

CASE REPORT- A CASE REPORT OF FAMILIAL ANIRIDIA**Asmita Mittal* MBBS, Saurabh Jain MBBS, Gaurav Sharma MS, Deeksha Kumari Dogra MBBS and Vishakha Dhatwalia MBBS**

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INTRODUCTION

The rare genetic condition known as aniridia (ANIRIDIA II, AN 2) is characterized by varying degrees of iris hypoplasia or absence along with other ocular characteristics, some of which are present from birth and some of which develop gradually over time.^{[1][2]}

Congenital partial or whole iris hypoplasia is the main diagnostic characteristic; foveal hypoplasia with decreased visual acuity is nearly always present and is linked to early onset nystagmus. Cataract, glaucoma, and corneal opacification and vascularization due to a lack of limbal stem cells are other commonly related ocular disorders, usually with a later onset.^[3]

In the vast majority of instances, aniridia occurs isolated without systemic involvement owing to dominantly inherited mutations or deletions of the paired box gene-6 (PAX6). It can also arise, in a minority of cases as part of the WAGR (Wilms tumor-aniridia-genital anomalies-retardation) contiguous gene syndrome in which the neighboring PAX6 and Wilms tumor (WT1) genes are both deleted.

CASE REPORT

A 32-year-old male presented to eye OPD with complaints of pain and redness after trauma to the left eye. The patient had a history of diminution of vision in both eyes since childhood. Two days back patient was walking on the road when he collided with a parked vehicle and underwent trauma in the left eye. The grade of ocular injury was Zone 3 Type B Grade 5. Then he was admitted and the open globe injury was repaired. On enquiring, it was found that the patient also had a family history of diminution of vision. On examination visual acuity in his right eye was Hand movements close to face and in the left eye there was no perception of light. On slit lamp examination, there was microcornea in both eyes (8.7mm horizontal diameter and 8.5 mm vertical diameter in both eyes) with significant opacity and vascularization (L>R). The left eye being completely opaque due to thickening and keratinization didn't allow further anterior segment examination. On further examination, it was found that there was a complete absence of iris in the right eye. There was also a nuclear cataract of grade 4 in the right eye. On fundus examination, only the optic disc could be visualized due to media haze and it was found to be pale. The patient also had bilateral jerk nystagmus. On examining his relatives, it was found that his mother, maternal aunt and maternal cousin also had microcornea with aniridia. To

rule out any abdominal tumor an Ultrasound scan KUB was ordered which did not show any abnormality. A family pedigree was drawn and the disease showed an autosomal dominant pattern.

DISCUSSION

Aniridia affects about 1.8 out of every 100,000 live births. There are three known phenotypes, with autosomal dominant aniridia being the most prevalent; it occurs in about 85% of cases and is unrelated to any other systemic symptoms. The second kind is congenital sporadic aniridia, which is linked to genitourinary abnormalities, Wilms tumor (nephroblastoma), and mental retardation (Miller's Syndrome). This second variety known as the WAGR syndrome (for Wilms' tumor, aniridia, genitourinary anomalies, mental retardation) is associated with partial deletions of the short arm of chromosome 11 (11p13). The third genetic kind of aniridia is autosomal recessive; it affects about 2% of cases overall and is linked to mental impairment and cerebellar ataxia (Gillespie syndrome). The PAX 6 gene is linked to hereditary aniridia. (4). In this case there was a clear autosomal dominant pattern of inheritance without any systemic involvement so we can say he falls in the first category of aniridia patients i.e. Autosomal Dominant Aniridia.

CONCLUSION

This case report highlights the importance of educating the patients and parents about aniridia and associated ocular anomalies and systemic problems. Genetic counseling is a must for such patients before bearing a child. Prenatal testing is indicated in pregnancies with a

risk of isolated aniridia. Hence there is a need to spread awareness among the general population about genetic conditions. Aniridia-associated keratopathy may require corneal grafting. Visual rehabilitation with the help of low vision aids may help in improving the quality of life for such patients.

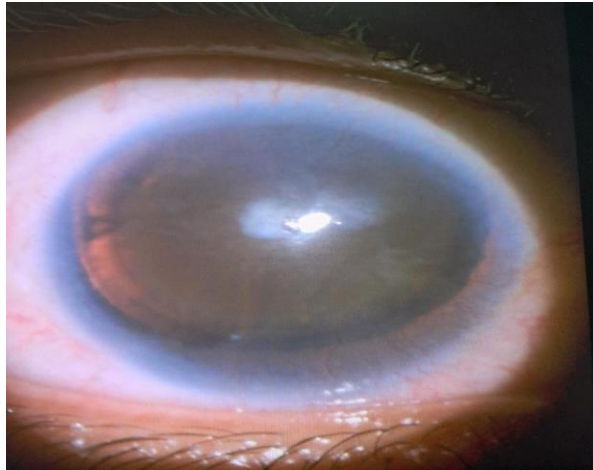
Figures

Figure 1: Aniridia and Nuclear cataract in right eye.

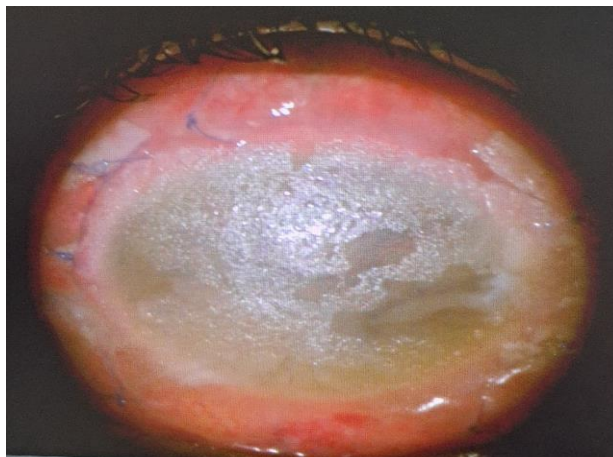


Figure 2: Aniridia associated keratopathy in left eye.

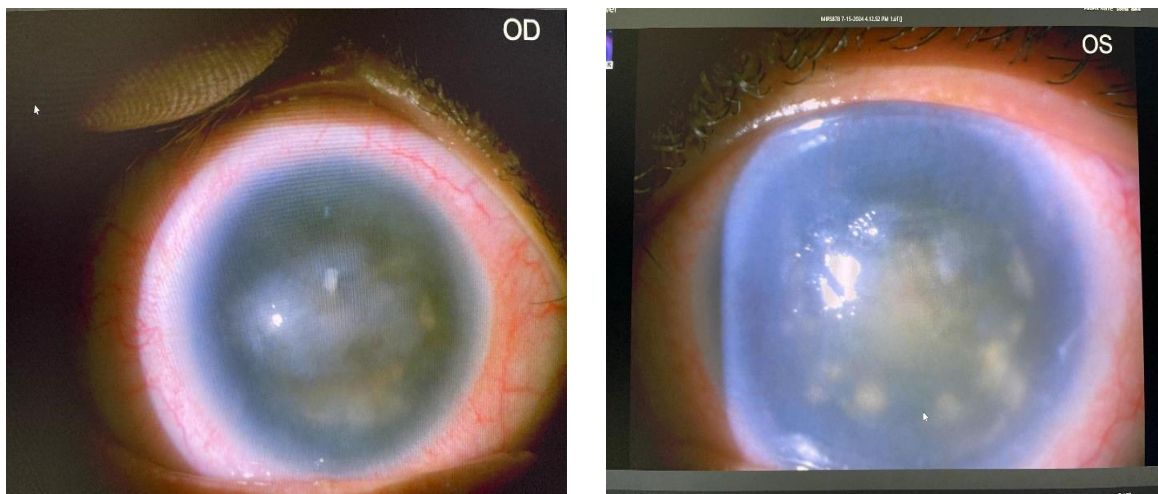


Figure 3: Eyes of patient's mother.

