



L V Prasad Eye Institute
L V Prasad Marg, Banjara Hills
Hyderabad, INDIA



**World Health
Organization**

Collaborating Centre for
Prevention of Blindness

Meera & L B Deshpande
Centre for Sight Enhancement
&

Dr P R K Prasad Centre
for Rehabilitation of
Blind and Visually Impaired

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Aniridia

Aniridia (Greek for “no iris”) is a congenital, hereditary, bilateral, extreme form of iris hypoplasia that may be associated with other ocular defects. Aniridia is an extreme form of iris hypoplasia in which the iris appears absent on superficial clinical examination. However, a gonioscopy can reveal the presence of the iris root. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time.

The pathogenesis of **aniridia** may be a primary developmental arrest of the neuroectoderm and a secondary alteration of all three neural crest waves of the mesenchyme. The disease can be inherited as an autosomal dominant trait and has an incidence of about 1/80,000 (Nelson, 1983). About one-third of all cases of aniridia are sporadic. Since the condition has a dominant transmission, proper genetic counseling is advisable. Mutations of the PAX6 gene, located on chromosome 11p13, are responsible for about 80% of cases of aniridia in humans (Ton, 1991).

Clinical manifestations may include some or all of the following:

- Aniridia alone
- Aniridia in association with systemic defects
 - ◆ Wilms tumor (20% cases)
 - ◆ Genitourinary abnormalities
 - ◆ Mental retardation
- Aniridia in association with ocular defects
 - ◆ Albinism
 - ◆ Ectopia lentis (50%)
 - ◆ Spontaneous lens dislocation
 - ◆ Arcus juvenilis
 - ◆ Keratoconus
 - ◆ Cataract (50-85%)
 - ◆ Glaucoma (30-50%)
 - ◆ Nystagmus
 - ◆ Strabismus
 - ◆ Optic nerve hypoplasia (75%)

(Singh DS)

Treatment

As of date there is no treatment for aniridia; only the associated disorders can be treated.

Implications

At LVPEI we have examined several patients with aniridia – some of the common modes of presentations include:

Reduced vision: Most patients are school-going children, with the common problem of being unable to copy from the blackboard. Patients with aniridia usually lack a foveal reflex, indicating poor macular development. True aplasia of the optic nerve can also occur. All patients need specialized management. Since they have reduced visual acuity and nystagmus, low vision devices prove very helpful, such as a hand-held telescope for distance vision tasks like board work.

Glare: The iris restricts the amount of light entering the eye; when it is absent patients are sensitive to bright outdoor light and may need eye protection. Tinted glasses or contact lenses with an artificial iris painted on it can be helpful. Sun visors, sunglasses, brimmed hats and shields are useful non-optical devices to reduce discomfort caused by glare. The teacher should avoid making such students stand with their back to the window. Doctors should bear in mind complications due to lack of stem cells while prescribing tinted contact lenses, though some of them do suggest tinted intraocular lenses (Singh, DS).

Nystagmus: This refers to an involuntary rhythmic oscillation of the eyes and is commonly noticed in congenital forms of vision loss, including aniridia. While the severity usually reduces as the child grows older, it does not go completely. Children with nystagmus often have the habit of tilting their head to attain maximum visual acuity.

Glaucoma: Glaucoma can occur in about 30-50% of patients with aniridia at any stage in life. Hence lifelong follow-up care is necessary. Both open- and closed-angle types of glaucoma can develop and medical or surgical treatment is advised to control intraocular pressure.

Keratopathy: Characteristic corneal findings in aniridia include a superficial grayish haze or opacification with a circumferential corneal epithelial pannus that advances centrally. In aniridia, pannus is thought to result from the absence of working stem cells and topical lubricants are advised to avoid further complications. In advanced cases penetrating

keratoplasty is advised but limbal stem cell deficiency needs to be considered (Mayer et al., 2003) while contemplating penetrating keratoplasty.

Cataract: Occasionally the lens in the eye may shift (subluxated) and a child with aniridia may have a cataract. This calls for cataract surgery with a special intraocular lens implant.

Tumor of Kidney: About one-third cases of aniridia occur by chance and children (usually before the age of 6) have a 1 in 4 chance of developing a malignant tumor of the kidney, Wilm's tumor. Frequent ultrasounds and other tests are necessary to try and catch the tumor early, which is highly treatable in the early stages. Most children with WAGR syndrome who develop Wilm's tumor are diagnosed before the age of 3. WAGR syndrome is a rare genetic syndrome in which affected children are predisposed to develop Wilm's tumor, aniridia, genitourinary anomalies and mental retardation.

Aniridia – Cerebellar Ataxia – Mental Deficiency (ACAMD): Aniridia, cerebellar ataxia, and mental deficiency, also known as **Gillespie syndrome**, is an extremely rare inherited disorder characterized by the absence, in whole (aniridia) or in part (partial aniridia), of the iris; impaired coordination of voluntary movements due to underdevelopment of the brain's cerebellum (cerebellar ataxia); and mental retardation. The condition usually affects both eyes though in a few cases only one eye is affected. Some individuals may have psychomotor retardation.

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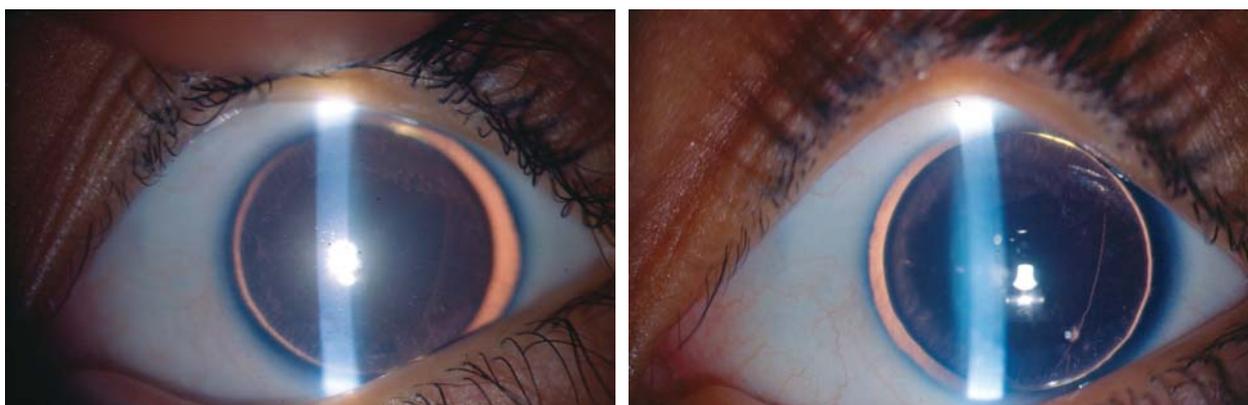
News and Events

- ◆ Low vision awareness program: The 14th low vision awareness program (LAP) for ophthalmologists, optometrists and rehabilitation professionals was conducted from March 31 – April 2, 2006, with 30 participants from across the country.
- ◆ Dr Raman (ophthalmologist), Sarojini Devi Eye Hospital, Hyderabad, Ms Asha Fofaliya (optometrist), Mumbai, and Mr Alok Bhandari (optometrist), Jyoti Eye Hospital, Visnagar, Gujarat, completed a 3-month short-term fellowship program in low vision care from April – June 2006.
- ◆ Ms Jasmine Davis completed a 3-month short-term fellowship program in low vision care from May – July 2006.
- ◆ Mr Sukanta Ghosh, optometrist from Calcutta Eye Research Institute, Kolkata, completed a one-month observation at the Vision Rehabilitation Centres in May 2006.

A case of aniridia with glaucoma and cataract

Anita, her parents' second child, was 4 months old when she was diagnosed with aniridia with glaucoma and cataract in both eyes. She underwent glaucoma surgery in the right eye at 6 months of age. When she was first examined at LVP she was 4 years old and her intraocular pressure in both eyes was still uncontrolled. Her best-corrected visual acuity was 6/60 and 6/38 in the right and left eye respectively. However, her near acuity was N8 at 10 cms. She read and watched TV from a very close distance and had to depend on friends for board work.

At the Vision Rehabilitation Centres her parents were reassured that reading or watching TV from a short distance would not harm Anita's eyes. She was advised to use a telescope for board work and underwent glaucoma surgery in both eyes. However she managed to complete board work with the assistance of her friends and teachers. While her intraocular pressure was under control the cataract in the right eye increased and she underwent cataract surgery with special intraocular lens implantation in the right eye.



Slit lamp photograph of a case of Aniridia

Anita then developed band shaped keratopathy in the right eye for which she underwent phototherapeutic keratectomy. She was advised to use topical lubricants for the right eye despite which she developed corneal scarring and lost significant vision in the eye. When last examined a couple of months ago her best-corrected visual acuity was 1/38 in the right eye (up to 1 meter) and 6/38 in the left eye.

Anita is now 15 and has done well in her tenth class exams. She was counseled at the Vision Rehabilitation Centres regularly and also given a letter requesting her teacher to allow her extra time for writing exams. Anita plans to become a Chartered Accountant. She has been going for a kidney ultrasound regularly, and is fine. She now feels the need for a telescope to cope with board work and use of the telescope would enable her to follow the teacher's instructions from the board like her normally sighted peers.

Anita's case demonstrates that life-long follow-up is essential in aniridia to deal swiftly with complications. A team of professionals comprising ophthalmologists from different subspecialties, low vision specialists, rehabilitation professionals and school teachers play a vital role in enhancing the child's quality of life.

Enrol now!

Short-term Fellowship Program in Low Vision care

Duration: 3 months

Program begins on: January 1, April 1, July 1, and October 1.

Minimum qualification: Diploma in optometry or Master's degree in ophthalmology – preferably institution-based

Registration is limited to two candidates per program.

Registration fee: Indian Rupees 4000

Low Vision Awareness Program

Duration: 3 days

Program begins in: March and September

Eligibility: Ophthalmologists, optometrists and rehabilitation professionals

Registration fee: Indian Rupees 1500

Vijaya K Gothwal

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Programs sponsored by Sir Ratan Tata Trust, Mumbai

Volunteers activities

- ◆ Volunteers of the Vision Rehabilitation Centres conducted spoken English classes for visually challenged children to boost their confidence and improve communication skills to enable them to access e-books and specialized software.
- ◆ A volunteer Mr Siddharth Singh gifted a software 'Spin it Again' that converts recorded material from audio-cassettes to compact discs. The advantages of the technology include reduced storage space, portability, printing option, and savings in time.

Some of the other activities that our volunteers have been involved in include:

Voice Recording: Ms Bharati Devi, Ms Ratnamala, Ms Sandhya, Ms Akhila, Ms Latha, Ms Neena Varghese, Mr Srinivas, Ms Rekha Manvi, Ms Madhuri Kiran, Ms Indira, Ms Praveena Gupta, Mr Sessaiah, Ms Shanti Thirumalai, Ms Varalakshmi, Ms Sugandham, Ms Bhagya Prasanna, Ms Kanyakumari, Ms Shamala, Dr Shanti Vijeyapal, Ms Aarathi Selvan, Ms Soumya Reddy and Ms Rajakumari.

English Classes: Ms Bharati Devi, Ms Ratnamala, Ms Neena Varghese, Ms Shakti Nigam

Typing: Ms Suma and Mr Ganesh

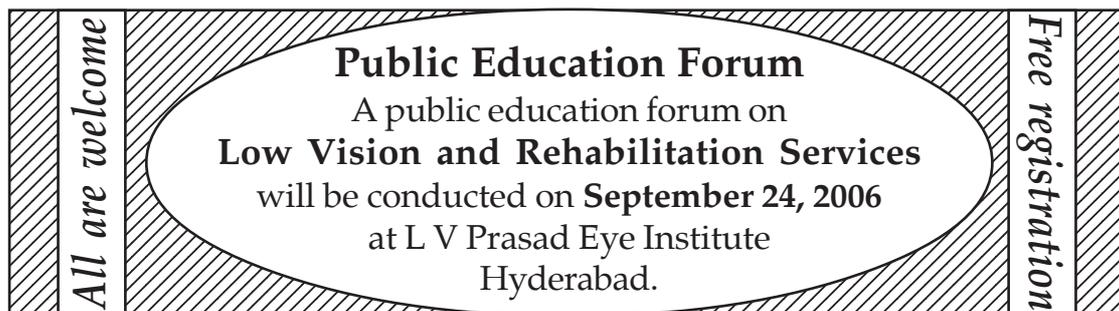
Translation of Assessment Tool: Ms Akhila Ravi

Editing Educational Material: Ms Taffy Johnson, Mr Siddharth Singh, Ms Rekha Manvi, Mr Ganesh

Placements: Mr Rajiv Kumar and Ms Sandhya Reddy

Donations

- ◆ Mr Praveen Kumar Gottipalli, an NRI and a committed supporter of the Vision Rehabilitation Centres, contributed \$ 1000 for the purchase of JAWS (version 7.0), a screen reading software for the visually challenged. The software converts text to speech so that individuals with visual impairment can work with computers like normal sighted persons.
- ◆ A volunteer Mrs V S Rukmini raised Rs 10,000 for the children's camp.
- ◆ Ms Taffy Johnson and Mr Karen Sawyer, LDS Charities, USA, donated material worth Rs 50,000, including audio-cassettes, CDs, and an audio recorder for preparation of talking books.



You can make a difference

Your contribution can help the Vision Rehabilitation Centres in several ways: provision of low vision devices to underprivileged children, training optometrists in detection and rehabilitation of the blind and those with incurable low vision, and conducting community programs for rehabilitation of persons with visual impairment.

Contributions to the Hyderabad Eye Institute and Hyderabad Eye Research Foundation are tax deductible. Donations above Rs 250 are exempt under Section 80G of the Income Tax Act 1961 for Hyderabad Eye Institute and under section 35(i) (ii) for Hyderabad Eye Research Foundation.

For more information please contact

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Greeting cards based on paintings by children with visual impairment are available for sale. Please contact Vision Rehabilitation Centres, LVPEI.