



Developmental Glaucoma

FACT SHEET

(The term 'glaucoma' refers to a characteristic pattern of damage to the optic nerve)

Developmental Glaucoma is an umbrella term for a glaucoma due to a relatively rare group of eye disorders characterised by abnormal development of the front part of the eye known as the anterior chamber. People with this underlying anterior segment dysgenesis (ASD) have a significantly increased risk of glaucoma developing.

ASD is often caused by a change (mutation) in a gene that drives the development of the front of the eye, usually affecting both eyes, they may also be found in varying severity in multiple members of a family.

Although present at birth some of these conditions may not be discovered until a routine eye examination of an older child or adult. Glaucoma can occur at any age.

Ophthalmologists may use the term Anterior Segment Dysgenesis to cover several subgroups, the terminology is a little confused at times and some authors adopt the term Axenfeld-Rieger Syndrome after notable Ophthalmologists that first described families with the condition and its glaucoma complications.

1. Axenfeld Anomaly
 - a. Strands connecting iris to Schwalbe's line (peripheral cornea)
2. Rieger anomaly
 - a. Abnormal pupil shape and location (corectopia)
 - b. Extra holes in the iris / multiple pupils (polycoria)
 - c. Thin Iris (Iris Hypoplasia)
3. Rieger syndrome – iris anomalies and other body anomalies:
 - a. Distinctive facial features – widely spaced eyes (hypertelorism)
 - b. Dental abnormalities – small teeth, fewer than normal teeth
 - c. Extra skin folds around the belly button (redundant umbilical skin)

Also included under the broad term anterior segment dysgenesis can be:

4. Aniridia
 - a. Partial or complete absence of iris development
5. Peters Anomaly
 - a. Cloudiness of the cornea - front of the eye
 - b. Iris strands connecting to the cornea

Monitoring and Treatment

This group of conditions all increase the risk of developing glaucoma – pressure induced damage to the optic nerve and silent sight loss.

This glaucoma can start at any time, including soon after birth. Commonly it is found in childhood or adolescence. It is commonly quoted that people with ASD have a 50% lifetime risk of glaucoma^{1,2}, but certainly glaucoma risk is higher in those with central iris changes and more pronounced anterior iris insertion.

Thus anyone with a diagnosis of ASD should have ongoing monitoring for raised intraocular pressure that precedes glaucoma development. This may be 6 or 12 monthly monitoring depending on the degree ocular changes present.

Once someone with ASD is old enough careful documentation of the health of the optic nerve (disc photos or scans from age 5) and visual field testing is important (from ~ age 10)

If glaucoma or significant ocular hypertension is confirmed the treating eye specialist will likely initiate medical treatment to lower intraocular pressure. In a small number of patients it is likely medicine is not effective enough and surgery will be necessary to control the pressures and prevent glaucoma vision loss.

Inheritance & Genetics

Several genes are known to cause these conditions³, and they may run in families in an autosomal dominant fashion where each person is at 50% risk of inheriting the gene. Importantly from one generation to the next there may be vast variety in the amount of changes in the front of the eye and hence variability in the risk for subsequent glaucoma.

In the future it is hoped genetic knowledge will inform our ability to predict glaucoma risk and the frequency of reviews be tailored appropriately.

It is very important to check eye pressure and check for glaucoma the parents, siblings and children of anyone who has been diagnosed with anterior segment dysgenesis.

References

- 1.. Fitch N, Kaback M. The Axenfeld syndrome and the Rieger syndrome. J Med Genet 1978;15:30 –34.
- 2.. Shields MB. Axenfeld-Rieger syndrome: a theory of mechanism and distinctions from the iridocorneal endothelial syndrome. Trans Am Ophthalmol Soc 1983;81:736 –784.
3. Alward WLM: Axenfeld-Rieger syndrome in the age of molecular genetics. Am J Ophthalmol 2000;130:107-115.

Our Mission: To eliminate glaucoma blindness

T: 02 9906 6640 F: 02 9439 8736 e: glaucoma@glaucoma.org.au w: www.glaucoma.org.au

JR0814130