

# Low Vision Assessment and Management of a Congenital Aniridia and Associated Cataract

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**Abstract**—*The case discusses the low vision assessment and management given appropriate for the 18-year-old male student with congenital aniridia and associated cataract and high myopia. Congenital aniridia is a rare, usually bilateral, panocular condition that mainly affects the iris and other ocular structures, leading to secondary conditions like keratopathy, cataract, lens subluxation, glaucoma, and foveal or optic nerve hypoplasia. Due to the abnormal development in the primary structures involving the visual function, individuals with aniridia become visually impaired. As no treatment exists for aniridia, management was directed on the associated condition/s and symptoms present to improve the patient's quality of life.*

**Keywords:** *aniridia, low vision, cataract*

## I. INTRODUCTION

Aniridia, also termed as irideremia, is a rare genetic disorder characterized by a total or partial hypoplasia of the iris tissue. It only affects 1 in 40,000 to 100,000 newborns worldwide, with no gender predilection. <sup>[1]</sup> As a panocular condition, secondary anomalies are usually associated with aniridia when the following ocular structures and physiology are affected: cornea, leading to corneal pannus and/or keratopathy; intraocular pressure, leading to glaucoma; lens, leading to lens opacification and/or subluxation; fovea, leading to foveal hyperplasia; and optic nerve, leading to optic nerve coloboma and/or hypoplasia. <sup>[2]</sup> Nystagmus and photophobia at birth are both typical presentations of aniridia. <sup>[3]</sup> Another manifestation is severely reduced visual acuity of usually 20/100 or poorer that worsens as affected individual ages, depending on the associated conditions present. Congenital aniridia occurs as a result of PAX6 gene mutation — the gene that plays a major role in the early development of the eye. <sup>[4]</sup> Nearly two-thirds (66%) of the aniridia cases are considered to be the inherited type, while a third (33%) is the sporadic type which occurs due to complications experienced during gestation period. <sup>[3]</sup>

Aniridia presents itself differently in each affected individuals. It may be an isolated type affecting only the ocular structures or it may be systemically involved — often seen in sporadic aniridia. Either way, they are considered visually impaired — a term also known as low vision. Low vision, also referred to as subnormal vision or partial sight, is defined by World Health Organization (WHO) as a person who has a visual acuity of less than 20/70 to light perception or a visual field of less than 10 degrees from the point of fixation, even after treatment and/or standard refractive correction. <sup>[5]</sup> Low

vision practitioners approach patients with the objective of improving not only visual status, but also overall health of the patient.

## II. MATERIAL AND METHOD

### A. Procedure

- **Clinical Evaluation.** Low vision assessment aims to *evaluate* the residual visual function of the patient, to *correlate* it with his daily living tasks including personal, social, educational, vocational, and other needs, and to *provide* interventions to enhance and maximize the remaining vision.
  1. Case History Taking
  2. Visual Acuity
  3. Ocular Health Examination (External and Internal)
  4. Refraction (Subjective and Objective)
  5. Visual Field
  6. Motility and Binocularity
  7. Contrast Sensitivity
  8. Color Vision
  9. Light Adaptability
  10. Magnification
- **Management and Rehabilitation.** Low vision management and rehabilitation provides the *corrective aids, measures and environmental modifications* that will maximize the patient's residual visual functions and reduce his disabilities.
- **Medical Recommendations.** This includes the necessary *referrals* to other healthcare providers and the available *management option/s* not given to the patient.

### B. Data Analysis

The case discusses the low vision assessment and management given appropriate for the 18-year-old male student with congenital aniridia and associated cataract and high myopia. Congenital aniridia is a rare, usually bilateral, panocular condition that mainly affects the iris and other ocular structures, leading to secondary conditions like keratopathy, cataract, lens subluxation, glaucoma,

**III. RESULTS**

**Clinical Evaluation**

**1. Case History**

*Case Report:* An 18-year-old male student of Chinese descent, diagnosed with congenital aniridia, was presented to our eye center for a low vision assessment last May 30, 2019. The patient was accompanied by his mother during the evaluation. Past ocular history and medical history are remarkable. Family ocular history is insignificant. He has been a spectacle wearer since age 5, but discontinued wearing at age 10 because of discomfort. He finally became dependent on his spectacles at age 14 until the present. His chief visual problems are severe blurring at distance, reading small prints, and discomfort during bright day light (photophobia). At present, he has no complaints with his current prescription photochromic lens with power of -7.00Dsph. The patient denied history of significant systemic diseases, ocular or head injuries, trauma and surgeries, and recent viral illness. He is also not under any medication.

*Birth and medical history:* Patient underwent normal spontaneous vaginal delivery. It was reported that his mother experienced unusual heavy bleeding during first trimester of gestation. Sign of nystagmus was reported to be first observed by his pediatrician when he acquired fever at 2 months old. He was referred to an ophthalmologist when his nystagmus persisted even after the fever was resolved. His pathology was then confirmed with genetic testing at 6 months old. Results revealed that a gene called PAX6 was missing and that history of aniridia was traced as negative from both parents. Renal enlargement was detected during infancy, but was not given management due to the complicated clinical procedure. Intraocular pressure was within the normal range and was regularly monitored every 6 months during infancy. At age 5, tiny dot-like lens opacities in both eyes were first detected. Mandatory follow-up visits to his ophthalmologist was discontinued at age 5 until patient's next ocular visit at age 17 when he was prescribed with Timolol ophthalmic drops due to abnormally increased intraocular pressure (IOP). However, he did not comply with the medication. The patient had his initial low vision evaluation last October 9, 2018 and was then referred to an ophthalmologist for further glaucoma screening, although IOP was measured within normal range. On December 2018, the ophthalmologist diagnosed him with bilateral aniridia and cataract. He took fundus photo and perimetry tests for both eyes as requested by the ophthalmologist on March 2019.

*Tasks related history:* Patient has no problem with mobility and navigation in familiar places. He is comfortable with his daily living skills like personal management. The patient reported transportation problems when alone as he has difficulties seeing the sign boards. The patient currently attends a regular school. He sits in the first row, takes photos and audio-records the lectures to follow. He has a history of bullying in school during childhood days. He has no difficulties with

learning and excels in his classes ever since starting school. He prefers normal room illumination and has poor dark to light adaptation. He reported difficulties while copying from the blackboard and watching television. He likes to play computer and mobile games and to paint during in his leisure time. More than 10 hours of gadget exposure was reported during the summer season.

**2. Visual Acuity**

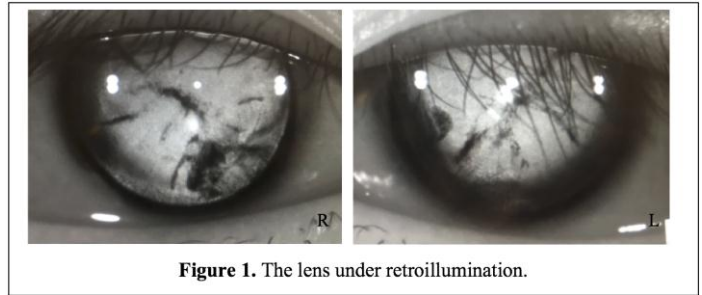
Unaided Distance: (Feinbloom chart) OD = 3m/107M, OS = 3m/107M, OU = 3m/91M

Unaided Near: (Lea numbers chart) OD = 5.0M, OS = 8.0M, OU = 5.0M @40cm

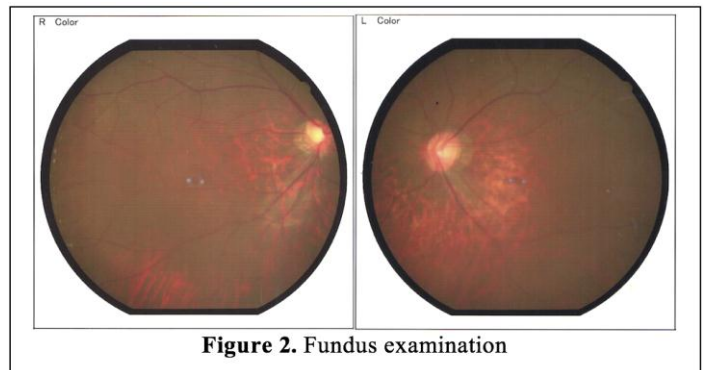
Aided Distance: (Bailey-Lovey chart) OD = 1.5m/40M, OS = 1m/40M, OU = 2m/40M

Aided Near: (Lea numbers chart) OD = 4.0M, OS = 5.0M, OU = 3.2M @40cm

- 3. Ocular Examination.** External examination revealed total iris hypoplasia for both eyes. Opacification on both lenses were also detected (see Fig. 1). IOP was measured OU = 14mmHg at 3pm using Goldmann applanation tonometer. No significant findings for the cornea, eyelids, and conjunctivas. Internal examination (see Fig. 2) interprets as follows: (1) tessellated fundus for OU; and (2) deep cupping for OD.



**Figure 1.** The lens under retroillumination.

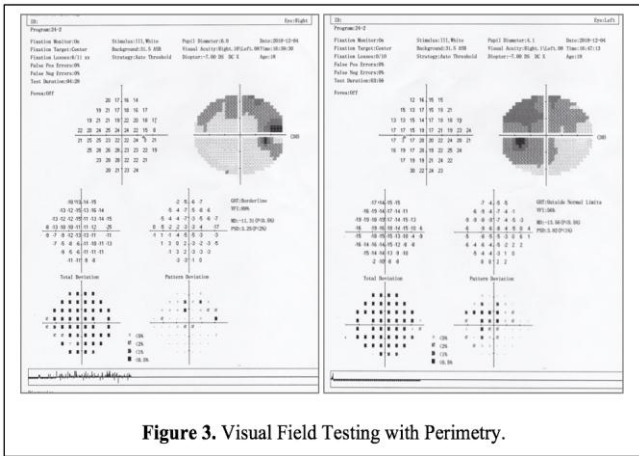


**Figure 2.** Fundus examination

- 4. Refraction.** Subjective results did not improve visual acuity with increased power. Refractive error on radical retinoscopy (objective) at 8 inches working distance yielded OD = -16.00DS and OS = -18.00DS with no

improvement in visual acuity. Patient preferred current prescription because contrast is better.

5. **Visual Field.** Both central and peripheral visual fields were not restricted bilaterally. Perimetry test as requested by the ophthalmologist also revealed insignificant results (see Fig. 3).



**Figure 3.** Visual Field Testing with Perimetry.

6. **Motility and Binocularity.** Patient exhibited nystagmus. Binocular vision using Stereofly Test was recorded as 800seconds of an arc.
7. **Contrast Sensitivity.** Using Hiding Heidi at 3m, results were recorded as OD = 10%, OS = 25%, and OU = 10%.
8. **Colour Vision.** Normal for both eyes using Farnsworth D15 and completed within 44seconds.
9. **Light Adaptability.** Patient reported that photophobia was only present when looking upwards during sunny days. He said he prefers normal indoor lighting and has difficulties with dark to light adaptation.
10. **Magnification.** Handheld video magnifier was tried out and results were remarkable. Patient's near visual acuity improved to OU = 0.80M at 40 inches and posture improved. Telescope with 8x magnification was also tried out and also yielded remarkable improvement with visual acuity of 3.9m/6.5M (Bailey-Lovey chart).

**Management and Rehabilitation**

Management

**Optical devices:** (1) Upgrade prescription lenses (same lens power) to photochromic-blue coated lens; (2) Telescope 8x and telescope training

**Non-optical devices:** (1) Bookstand; (2) cap; (3) umbrella, (4) lamp; (5) darker pen; (6) larger prints (minimum letter size: 3cm)

**Patient Education:** (1) Patient's condition and its complications; (2) visual and eye care/hygiene; (3) negative effects of long exposure to digital gadgets; (4) avoid vigorous/extraneous contact sports like basketball, volleyball, football, soccer, and the likes. Limited to sports like table tennis, bowling, chess, and the likes.

Rehabilitation

**Optometric rehabilitation:** (1) Counselling; (2) meal management; (3) money management

**Environmental modifications:** (1) Have curtains for windows; (2) organize documents by color coded; (3) avoid sitting beside window light

**Orientation and Mobility:** (1) Sighted guide technique; (2) Memorize landmarks

**Medical Recommendations**

**Available management options:** (1) Prosthetic contact lens (printed iris) with prescription; (2) handheld video magnifier; (3) Talk-back App

**Referrals:** (1) Regular visits to ophthalmologist every 6months for IOP monitoring and fundus evaluation; (2) referral to nephrologist to check overall health of kidneys

**IV. DISCUSSION**

Low vision individuals are considered visually impaired as a consequence of a visual disorder or when there's a disturbance in the normal anatomical structure. It is a condition wherein the visual functions such as binocularity, visual fields, contrast sensitivity, light sensitivity, colour vision, and more is reduced or affected. This results in poor visual performance that becomes a hindrance in the person's daily living.

Conducting a low vision assessment takes more patience and time compared with the standard clinical method. Aside from taking a detailed case history, observation of when the patient comes in is also important. One should take note of the patient's behaviors, mobility, fixation and posture. Another sign to look for is the psychology of the patient and how the patient reacts to his present condition. Importantly, one should be able to determine if the patient has realistic expectations and is receptive to help. In providing the most appropriate treatment and management, knowing and understanding the pathology matters. This will all be beneficial in how you will approach the patient and conduct the assessment.

In the case of the patient presented with congenital aniridia, he falls under the sporadic type since it was acquired during gestation and since history of the condition is not found for both parents. The first trimester of gestation is considered to be the most crucial period because it encompasses the development of organs. PAX6 gene is not only responsible in providing genetic information for the early development of the eyes, but also with the spinal cord, brain, pancreas and kidneys. [1] Since sporadic aniridia is an acquired genetic abnormality, it happens during the embryonic development and this poses more danger to affected individuals as compared to the inherited type. There is a 30% chance for sporadic aniridia to develop Wilm's Tumor, a condition associated with WAGR syndrome (Wilm's tumor, Aniridia, Genitourinary abnormalities, and mental Retardation). [3] Thus, genetic testing is essential as early as possible to confirm the genetic alteration and the type of aniridia.

Aside from the systemic complications of aniridia, secondary ocular conditions include at birth like foveal hypoplasia and optic nerve hypoplasia/dysplasia. Secondary anomalies such as corneal opacifications, lens subluxation or opacification, and glaucoma usually arise on later age onset which will result to further deterioration of visual function. Secondary condition diagnosed in the patient is bilateral cataract. Although patient's severely decreased vision could easily be identified as a symptom of foveal hypoplasia, it has to be validated with optical coherence tomography (OCT) scan. However, it cannot be completed in the patient's case due to the presence of lens opacity obstructing light entry of the instrument.

Since there is no treatment for aniridia itself, management was directed towards the patient's associated conditions and symptoms. For optical aid, an upgrade of lens coating to photochromic-blue lens with the same power was prescribed to the patient to match with patient's task related history which is long exposure to gadgets. Possible impeding conditions were also critically considered. The patient was advised to limit extraneous physical activities to avoid more retinal stretching. He was also informed of other available management options that can further aid him. Handheld video magnifier that showed significant results is highly recommended because the device has features for contrast sensitivity problems. Mandatory visit to a retinal specialist every 6 months is important to monitor not only patient's IOP but also for conscientious fundus evaluation since OCT, which is a routine method for glaucoma patients cannot be done. Visit to nephrologist was also requested as the patient never had another examination after detection of kidney enlargement.

## V. CONCLUSION

Due to the abnormal development in the primary structures of the visual function, individuals with aniridia becomes visually impaired and sometimes disabled. Providing the best management for them will be dependent on the associated condition/s and symptoms. By following the standard procedures of low vision assessment, the patient was provided with the best management available, recommendations and practical adaptations to maximize his remaining vision, reduce his disabilities (relieve the symptoms of photophobia) and improve the quality of his life.

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