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OCULAR COMPLICATIONS IN MARFAN'S SYNDROME PRESENTING FOR VISUAL DISABILITY CERTIFICATE : CASE REPORT.

Ophthalmology

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ABSTRACT

Ths report is of a 31 year old female who presented to the outpatient ophthalmology department of civil hospital Palampur. Patient had come for the visual disability certificate. Examination of the patient revealed features which were in keeping with Marfan'ssyndrome and ectopia lentis right eye and history of retinal detachment in left eye. The patient was refracted and recommended glasses which improved her vision were prescribed. The patient was also referred to the cardiologist for further evaluation and management. Multi disciplinary approach should be adopted in the management of Marfan's syndrome so as to prevent complications most especially life threatening ones and improve quality of life

KEYWORDS

Marfan's Syndrome; Ectopia Lentis; Retinal detachment

INTRODUCTION

Marfan's syndrome is a genetic disorder of the connective tissue. It is an autosomal dominant disorder involving the cardiovascular, skeletal and ocular systems.[1] It was first described by Antoine – Bernard Marfan in an 1896 case report of a young girl with unusual musculoskeletal features.[2] In 1991the firillin-1(FBN 1) gene mutation on chromosome 15 was identifid as a cause of Marfan'ssyndrome. Fibrillin, a glycoprotein provides force bearing structural support and elasticity of theocular connective tissues . The main ocular features of Marfan syndrome, all of which can result in decreased vision, include bilateral ectopia lentis (lens dislocation), myopia and retinal detachment . About 50% of patients with Marfan syndrome are diagnosed by an ophthalmologist; some individuals may present with isolated ocular signs suggestive of this syndrome .[3] Progressive aortic dilatation, usually maximal at the sinus of Valsalva, associated with aortic valve incompetence leads to aortic dissection or rupture and is the principal cause of mortality, but mitral valve prolapse with incompetence may be significant, and lens dislocation, myopia and arthritis associated with chronic joint laxity can cause substantial morbidity.[4] The ophthalmic features of Marfan's syndrome were first described in 1914 by Boerger a Peadiatrician. Worldwide, the incidence of Marfan syndrome is approximately 7-17/100,0004.[5]

Clinically affected individuals often have tall stature and dolichostenomelia, lens dislocation, aortic dilatation, skeletal manifestations as precuts deformities and/or scoliosis. Ocular features of this syndrome have been repeatedly reported. Ectopia lentis,the most common ocular feature, occurs in 70 to 80 % ofcases. Isolated reports of spontaneous complete posterior crystalline lens luxation, or developing secondary complications like glaucoma or uveitis have also been described.[6]MFS is usually associated with normal intelligence. The diagnosis and management of Marfan syndrome requires a multidisciplinary team approach, in view of its multisystem effects and phenotypic variability. A DNA diagnostic service for FBN1 gene testing for MFS and related clinical entities is available .[7]

A case report

This paper presents 31 years old female who, presented to out patient department of ophthalmology at civil hospital, Palampur,HP for visual disability certificate. She was tall, of thin built. She had disproportionately long limbs compared with the trunk, long spider-like fingers (arachnodactyly)(fig 1), long toes with prominent finger joints , elongated face and mild joint hypermobility.

The examination of CNS intact , Chest was clear and no abnormality detected in abdomen . On examination of cardiovascular system revealed left parasternal heave, both S1 and S2 sounds were audible. The chest was clear and no abnormality was detected in the abdomen. Ocular examination demonstrated a visual acuity of 1/60 in the both eyes which improved to 6/36 with pin hole in both eyes , with further no improvement in visual acuity after refraction.. Intraocular pressure was 13 mm Hg bilaterally. The anterior segment was quiet normal in both eyes ,lens was clear, pupils briskly reactive. Undilated anterior segment evaluation on slit lamp biomicroscopy revealed a superiorly subluxated lens in the right eye(fig 2) and aphakia in left eye. Examination of the right eye under mydriasis revealed a deep and irregular anterior chamber with superiorly subluxated lens from 8 to 2o'clock .She underwent surgery for retinal detachment in left eye 1 year back. Posterior segment revealed pink optic disc, cup disc ratio was 0.3:1 in both eyes with peripheral chorioretinal degeneration in both eyes. A subjective refraction and cycloplegic examination was done patient accepted +6.00 Ds R/E , +5.00 DS L/E with improvement to 6/36 in both eyes. The patient did not have any detected systemic illness.

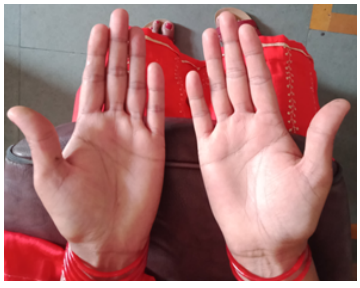

Fig 1. Arachnodactyly (long fingers)

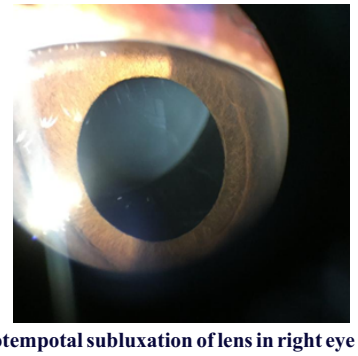

Fig 2. Superotempotal subluxation of lens in right eye

DISCUSSION

Ocular complications are commonly found in Marfan Syndrome. Of them, bilateral spontaneous superotemporal subluxation of lens is a common manifestation . Posterior dislocation of lens a rare manifestaion , may be silent and rarely causes glaucoma or uveitis. Commonly observed changes include liquefaction of the gel and posterior vitreous detachment(PVD). Posterior dislocation of lens could be due to the disruption of anterior hyaloids face resulting in loss of support. This causes more severe retinal complications as total rhegmatogenous retinal detachment, secondary glaucoma, which need prompt and aggressive treatment to minimize the degree of visual loss.[8] Ectopia lentis is potentially visually debilitating but visual acuity varies with the degree of malposition of the lens.The presence of ectopia lentis is a major criterion for the diagnosis of Marfan's syndrome which unequivocally establishes the diagnosisof Marfan's

syndrome in 86% of cases.[4] Common non-surgical management in ectopia lentis include refractive correction and pharmacological manipulation of the pupil. There is often amblyopia even with a lens of normal transparency and it is caused by optical changes due to the fact that the equator of the lens occupies the optical centre of the lens .[9] Our patient had a visual acuity of 6/36 after full cycloplegic correction with spectacle. She was given 40% disability as per the guidelines by government for gradation and certification of visually handicap.[10] The patient was advised to follow up regularly for further early detection of ocular complication. Follow up with cardiologist was advised to detect life threatening complications like dissection of the aorta early. The family members were also examined to rule out the cases of Marfan syndrome.

CONCLUSION

As our patient presented for visual disability certificate, so there is need to effectively manage the ocular features of Marfan's syndrome so as to prevent amblyopia and loss of vision. There should be a multidisciplinary approach to the management of Marfan's syndrome so as to prevent life threatening situations that can arise and further improve the quality of life of the patient.

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