Case Report

Association Of Retinitis Pigmentosa(Rp) With Mental Retardation(Mr) and Subluxated Lens

Dr. Sneha T. Shah*(Corresponding Author) ,Dr. Nilesh V. Parekh, Dr. Neepa R. Gohil***, Dr. Aditya Parekh******

*2nd Year Resident Doctor, **Professor and Head of Department of Ophthalmology, ***Assistant Professor of Ophthalmology, Department of Ophthalmology, Sir T. Hospital, Bhavnagar 364001,Gujarat, India.

****MBBS,22 Behind CSMCRI,Bhavnagar 364001,Gujarat,India.

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ABSTRACT

Retinitis Pigmentosa is pigmentary retinal dystrophy characterized by triad of perivascular pigmentary changes resembling bone corpuscles,presence of pale and waxy optic disc and attenuated thread like retinal blood vessels on fundus examination which is associated with common syndromes.We report a case of 35 year old Mentally Retarded Male having Subluxated lens with Retinitis Pigmentosa.

INTRODUCTION

Retinitis Pigmentosa is slowly progressive bilateral primary pigmentary retinal dystrophy due to apoptosis affecting rods more than cones resulting in night blindness(earliest feature), which is inherited as Autosomal Recessive condition, affecting Males more commonly, showing characteristic triad of perivascular pigmentory changes resembling bone corpuscles,presence of pale and waxy optic disc and attenuated thread like retinal blood vessels on fundus examination which is associated with common syndromes.

CASE REPORT

35 year old Mentally Retarded Male presented with chief complaint of Left eye Redness,Pain and Watering since 3 days.Torch Light findings were confirmed by Slit Lamp Examination on which Left eye was Edematous Congested having Corneal Edema,Shallow Anterior Chamber,Semidilated Fixed Pupil and Anteriorly Subluxated Lens with Intraocular Pressure(IOP) not recordable on Non Contact Tonometer(NCT) and patient being uncooperative for applanation tonometry.Right eye having Inferotemporally Subluxated Lens with 15 mmHg IOP on NCT.Due to Mental Retardation,Patient was unable to cooperate for Visual Acuity testing.Glow not seen on Fundus examination with Subluxated Lens in both eyes.Both eye having Vitreous Degeneration on B Scan Ultrasonography.

Surgical Management planned after written and informed consent from legal guardian.Psychiatric evaluation of patient was done.Preoperatively Tobramycin eye drops(eds) four times/day(QDS),Prednisolone eds 6 times/day and Hypertonic saline eds 5 times/day was started in Left eye with Intravenous 500ml 20% Mannitol Stat and Tablet Acetazolamide (250 mg) QDS with banana or coconut water.On IOP control next day Left eye cataract extraction with Iris Claw Lens Implantation was done under General Anesthesia(Figure 1).Postoperatively Fundus Examination of Left eye had tilted disc with pallor,generalized vessel attenuation and bony spicules indicating Retinitis Pigmentosa(Figure 2).5 months later Elective Right Eye Cataract Extraction with Iris Claw Lens Implantation was done under General Anesthesia(Figure 3,4).Postoperative fundus examination of right eye revealed similar picture of Retinitis Pigmentosa(Figure 5).

DISCUSSION

Common Syndromes associated with Retinitis Pigmentosa:

1. Laurence Moon Biedl Syndrome
   Most Common
   Mental Retardation + Polydactyly + Obesity + Hypogonadism
2. Cockayne Syndrome
   Mental Retardation + Ataxia + Nystagmus
3. Kearns-Sayre Syndrome
   Ocular Myopathy + Heart defects

Correspondence Address : **Dr. Sneha T. Shah**
E-mail : tosnehashah@gmail.com
Mob no. : 07977893689/09969673287

4. Refsum disease

   Polyneuropathy + Cerebellar Ataxia + Deafness

5. Usher's Syndrome

   Labyrinthine deafness

6. Barren-Kornweig Syndrome

   Abetalipoproteinemia + Acanthocytosis

7. NARP Syndrome

   Neuropathy + Ataxia

But association of Retinitis Pigmentosa in Mentally Retarded with Subluxated Lens is a new presentation.

Have we found a New Syndrome?

CONCLUSION

Common Syndromes are associated with Retinitis Pigmentosa but we present case of 35 year old Mentally Retarded having Subluxated Lens with Retinitis Pigmentosa which is a new presentation.

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