AOO imaging of Fabry disease
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Fabry disease

• X-linked storage disorder characterized by deficient activity of the α-galactosidase A with subsequent lysosomal accumulation of sphingolipids.
• Incidence ~1/100,000 male births
• May lead to multivisceral damage primarily affecting the brain, heart and kidney.
• Ocular manifestations of FD
  – Cornea verticillata 75%
  – cataract 30%
  – retinal vessel tortuosity 50%
• Enzymatic therapy available, the effect of which on long-term prognosis is still debated.
Striated pattern of affected arterioles showing the presence of sphingolipids in smooth muscle cells
arteriole

venules
In the most severely affected cases, there may be a diffuse infiltration of the arterial wall.
Relationship with end-organ damage

• The interest of grading retinal phenotype relies on its value as a surrogate of sphingolipid infiltrations and/or of clinical severity.
• There is a more severe retinal phenotype in men, in accordance with the overall severity of the disease.
• 4 of the 5 patients with kidney disease had grade 4 or 5 retinal lesions
• Prospective studies are ongoing to confirm the prognostic interest of AO imaging in these patients.
• AOO may help to assess the effect of enzymatic therapy
To sum up

- The sphingolipids deposits of Fabry patients can be seen with high precision using AOO
- This may give insight into the understanding of complications and may contribute to management
- Storage diseases with vascular or neuronal deposits may benefit from AOO imaging of the retina
Back up
Grading: from small to large vessels, and from arteries to veins

- First vessels affected: small arterioles 20µm diameter
- Last vessels affected: largest veins close to the disc

Grade 2

Grade 3

Grade 4

Grade 5