

Anirida and Ptosis – A Coincidental Association or Another Syndrome Complex?

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Abstract: The misfortune of aniridia rarely comes alone - it is usually associated with a host of other ocular and systemic disorders. A case of aniridia presenting with bilateral ptosis and vitreous opacification is reported here.

Keywords: Aniridia, ptosis, PAX6

A 38 year old man came to us with complaints of dimness of vision BE since childhood. He also gave us the history that he had visited many eye specialists since childhood but with no avail. On examination, he had best corrected visual acuity of 3/60 BE. Intraocular pressure as measured by GAT was 16 mm BE. In addition to bilateral aniridia, he had 2 mm ptosis in his RE with reasonably good levator function. There was also developmental cataract with phacodonesis in the same eye. The posterior segment could not be visualized due to the presence of cataract. The LE also had ptosis of slightly greater degree (3mm) but again with good levator function. There was developmental cataract but no phacodonesis in this eye. The fundus details in this eye too could not be visualized due to the coexistent cataract.

Gonioscopy done on both eyes revealed normal angle architecture. As the fundi were not visible clinically, USG was advised on BE. It showed multiple echogenic foci of low internal reflectivity in the vitreous cavity BE suggesting vitreous opacification. The ONH were anatomically normal in BE.

The patient was sent for nephrological evaluation where no anatomical lesions were seen clinically. USG and X-ray KUB were also non contributory.

As there was some useful vision – and amblyopia was indeed a strong proposition – surgical intervention for either cataract removal or ptosis correction was not considered.



Discussion

The incidence of aniridia is approximately 1 in every 100,000 live births. Three variants have been described AN 1, 2 and 3.

AN1 is the commonest variety – seen in nearly 85% of the cases. It is also called familial aniridia, and is transmitted in an autosomal dominant fashion with complete penetrance but variable expressivity. Some researchers believe the defect is in chromosome 2, but others disagree with this.

AN2 is also called Miller's syndrome or sporadic non familial aniridia. Sporadic aniridia mutations may affect the WT1 region adjacent to the AN2 aniridia region, causing nephroblastoma (Wilms' tumor). These patients

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often also have genitourinary abnormalities and mental retardation –WAGR syndrome. Often this is associated with childhood onset obesity – the so called WAGRO syndrome.

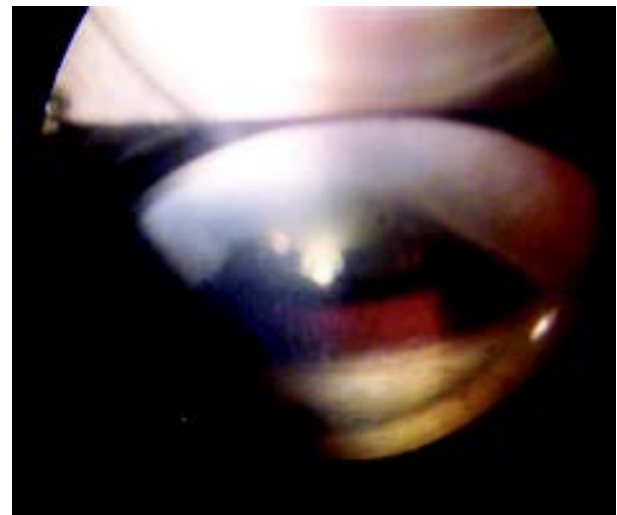
The AN2 region of the short arm of chromosome 11 (11p13) includes the PAX6 gene (named for its PAired boX status), whose gene product helps regulate a cascade of other genetic processes involved in the development of the eye (as well as other non-ocular structures). Interestingly, this PAX6 gene is around 95% similar to the PAX gene found in zebra fish, a creature which diverged from the human ancestry around 400 million years ago. Thus, the PAX6 gene constitutes an important evolutionary link to mankind's distant ancestors.

Several different mutations may affect the PAX6 gene. Some mutations appear to inhibit gene function more than others, with subsequent variability in the severity of the disease. Thus, some aniridic individuals are only missing a relatively small amount of iris, do not have foveal hypoplasia and retain relatively normal vision. Others may present with other ocular anomalies like glaucoma (due

to progressive angle closure from synechia) and AAK (aniridia-associated keratopathy). This due to alterations in corneal cytokeratin expression, glycoconjugate expression and stem cell deficiency. There is cataract development, and this is associated with a fragile lens capsule. Around 56% cases show lens subluxation, which explained phacodonesis in this patient.

AN3 is also called Gillespie's syndrome. This is autosomal recessive condition first reported in 1965 is associated with cerebellar ataxia, mental retardation and foveal hypoplasia. Wilms' tumor is conspicuously absent in these cases.

Other systemic anomalies like absent patella have also been reported. But a search of the literature did not reveal any report where bilateral ptosis and vitreous opacification coexisted with aniridia. Whether the ptosis was reactionary due to photophobia is a point to ponder, but is again not supported by available literature. This particular patient therefore does not fit into any of the specific aniridia prototypes.



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