

## Case Report

### A Case of Autosomal Dominant Bilateral Familial Aniridia

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#### ABSTRACT

Aniridia is a familial or sporadic disorder affecting not only the iris but also the cornea, angle structures, lens, optic nerve, and fovea. This disorder may be associated with many other systemic abnormalities such as urogenital malignancies. We report a family of aniridia with different clinical spectrum of features.

**Keywords:** Aniridia, cataract, fovea hypoplasia, glaucoma, keratopathy, nystagmus, optic disc hypoplasia, panocular, PAX6.

#### INTRODUCTION

Aniridia is a rare congenital, panocular disorder characterized by varying degrees of iris hypoplasia, reduced visual acuity and nystagmus secondary to foveal hypoplasia. Other ocular findings include cataract, glaucoma, aniridic keratopathy, and optic disc hypoplasia. It may occur as an isolated finding or may be associated with systemic involvement. The prevalence of aniridia has been reported to be in 1 in 40,000 to 1,00,000 ; with no gender predilection.<sup>1</sup>

**CASE:** A 6-year-old male child was brought by mother to one of teaching hospitals of western Gujarat, to Ophthalmology department with complaints of involuntary movements of both eyes since birth. History of photophobia was present .No history of other systemic abnormalities. Birth history revealed he is first born child, FTND (full term normal delivery). Family history revealed defective vision with nystagmus in father since childhood.

**EXAMINATION OF CHILD:** Vision without glasses: (Both eye) 1/60, not improving with glasses. Examination of both eyes showed full extraocular movement with pendular

nystagmus in all gazes, fast component in lateral gaze. Anterior segment examination showed bilateral aniridia with clear lens and clear cornea (Figure 1). Fundus examination of both eyes showed clear media with temporal pale disc and normal vessels with normal macula. Child was referred to pediatrician to rule out any systemic anomaly. Regular follow up was advised for the child.

**EXAMINATION OF FATHER:** Vision without glasses : (Right eye): (hand movement) HM+, (perception of light ) PL+, (projection of ray) PR(+4), Not Improving with pinhole; (Left eye): (counting fingers near face) CFNF, HM+,PL+,PR (+4) ; with Bilateral moderate ptosis with fair LPS (levator palpebral superioris) action (figure 2), he showed pendular nystagmus in both eyes with mild divergent squint in both eyes. Ocular examination revealed mild microcornea, early corneal degeneration+ with arcus senilus+ & aniridic keratopathy, Bilateral aniridia with both eye aphakia (Figure 3). (Both Eye) fundus examination showed hazy media, (Both Eye) optic disc hypoplasia, mild tessellated background , Foveal reflex dull. Past History Significant Of (Both eye) surgery done at age of 3-4 years for both eyes subluxated lens.

**Figure-1: Typical form of bilateral aniridia, marked by almost complete lack of iris tissue**



**Figure-2: Both eyes moderate ptosis with mild divergent squint**



**Figure-3: Both eyes aniridic keratopathy**



## MANAGEMENT

Refractive correction significantly improves visual acuity in these patients. Light sensitivity (photophobia) can be managed with tinted or photochromic glasses, and wide-brimmed hats may be helpful. Patients with mild keratopathy can be managed with topical preservative-free lubricant. Moderate keratopathy is treated with autologous serum and amniotic membrane transplant. The Boston Keratoprosthesis has also been used successfully in AAK. Cataract extraction improves vision, but the surgeon should be prepared to manage complications in eyes with weak zonular fibers. Postoperatively, there may be rapid progression of keratopathy, secondary glaucoma, and macular edema.

Trabeculectomy is generally the first surgery performed in aniridic glaucoma that is not controlled on medical treatment; however, glaucoma drainage devices are a reasonable alternative. Specialized intraocular lenses that incorporate an artificial iris diaphragm can be used to correct aniridia at the time of surgery.

## DISCUSSION

This family provided a good illustration of the different clinical spectra of ocular involvement in congenital aniridia and its management. We also found similar case reports of Ten patients (30%) who had sporadic aniridia, with no previous family history; Wilms' tumor did not develop in any of them during the follow-up period. In the autosomal dominant group, ocular and systemic findings present in combination with aniridia were observed in 20 patients in the two families. Family I had aniridia and developmental delay or behavioral disorders in three generations as well as high myopia (greater than 6.00 diopters) in all affected adults. Family 2 presented a wide phenotypic variability of aniridia with myopia in three generations. Open-angle glaucoma developed in three young adults in this family, and two members were found to have gynecologic abnormalities (hypoplastic uterus and imperforate vagina). Myopia was the most prevalent refractive error (64%) in the 33 patients. Refractive correction significantly improved the visual acuity in half of these cases. Glaucoma was present in 10 patients (30%) and was the main cause of vision loss, provoking blindness in two cases (6%). Affected patients manifested progressive angle closure or presented with open angle glaucoma.<sup>2</sup>

In addition, Friedman<sup>3</sup> suggested, frequent abdominal sonography of aniridic individuals to facilitate early detection of Wilms' tumor. He followed up 11 patients with sporadic aniridia, of which two developed Wilms' tumor subsequently.

Aniridia is a rare, bilateral, hereditary absence of the iris and usually presents with a rudimentary stump of iris. Three phenotypes are recognized, of which autosomal dominant aniridia is most common (85%) and is not associated with systemic manifestations. The second phenotype is congenital sporadic aniridia, found in association with Wilms tumor, genitourinary anomalies, and Mental retardation (WAGR syndrome). Third type is Autosomal recessive aniridia associated with cerebellar ataxia and mental retardation (Gillespie syndrome). Ocular features include pendular nystagmus, decreased vision, strabismus, rudimentary iris, cataract, ectopia lentis, glaucoma, microphakia, corneal opacification, foveal and optic nerve head hypoplasia<sup>4</sup>. The visual function associated conditions like cataract, glaucoma, foveal hypoplasia, corneal opacity.<sup>5</sup>

## CONCLUSION

Aniridia is a genetic disease with possible association with other ocular and systemic disorders, which may compromise visual and vital prognosis. Genetic advice is very important in such families.

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