

# Curtainless Windows - Approach to a Case of Aniridia

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**Abstract:** *Introduction:* Aniridia is a rare familial or sporadic disorder incidence between 1:64000 and 1:100000 affecting not only the iris but also the cornea, angle structures, lens, optic nerve and fovea and is associated with PAX-6 gene mutations. <sup>[1]</sup> In upto 85% cases, it is inherited as an autosomal dominant manner, with high penetrance, which consists of at least two-third cases of congenital aniridia. However, in up to 30% of aniridia patients, disease results from chromosomal rearrangements at the 11p13 region. <sup>[2]</sup> Congenital aniridia is described as partial or total absence of iris, but gonioscopy differentiates the two. <sup>[3]</sup> Sporadic congenital aniridia whether complete or partial is due to the de-novo mutation in the aforesaid PAX-6 gene and may consist of 13-33% of cases of aniridia. <sup>[4]</sup> It can be even traumatic in etiology but usually bilateral and symmetrical presentation will never be seen. This disorder may be associated with many other systemic abnormalities such as urogenital malignancies or WAGR syndrome (Wilms tumor-Aniridia-Genital Anomalies-Retardation).

**Keywords:** Aniridia, WAGR syndrome, gonioscopy

## 1. Case Report

A 15 year old female presented to our outpatient department with chief complaints of glare from the last 5 years. Her UCVA for both eyes was 3/60 for which refraction yielded 20/20 both eyes, anterior segment examination was normal, crystalline lens was normal, IOP was 12 and 14 mm Hg in right and left eye respectively. Then, gonioscopy was then conducted to rule out complete or partial aniridia, which further revealed iris stumps in all the quadrants. Fundus examination was done which revealed normal study.

Her systemic examination was done, as the association of any organomegaly was ruled out. She underwent USG whole abdomen and gynaecological examination was normal. There was no history of any developmental delays, menarche was at the age of 13 years. As the age of her presentation is in the second decade, the association with WAGR Syndrome is a rarer entity. As per MMSE, her score of well above 25 suggestive of no cognitive impairment.

The inference was that this was a case of congenital partial aniridia with sporadic variety with no systemic association which marks it as an atypical rare entity. As she could not afford aniridia IOL implantation, so she was trained to use coloured contact lenses (aniridia lenses) or the photochromatic glasses.

## 2. Discussion

In nutshell, aniridia is an extremely rare condition occurring in approximately one in 60,000-100,000 individuals <sup>[5]</sup>. Fraumeni and Glass <sup>[6]</sup> found seven Wilm's Tumor patients among 28 cases of aniridia which suggests that Wilm's Tumor is more common in patients of aniridia. Therefore, every case of aniridia even if it is a sporadic variety, in first

few years of life, Wilm's Tumor should be suspected <sup>[7]</sup> unless proven otherwise and a close follow-up with counselling of parents should be done and is a must-must scenario especially in developing nations as in ours where awareness is also rare.

## 3. Conclusion

Therefore, whenever we come across a case of aniridia, the take home message is to clinically rule out complete from partial aniridia using gonioscopy and nevertheless forget its local and systemic associations with neurological examination as was done in this case.

## References

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Figure 1

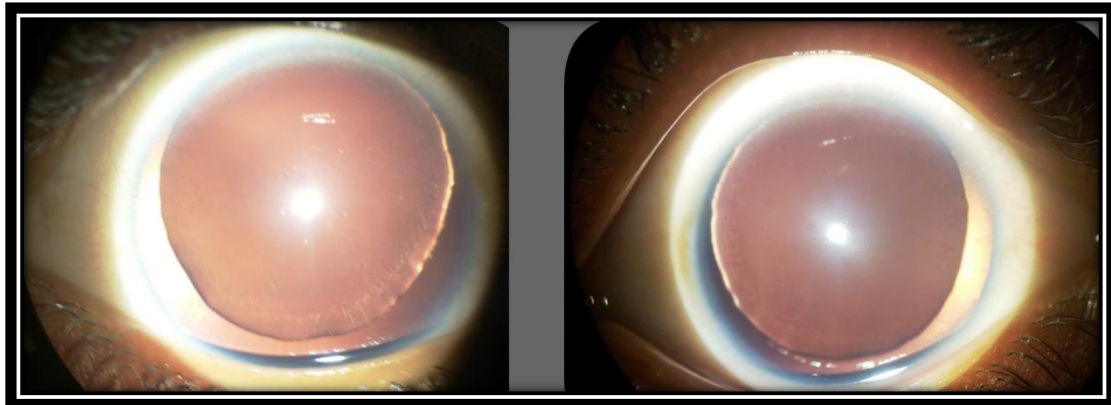


Figure 2

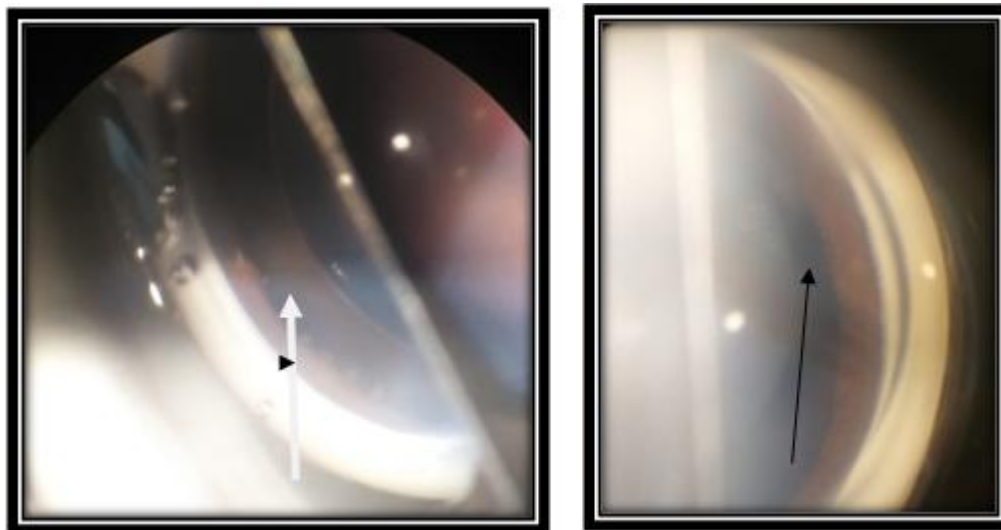


Figure 3: Gonioscopic findings- Arrows indicate iris stumps S/O partial Aniridia

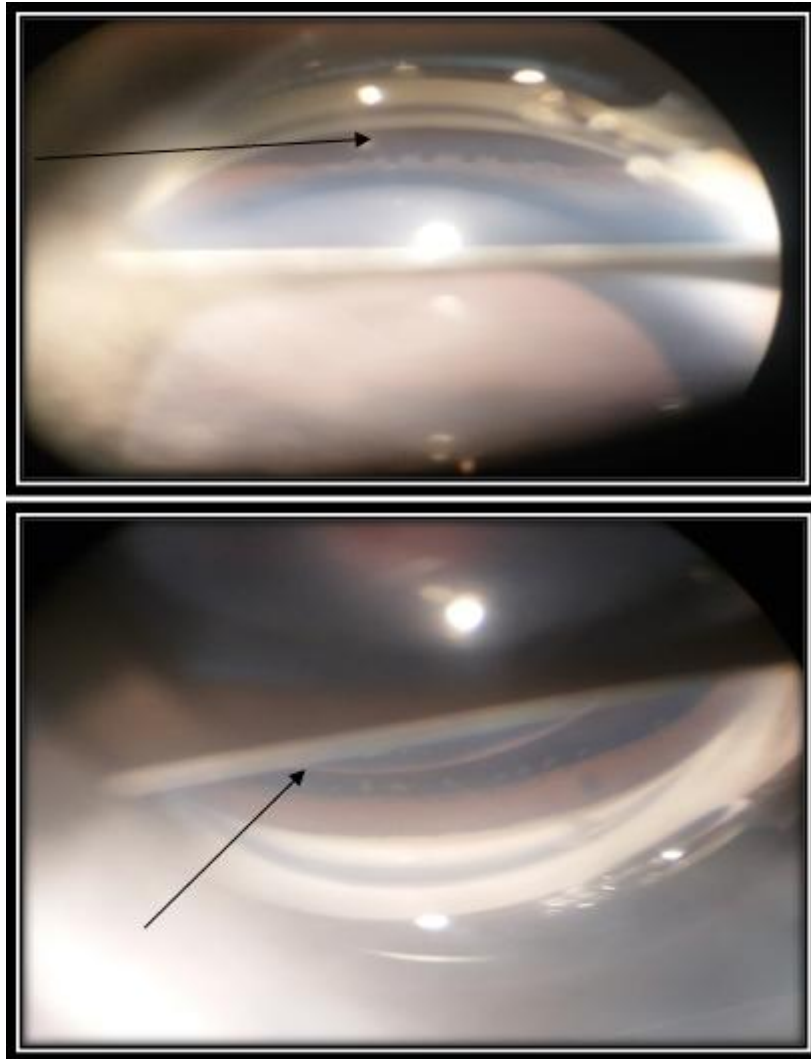


Figure 4