Welcome to Retina India.

All about Retina, for those with an interest in Retina & Retinal Ailments.

June 2012

Honorary Editor: **Giridhar Khasnis** **June 2012**

"I have interacted with many visually challenged players and I can say honestly that these players have a special talent to see the pieces in their mind when they play. For us, it is very difficult to be able to imitate that special skill."

- Grand Master Viswanathan Anand -
World Chess Champion

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C O N T E N T S:

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GENETICS OF RETINAL DISORDERS: ISSUES FOR PATIENTS & THEIR FAMILIES

by Dr Meenal Agarwal & Dr Shubha R Phadke

Introduction:

Retina is the innermost layer of the eyeball which works like a camera and converts light rays into neuronal signals. These neuronal signals are sent to brain via optic nerves and an image is interpreted. Retina itself is a 10 layered structure comprising of photosensitive cells, neuronal cells and their network and supportive cells.

Retinal disorders can be acquired due to trauma, drugs, diseases like diabetes and hypertension or can be age related (age related macular degeneration). Many of retinal disorders are genetic in origin and multiple members may be affected in a single family.

Specialized tests like electroretinogram (ERG), electro-oculogram (EOG), dark adaptation test, visual field test and fluorescein angiography can help in establishing specific diagnosis and require interpretation by an ophthalmologist specialized in retinal disorders.

Careful history and detailed examination of family members can point towards specific mode of inheritance and hence guide for genetic testing. In many of the retinal disorders with genetic causes, only single member is affected (sporadic cases). For e.g., the risk of recurrence of retinitis pigmentosa in a family with only one patient can vary from 0-50%.

In a family with more than one case of retinal disease, drawing a family tree (pedigree) helps in concluding about the mode of inheritance of the disorder in concern. The pedigree also helps in giving information about possible or definite carriers of the disorder in the family. Of course it is necessary to consult a medical geneticist for getting accurate information about the risk of recurrence and genetic tests as there are many issues like variable expression, non-penetrance of the disease, genetic and phenotypic heterogeneity of the disease, etc. The process of consultation with a medical geneticist regarding information about genetic aspects of the disease is known as genetic counseling.

For the sake of explanation of how it works, we will consider the example of one retinal disease, Retinitis Pigmentosa (RP).

Retinitis pigmentosa:

RP is the most common cause of hereditary retinal degeneration. It is also called pigmentary retinopathy or rod cone dystrophy. It is characterised by primary degeneration of rod photoreceptors thus presenting with night blindness and loss of peripheral vision. Central vision may be preserved till late stages of disease. In later stages, cones are also affected affecting visual acuity and many patients become legally blind by adult age. Age of onset may vary from infancy to adulthood.

RP is mainly isolated problem without any effect on any other organ of the body (nonsyndromic RP) but in few cases other organ systems can be affected (Syndromic RP). To rule out syndromic forms of RP, a thorough clinical examination of patients and their family members are required preferably by a clinical geneticist.

Genetic testing

Molecular genetic testing for RP is mainly guided by inheritance pattern in the family as clinical presentation in all types of RP is almost same. Out of all the cases 50% are sporadic i.e. only single member in the family is affected. Once the genetic defect in a patient is identified, other family members who are at risk can also be tested or prenatal diagnosis in pregnancy can be provided with certainty.

The ophthalmological findings usually do not provide any clue for the gene to be tested first. Hence, one gene after the other needs to be tested till the disease causing mutation is identified. This mammoth task has been eased out by the development of next generation DNA sequencing testing as it can simultaneously look at many genes and identify the mutation.

The DNA based testing may not be necessary for the diagnosis of cases of RP with classical manifestations. But it plays an important role in diagnosis in early stages of disease and cases without similarly affected family members. The DNA based testing is becoming more important as some treatments based on the specific genetic defects are in the pipeline.

The mutation detection in the patient also helps in testing possible carriers of the disease and providing them genetic counseling and prenatal diagnosis of RP to prevent recurrence. Mutation detection is also useful for the presymptomatic diagnosis of family members. However, the presymptomatic diagnosis and prenatal diagnosis for RP is ethically debatable and has many psychosocial and legal aspects to be thought of and counseled for.

Management

Apart from the current management paradigms for RP, which are mainly supportive in nature, a significant amount of research work in the field of molecular and gene therapy may herald a new way of treating this untreatable condition.

Success of gene therapy for at least some genetic types of RP has been convincingly shown in the research settings. These options have opened the possibilities and raised hopes for the patients of RP in recent future.

Genetic counseling is essential in all of these cases as to look for any syndromic association, examination of other family members, molecular testing and ultimately predicting risk of recurrence and early detection of disease in relatives who are presumed to be at high risk.

Summary

RP is an important cause of inherited visual loss. Its progressive nature and currently non-availability of preventive or curative treatment raises many complex issues for the patient and families. This makes genetic counseling imperative for the family. Family history data and DNA based tests play an important role in genetic counseling and management of the patient and the family. RP is a rapidly evolving field and ongoing enormous research in the field of gene therapy and other molecules for therapy, which are based on better understanding of the pathophysiology of the disease. There is a significant amount of hope for RP treatment in the near future.

***Note**: This is the summary of an article written specifically for RI Newsletter by Dr. Meenal Agarwal and Dr. Shubha R Phadke, Dept. of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India. RI Newsletter expresses its gratitude to the writers.*

If you wish to read the complete article, please click here.

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ELECTRONIC IMPLANTS RESTORE RUDIMENTARY VISION TO TRIAL PATIENTS

Recently, the newspaper "TheGuardian", UK (3 May 2012) has reported that doctors, led by Tim Jackson and Robert MacLaren, surgeons at Kings College and Oxford respectively, were able to restore rudimentary monochrome vision to two trial patients, only weeks after surgery.

Chris James, 51, who lost his sight to the disease retinitis pigmentosa (RP); and Robin Millar, a 60-year-old music producer from London were the first blind patients to be fitted with electronic eye implants in a UK clinical trial.

While Millar revealed that he had been able to detect light and distinguish the outlines of certain objects, James reportedly said that "it'll give me some imagery rather than just a black world."

The surgery involved implanting a 3mm-by-3mm chip (similar to the camera in a mobile phone and containing 1,500 light-sensitive elements that replace the damaged cells in the patient's eye) through a small flap in the delicate retina at the back of the eye of the patients.

Delighted with the initial results, MacLaren hoped that the electronic chips would provide independence for many people who are blind from retinitis pigmentosa. Made by the German company Retina Implant AG, the chips have been in trials for more than six years.

Developers were expected to seek approval for the device if the latest trials became a success. David Head, of the charity 'RP Fighting Blindness' reportedly said: "The completion of the first two implants in the UK is very significant and brings hope to people who have lost their sight as a result of RP."

For Nick Astbury, a consultant ophthalmic surgeon and chair of 'Vision 2020 UK', the trial was the first step on a long journey to help people with sight loss to see again and live independently.

Source

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Online Braille library

Using technology to empower the physically challenged, the government has set up an Online Braille library to make books and reading material in digital format accessible to visually impaired persons.

Launched on January 4, 2012, the Online Braille library, established by National Institute for Visually Handicapped (NIVH), Dehradun, has books which can be accessed in real time from any location.

For assisting visually challenged people in locating their books, an online catalogue of Braille books has been hosted by NIVH which enables visually-impaired persons to ascertain the location and availability of a desired Braille book, without going to Braille libraries and presses. NIVH has also launched an Online Directory of Services for the Visually Impaired Persons.

NIVH is located in an area of about 43 acres on Mussoorie-Dehradun Highway. An autonomous body under the Ministry Of Social Justice and Empowerment, Government Of India, the Institute is a training centre for blind. IT has a Model School for the Visually Handicapped which provides education to blind, partially sighted and low-vision children from Nursery to Class X. The blind children are taught through Braille and tactile sensation whereas the partially sighted and low vision children make use of magnifying devices to read the printed text. The scholars are provided with free board, lodging, clothing, tuition and other facilities besides monthly pocket money.

NIVH is also engaged in production of Braille literature, Aids and Appliances for Visually handicapped. The Institute has its regional center in Chennai to provide rehabilitation services in the southern states of the country. It renders vocational training at par with NIVH. It also provides rehabilitation services to the rural-based visually handicapped persons through its Community Based Rehabilitation activities.

NIVH also has two Regional Chapters at Kolkata (W.B.) and Secunderabad (Andhra Pradesh) providing peripheral services.

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If you wish to read more about NIVH, please click here & here.

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Smart shoes to guide the blind

A group of students from Mangalore are trying to make walking easier for the visually impaired with the help of a smart shoe with GPS-enhanced sensory heels.

The smart shoe has three sensors around the front end of the shoe. The sensors, connected to the microcontroller, will start vibrating on detecting an obstacle, the intensity depending on the distance. The user can get an approximate idea about the width of the object as sensors are fitted in both shoes. All the components including the IR sensors, GPS, vibrator and buzzer are connected to a microcontroller.
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On detecting an obstacle, IR sensors in the shoe pass on the information to the microcontroller which analyses which sensor found the obstacle. A voice module directs the user based on GPS readings and the algorithm in the module.

Moisture-proof shoes can be built for use during the monsoon, and the project can be further enhanced by adding a GSM module which can store important telephone numbers. A video camera can also be attached to the shoe so that the user can be monitored from any part of the world and guided if an error occurs in the GPS module. If the GSM module supports 3G, there can be video transmission in real time.

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TECHNOLOGY & VOLUNTEERS HELP VI STUDENTS

Technology has made a world of difference to the lives of a group of visually impaired students by helping them fare well in the II PU exam. Leaving aside time consuming classes in Braille, Samarthanam Trust for the Disabled, Bangalore made all the lessons available on the computer, enabled with screen-reading software, to them and it paid off too. Yogaraj R., Sameer Mubarak Sanadi, Ashok R., Bhagyshri and passed the examination, with the first two students securing distinction.

“None of us here look at ourselves differently because of our disability,” said Yogaraj, who is also a national-level athlete. “We take it as a challenge and never let it come in the way of our growth.”

Students have dedicated a large part of their success to the volunteers who spent a lot of time with them. “Volunteers sat with us from 10 a.m. to 4 p.m. Lessons became simple when they read them out to us and cleared our doubts,” said Yogaraj.

“We give recordings of the lessons to the students and repeat important points in class to make sure that it stays in their memory,” said Shobha Chittaranjan, a volunteer. “The integrated approach bringing the visually impaired to the mainstream has helped everyone in the classroom.”

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FREE RESIDENTIAL MUSIC SCHOOL for blind girls

The Pandit Puttaraj Gawai Trust for the Disabled set up by Deva Reddy N. Chinchali in 2003, runs a free residential music school in Bangalore for visually challenged girls aged between 14 and 25. Students at the residential school have the opportunity to learn Carnatic, Hindustani, Hindi and Kannada light music, and train in veena, keyboard and the harmonium. The Trust sponsors accommodation, meals, clothes and other essentials

for its wards who all hail from economically backward families in rural Karnataka.

A disciple of the late Pandit Puttaraj Gawai, 42-year-old Reddy is a blind Hindustani musician who also teaches music at a Government Junior College. He holds graduate degrees in History (Bangalore University) and Music (Karnatak University, Gadag).

On June 22, 2012, the Trust added another feather to its cap by releasing the Carnatic Junior music exam syllabus book in Braille. It is working on transcribing both Carnatic and Hindustani music syllabus books in Braille by 2014.

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BUDDING MUSICIAN

Krutika Janginmath, 14, plays Hindustani music on flute. The soft-spoken, visually impaired girl has participated in over 60 concerts in almost all districts in Karnataka, and in Andhra Pradesh and Maharashtra . She was just three years old when her parents saw her responding to the notes of the harmonium during a religious programme. She started training in Hindustani music under Danamma Gidaveera and Ramanna Hadapada, both visually impaired artistes. Babaji Naikwad taught her the first lessons in flute. Now, she is being trained by Chandrashekar P.

While her mother spent two months to learn Braille, her 18-year-old brother Kartik learnt the tabla so that he could accompany her.

She has performed at events such as Navaraspur Utsav in Bijapur and Sharana Samskriti Utsava in Chitradurga and met stalwarts such as Gangubai Hangal, Pandit Puttaraj Gawai, Pandit Hariprasad Chaurasia and flautist Praveen Godkhindi.

An admirer of Bhimsen Joshi, Krutika is a now Class 9 student. She plans to learn keyboard and is looking for a teacher.

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VI ON WHEELS

About 150 participants were seen in action at a car rally organised by the Bangalore Knights Round Table (BKRT) 174 and the National Association for the Blind on Sunday, 17 June 2012.

Visually impaired participants from the NAB were paired with other registered participants who would have to drive the cars on a 50-km long obstacle course based on navigation instructions given by their visually impaired

partners reading from a map in Braille.

The rally was designed to be a time-speed-distance test and the participants lost points for arriving too early or too late at a checkpoint. The youngest of all participants was 14-year-old G.V. Ganaraj Gowda from Mathru Education Trust. The rally was followed by a fashion show in which four visually impaired participants from NAB walked the ramp with other models. The proceeds of the programme would be used by NAB to construct a free school for the underprivileged in Whitefield.

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Donate your expertise or your time. Volunteer for Retina India. You can take up projects, mentor someone or choose to be a reader for a visually challenged. Take that first step. Make a difference in someone's life. Write to us at info@retinaindia.org.
Donate money: You can send a crossed check, demand draft payable at Mumbai, or money order for any amount you wish to donate in the name of "Retina India", with a cover letter, to the following address: Retina India, Ridhi Sidhi Bhavan, 2nd floor 2/12, Babu Genu Road, Mumbai 400 002, India.

Please mention your full name, address and contact details so that we can send you a receipt for your donation. If you wish to discuss donation to Retina India, you can call +91 98200 19584, or +91 89399 11897.

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