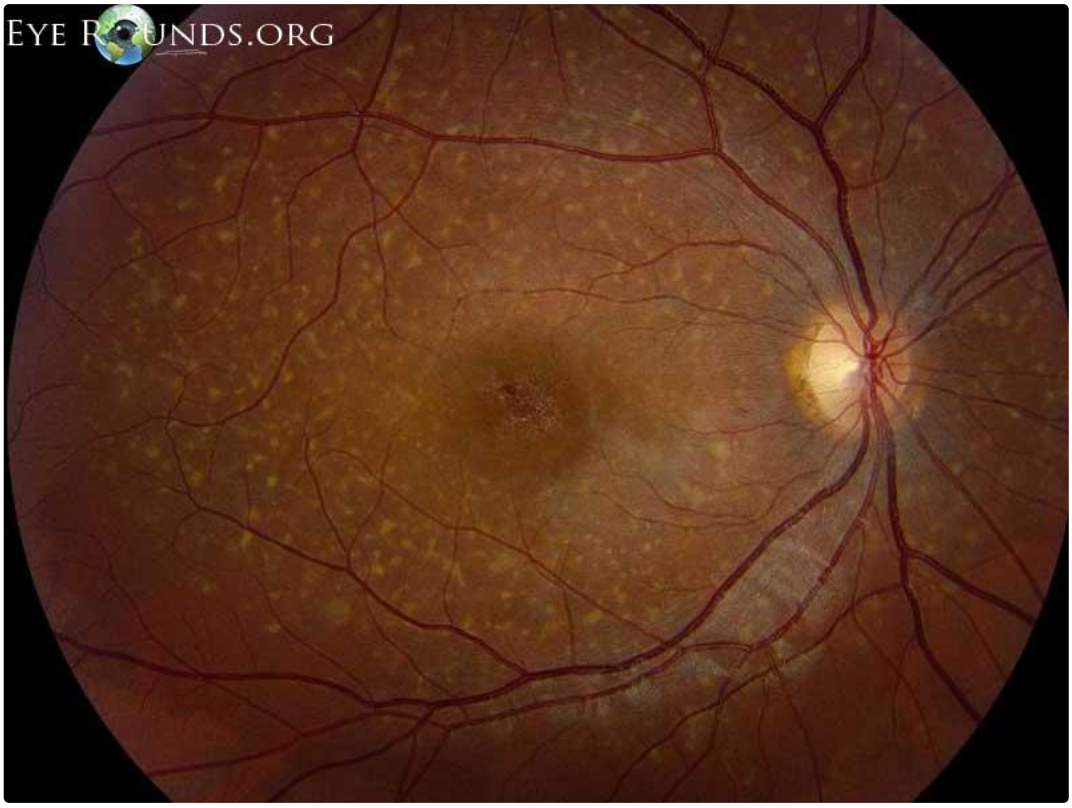
Category(ies): Genetics, Inherited Eye Disease, Retina

Contributor: Jeffrey D. Welder, MD

Photographer: D. Brice Critser, CRAt

20-year-old male with molecularly confirmed Stargardt disease (ABCA4 mutation). Note the diffuse yellow pisciform flecks within the arcades and central macular mottling in both eyes.

OMIM #248200, #600110, #603786



Right Eye

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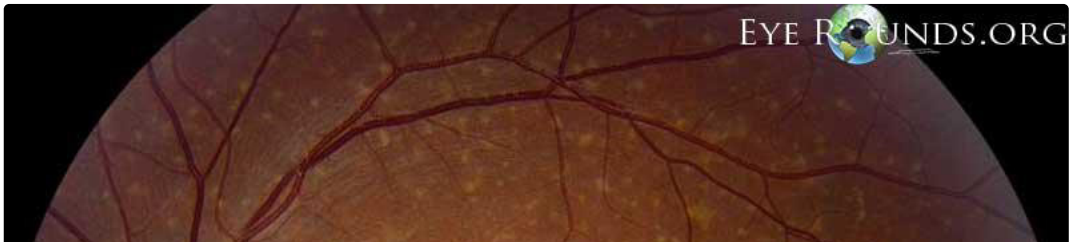

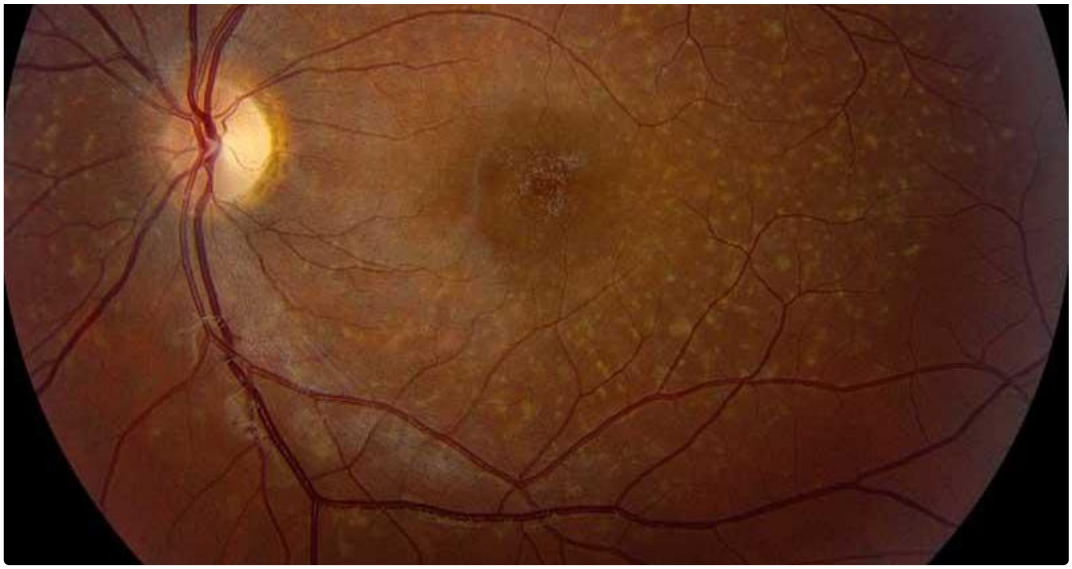

Left Eye

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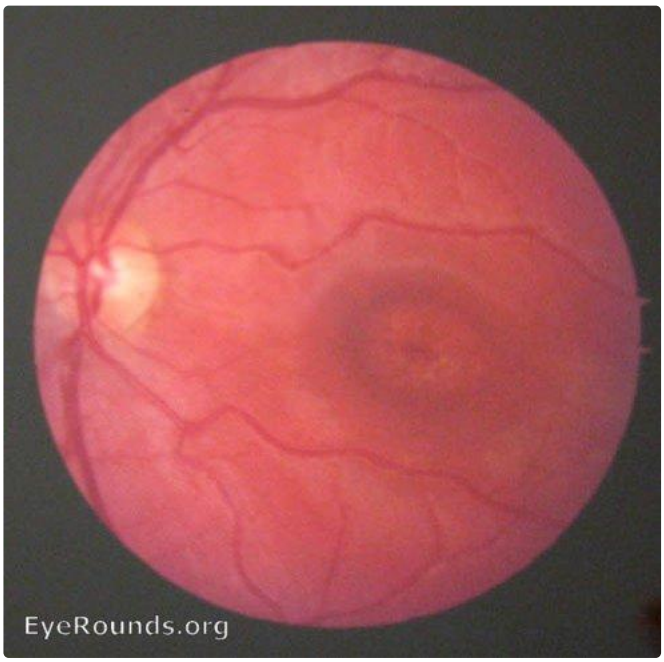
Stargardt's Disease (macular dystrophy)

Contributor: Andrew Doan, MD, PhD



1) Bullseye maculopathy.

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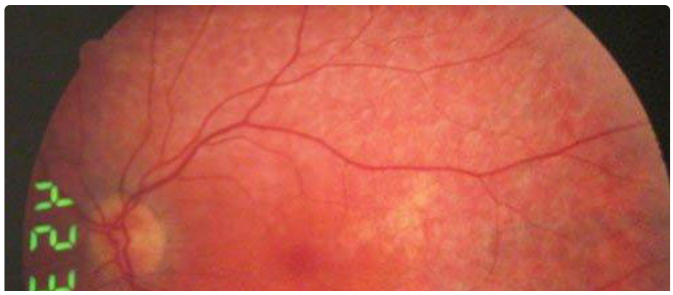

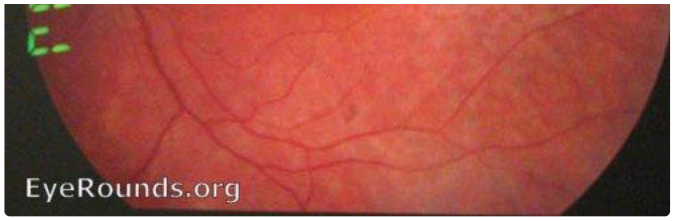

2) Pisciform flecks in the retina.

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Ed Stone, MD, PhD teaches the following:

A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Stargardt macular dystrophy.

Most common mutation is Gly1961Glu

Clinical features:

- bullseye maculopathy
- vermillion (orange) fundus and masked choroid
- pisciform flecks (due to lipofuscin A2E deposits) → If flecks are seen, then patients will usually decline to 20/200 vision within 1 year
- patients with no flecks do much better, but may develop flecks later-- less than 20/200 vision in 5 yrs after becoming 20/40.

If patients are 20/40, then they'll be 20/200 in 5 years on average.

Patients have exuberant response to incidental ocular trauma- keloid scars in macular. Avoid contact sports.

FFA demonstrates masked choroid.

Heidelberg autofluoresence is present.

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