

## [Alport Eye Abnormalities: A Patient Guide for Ophthalmology Appointments](#)

The following information was prepared by Alport Syndrome Foundation with the guidance of Medical Advisory Committee member, Dr. Laith Al-Rabadi (University of Utah), and Dr. Paul Bernstein (Moran Eye Center, Utah).

### Symptoms and Who Should Be Screened

As collagen IV is present in the cornea, lens, and retina of the eye, these areas of the eye can be affected by Alport syndrome. **Common eye symptoms in Alport patients include blurry vision and eye pain.** Eye changes are more severe in X-linked males and equally severe in both males and females with ARAS (Autosomal Recessive Alport syndrome). Eye abnormalities are uncommon in Autosomal Dominant (ADAS) patients. **All Alport patients are encouraged to do at least one baseline exam.** After an initial exam, patients may be referred to a cornea or retina specialist at the ophthalmology clinic, pending findings. An initial consult will also help establish the frequency of follow-up appointments.

### Eye Abnormalities to Check For

**Not all ophthalmologists may be familiar with the wide range of Alport-centric eye abnormalities. Please have your ophthalmologist check for the following:**

**Cornea:** *Recurrent Corneal Erosions* and *Posterior Polymorphous Corneal Dystrophy* (white dots in the cornea)

**Lens:** *Lenticonus* (anterior/posterior). This may appear as a dimple in “red reflex” on ophthalmoscope or slit lamp exam and may be mistaken for a cataract. Cataracts have been documented in Alport patients, including adolescents, but are commonly observed in those post-renal transplant.

**Retina:** *Peripheral Fleck Retinopathy* and *Temporal Macular Thinning*. This thinning of the macula can result in *Giant Macular Holes* in a small percentage of patients. These holes are difficult to repair. Alport patients with macular holes are encouraged to contact Alport Syndrome Foundation staff by email ([info@alportsyndrome.org](mailto:info@alportsyndrome.org)) as their experience can help ongoing research.

### Diagnostic Tools Your Ophthalmologist Will Require

**Please call your ophthalmology clinic in advance to ensure they can perform the following during your visit.**

- Retinal Photographs
- Ocular Coherence Tomography (OCT)
- Wide Field Color Imaging

Ophthalmologist will also likely perform the following routine examinations: Slit lamp exam; Dilated eye exam; Ophthalmoscopy

For more detailed descriptions of Alport Eye Abnormalities, including a 60-minute presentation/Q&A on this subject, please visit the ASF website: <https://www.alportsyndrome.org/eye-abnormalities/>

