OCULAR PRESENTATION OF MARFAN SYNDROME- DIAGNOSIS AND MANAGEMENT

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ABSTRACT

BACKGROUND

Marfan syndrome is an inherited systemic connective tissue disorder primarily associated with skeletal, cardiovascular and ocular pathology. Clinically affected persons have overgrowth of the long bone, thickening of atrioventricular valve and lens dislocation.

MATERIALS AND METHODS

A hospital-based clinical study was carried out on patients complaining of diminished vision in the Outpatient Department of Ophthalmology, Patna Medical College and Hospital. Patients of all age group with inclusive criteria of Marfan syndrome were studied.

RESULTS

This syndrome has no ethnic or gender preference and shows high penetrance but variable expression, both interfamilial and intrafamilial clinical variation is common.

CONCLUSION

The ocular complications commonly occur in Marfan syndrome as bilateral spontaneous posterior lens dislocation, retinal detachment and secondary glaucoma.

KEYWORDS

Marfan Syndrome, Ectopia Lentis.

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BACKGROUND

Marfan syndrome is an autosomal dominant genetic disorder caused by misfolding of extracellular matrix protein, Fibrillin-1 (Gene: FBN1).[1,2] It is a connective tissue disorder with varied expressions in the body with ocular and systemic involvement. The incidence is reported to be 1 in 10,000 live births and approximately one-fourth of cases are sporadic.[3] The prevalence of Marfan syndrome is at least 1/5000,[4] 2 - 3 per 10,000 individuals[5] or 1 in 9800.[6]

MATERIALS AND METHODS

A hospital-based clinical study was carried out on patients complaining of painless diminished vision from July 2016 to June 2017.

Inclusion Criteria

Patients were selected on the basis of following criteria: Tall stature and long bone overgrowth with lens dislocation, unilateral or bilateral along with cardiac abnormalities attending to outpatient department of ophthalmology and patients referred to the department of ophthalmology from other departments.

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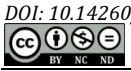

Exclusion Criteria

1. Non-compliant, immunocompromised patients.
2. Urine metabolic screen or fasting plasma amino acid to exclude homocystinuria.
3. Chromosome karyotype to exclude Klinefelter syndrome.

Materials Used

1. Visual acuity by Snellen's chart.
2. IOP measurement by Schiotz tonometer.
3. Slit lamp examination.
4. Gonioscopy.
5. Direct ophthalmoscopy.
6. Indirect ophthalmoscopy.
7. Fundoscopy using +90D lens.
8. Lab investigations.

RESULTS

A total of 108,000 patients attended PMCH eye outpatient department in the above-mentioned period. Out of these, 4 cases (of the same family) presented with bilateral lens dislocation and secondary glaucoma. Three patients showed bilateral supero-temporal lens dislocation and one patient showed supero-nasal bilateral lens dislocation. All patients had long, thin extremities and increased arm span. We also came across five cases of lens dislocation due to other causes (Trauma, Homocystinuria, Weill-Marchesani syndrome, Hyperlysinaemia).

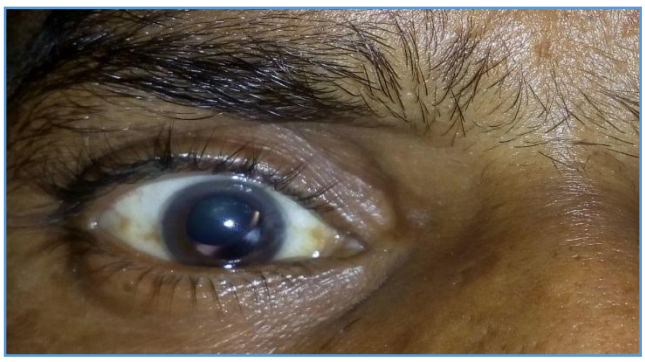

Figure 1. Temporal and Upper Subluxation of the Lens as well as Enlarged Axial Diameter of the Eye

DISCUSSION OF CLINICAL PRESENTATION

Marfan's syndrome is a systemic disease affecting cardiovascular, musculoskeletal and ocular system. The ocular features of Marfan syndrome results in decreased vision including bilateral ectopic lentis 80%, (subluxation is most frequently superotemporal), myopia and retinal detachment. Subluxation usually develops in early childhood, but may first appear in the second decade. Lens dislocation into anterior chamber may occur. Patient with Marfan syndrome may present with flatter cornea and megalocornea may present. Hypoplastic iris or ciliary muscle caused decreased miosis can also be seen and considered minor criteria for ocular system involvement in Ghent nosology.[7] Myopia is associated with an increased risk of retinal detachment. With the disease primary open angle glaucoma is most common, but glaucoma can also be secondary to anterior lens dislocation, anterior chamber angle abnormalities or surgery.[8]

DIFFERENTIAL DIAGNOSES

- Trauma.
- Homocystinuria.
- Weill-Marchesani syndrome.
- Hyperlysinaemia.
- Sulfite oxidase deficiency.
- Isolated familial ectopia lentis.

DIAGNOSIS

The diagnosis of Marfan syndrome to be locus heterogeneity as well as the larger size of the FBN1 gene and lack of family history in about one-third of cases, also molecular testing is of limited help.[9] Clinical diagnosis depends on a combination of major and minor signs as defined in revised 1996 Ghent nosology.[7] The existence of ectopic lentis is considered a major criteria for the diagnosis of Marfan syndrome in this nosology, which unequivocally diagnose or excludes Marfan in 86% of cases.[6] Slit lamp examination with full dilated pupil for the detection of ectopia lentis and other ocular abnormalities should be part of every initial evaluation of Marfan syndrome.

DISCUSSION OF MANAGEMENT

Common non-surgical intervention in ectopia lentis includes refractive correction by glasses and pharmacological manipulation of pupil. Indication for surgical lens extraction include lens opacity with poor visual function, anisometropia or refractive error not responsive to optical correction, impending complete luxation and lens-induced glaucoma or uveitis.[10.11] With the development of new technique and instrument, surgical option has increased to improve visual acuity in patients with ectopia lentis. The principle surgical method in Marfan syndrome is lens extraction with either IOL placement or contact lens correction. Long-term follow-up after lensectomy with limited anterior vitrectomy and subsequent correction of aphakia with glasses or contact lens showed this procedure to be safe for children with hereditary ectopia lentis.[11] Because of zonular weakness and the resultant capsular instability, correction of aphakia could be carried out by ACIOL or scleral fixated PCIOL.[12]

CONCLUSION

About 50% of patients with Marfan syndrome are first diagnosed by an ophthalmologist. Some patients may present with isolated ocular signs. Timely diagnosis and treatment of refractory complaints, retinal detachment and glaucoma and timely surgical intervention can prevent amblyopia and help the patients to preserve the vision.

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