Bilateral Ectopia Lentis: Presentation and Management in Child with Marfan Syndrome.

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Case Report
Subject: Ophthalmology

Abstract:
Marfan syndrome is inherited connective tissue disorder involving cardiovascular, ocular and musculoskeletal system. Incidence varies from 1/5000 to 1/20000. It is characterized by weakness or incompetence of the connective tissue due to defects in fibrillin. It is described by Antoine Marfan in 1896. Aortic root aneurysm and ectopia lentis are the cardinal clinical features. In the absence of family history, the presence of these two manifestations is sufficient for confirmatory diagnosis of Marfan syndrome. Patients with ocular manifestation should be screened for cardiac manifestation to support diagnosis. FBN1 mutation analysis helps in prompt diagnosis. There's no cure for Marfan syndrome, so treatment focuses on managing the symptoms and reducing the risk of complications. Recent advances in diagnosis, improved surgical technique and application of prophylaxis has contributed in preservation of sight in patients.

Keywords: Marfan syndrome, ocular Manifestation, Management etc.

Introduction:
Marfan syndrome is an autosomal dominant condition characterized by abnormal biosynthesis of fibrillin (major constituent of micro fibrils) due to mutation of FBN1 gene on human chromosome 15. This results in body's ability to stretch abnormally in response to stress [1]. In three-quarters of cases it is inherited from parents and in remaining quarter cases is sporadic [2]. Men and women are equally affected. Ocular manifestations include bilateral ectopia lentis, myopia and retinal detachment. 50% of patients are diagnosed by ophthalmologist due to early occurrence of ocular signs and symptoms and some patients may present with ocular involvement only [3].

Case History:
Seven year old male child presented to our ophthalmology department on 3rd March 2012 with complaints of progressive diminution of vision with glare in both eyes since past 8 months. Parents denied any other medical, surgical, family and trauma history. Ocular examination revealed visual acuity in right eye was 6/12 (with correction of +6.5 D spherical; 2.0 D cylinder 1300) and left eye was 6/18 (with 8.5 D spherical; 3.5 D cylinder1450). Ocular motility was full and free in all direction of gaze. Slit lamp bio microscope examination of anterior segment revealed clear cornea in both eyes without any corneal ectasia and megalocornea. Both eyes angle were deep without any opposition to cornea in either side. No evidence of anisocoria and relative afferent papillary defect. Superotemporal subluxation of lens was noted in both eyes. Intraocular pressure was 16 mm Hg (with Tonopen) for both eyes. Dilated fundoscopy examination showed normal Posterior segment without peripheral retinal degeneration or detachment.
General examination revealed skeletal abnormalities like long, thin extremities, arm span greater than height, arachnodactyly with prominent finger joints and high-arched palate. Patient was referred to cardiologist and echocardiography revealed mitral valve prolapsed with mild dilatation of aorta. Patient was started on beta blocker for the same. Diagnosis of Marfan syndrome was made on clinical findings due to non-availability of genetic study at our institute.

Patient was operated for extraction of clear lens followed by foldable intraocular lens placement in capsular bag with capsular tension ring under general anaesthesia. His intraocular lens power was 23.0 D and 23.5 D for OD and OS respectively. But intra operatively lens with power of 21 D and 21.5 D power was implanted for OD and OS respectively (as 10% reduction in lens power was considered due to paediatric age group). His postoperative course was unremarkable with excellent improvement in his visual function. His postoperative visual acuity (unaided) was OD 6/12 P and OS 6/12. On slit lamp examination intraocular lens implanted was well centered.

He was advised frequent follow-up due to the potential of the development of future ocular complications like glaucoma, retinal detachment which requires prompt and aggressive treatment.

**Discussion:**

This patient had presenting complaint of decrease in vision and glare attributed to bilateral Superotemporal subluxation of lens with poor improvement in vision following refraction correction. Parents denied such history in family members. Patient was referred to cardiologist with suspicion of Marfan syndrome for further evaluation. Echocardiography finding of mitral valve prolapsed and mild dilatation of aorta supported the diagnosis of Marfan syndrome in absence of family history. Mutation analysis of FBN1 gene helps in prompt diagnosis but it was not available at our institute [4].

The presence of ectopia lentis is considered as major criteria in revised Ghent nosology for diagnosing Marfan syndrome and it alone can diagnose 86% of patients with this syndrome [3, 5]. It is usually bilateral, symmetric, super temporal and non-progressive present in 50-80% of affected individual [6]. It varies from mild
asymptomatic displacement to significant subluxation resulting in monocular diplopia [3]. Anterior dislocation of lens results into pupillary block glaucoma or chronic angle closure glaucoma. Posterior dislocation results in posterior uveitis or chorioretinal inflammation due to leakage of lens proteins [3]. Non-surgical management includes refractive correction and application of miotic drugs.

Surgical indication for lens extraction include lens opacity, anisometropia, non-correctable refractive error, impinging total luxation of lens and lens induced glaucoma or uveitis [3, 7]. Presence of zonules weakness and capsular instability makes implantation of an intraocular lens difficult with amplification of usual complication of lens extraction. Surgical options include anterior chamber IOL, ciliary sulcus posterior chamber IOL fixed to the sclera and/or to the iris, and scleral fixated capsular tension rings. Capsular tension rings, is suitable option as it allows preservation of the capsular bag and primary implantation of IOL. It is 270open PMMAring causes even distribution of centrifugal forces thought the zonules. These rings contain holes that allow centering and fixation of the capsule bag to the scleral wall. Recent reports shows good visual outcomes without any serious complications in surgery [3, 8], our patient had excellent improvement in vision following surgery.

Myopia is considered as minor criteria in revised Ghent nosology for diagnosing Marfan syndrome and is present in 34-44% of affected individuals [3, 9]. Ectopia lentis and retinal detachment are associated risk factors.

Retinal detachment is severe complication seen in 5-11% patients. It occurs due to vitreous traction by ectopia lentis. Other associated risk factors are globe elongation, vitreous liquefaction, posterior vitreous detachment, retinal thinning and lattice degeneration. Surgeries for retinal detachment in Marfansyndrome are challenging because of thin sclera, poor dilated pupils, ectopia lentis and multiple retinal breaks [3].

Strabismus is seen in 19-45% patients with Marfansyndrome and if uncorrected may lead to amblyopic. It occurs due to instability of extra ocular muscle pulley with delayed and inadequate correction of refractive errors. Surgery is treatment of choice for strabismus [3].

Our patient did not show any other ocular finding on examination. He was advised frequent follow up with ophthalmologist for early detection of other ocular features and follow up with cardiologist and orthopaedician was also suggested to improve quality of life and to help early detection of life threatening complications like dissection of aorta.

**Conclusion:**

Ophthalmologist plays a vital role in detection of Marfan syndrome. Timely diagnosis and treatment of ocular manifestations helps in preserving better eye sight in patient.

**References:**


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