

THE EYE AND VISION

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A number of eye and eye movement disorders are seen in people with Ataxia-Telangiectasia (A-T). These include (1) conjunctival telangiectasia, (2) eye misalignments (strabismus), (3) abnormal eye movement, (4) problems with accommodation (changing focus from far to near), and (5) problems with convergence (aiming the eyes together to look at near objects).

The most frequent and noticeable eye finding is prominent blood vessels on the white part of the eye wall. These vessels are called conjunctival telangiectasia and are harmless. It is not known why these vessels grow abnormally in patients with A-T. They are not present at birth, but usually become noticeable when children are 4-8 years old. In approximately 10 percent of A-T patients, telangiectasia never develop at all, and their absence can make the diagnosis of A-T more difficult. Telangiectasia have no effect on vision.

Except for the presence of telangiectasia, the eyeball is normal. Children with A-T have the same visual acuity at far distance (ability to see objects in focus) and color vision as other children, and need corrective lens with the same frequency as others. The average visual acuity at far distance of patients with A-T has been 20/30; just minimally less than a perfect 20/20. Near vision, however, may be hindered by a difficulty in accommodation. Accommodation is the ability of the eye to change focus from far to near. A delay or absence of accommodation occurs in many A-T patients. This lack of focusing at near may be compensated with the use of reading glasses.



Fig. 9.1: Telangiectasia are clusters of abnormally twisted blood vessels. They make the eye appear bloodshot.

Eye misalignments (strabismus) are seen in a much higher percentage of A-T patients than normal individuals. In most cases the eyes turn in (esotropia), but sometimes the eyes turn out (exotropia). In either case, the eyes don't work together, and the brain uses only one of the two images presented by the wayward eyes. Often, convergence is impaired as well, which means the eyes cannot be properly aimed at near targets such as books. Strabismus can be a small problem needing no treatment, or a large problem that can have major effects on vision.

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Glasses may be helpful. In some cases, strabismus requires eye muscle surgery, performed by an ophthalmologist as an outpatient procedure. The results on patients with A-T are promising with good eye alignment in most patients.

Abnormal control of eye movements is seen in most A-T patients (See Neurology Chapter for details). The cause of these problems is not clear, but it is likely to be due to progressive abnormalities in the cerebellum and the brainstem.

Many patients with A-T complain of difficulty reading. This is probably due to their inability to properly control several aspects of eye movement. This makes it difficult to move from word to word on written line or from one line of text to the next. The difficulty with convergence, properly aiming the eyes at near targets, contributes as well. While there is as yet no way to fix these problems, many adaptations can be made to improve the ability to read (see Assistive Technology chapter). Furthermore, poor accommodation may be corrected with the use of reading glasses, bringing reading materials to better focus.

Overall, patients with A-T function well on visual tasks. The major problem appears to be difficulty reading, perhaps from eye movement limitations. These often fall into the category of convergence insufficiency. Another abnormality that is encountered is misalignment of the eyes, which may require surgery. The eyeballs, aside from the dilated vessels on their surface (conjunctival telangiectasia), are otherwise normal in nearly all patients and most patients retain excellent visual acuity.

THE A-T EYE

- Most patients have telangiectasia (prominent but harmless blood vessels) on the white part of the eye (conjunctivae).
- Vision (ability to see objects in focus) is normal.
- Eye misalignments (strabismus) are common, but can be repaired.
- There is difficulty moving the eyes together to focus on close objects (convergence).
- Reading is a major problem because eye movements are not smooth, and there are problems coordinating eye movements with head movements.