Original Research Paper

Volume-10 | Issue-2 | February - 2020 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Architecture

MARFANS SYNDROME - A CASE REPORT

Dr. K. Srujana* Post graduate Department of Ophthalmology, Rangaraya Medical College, Kakinada ,DR.NTRUHS, Vijayawada, India *Corresponding Author

Dr. P. Viswamithra Professor, Department of Ophthalmology, Rangaraya Medical College, Kakinada, DR.NTRUHS, Vijayawada, India

ABSTRACT Marfans syndrome is a rare genetic disorder with an incidence of 1 in 5000 world wide. It is an autosomal dominant connective tissue disorder. It is a systemic disease that classically affects cardiovascular, musculoskeletal and ocular systems. Our paper reports the diagnosis of this rare syndrome in a 8 year old male child based on clinical features.

KEYWORDS : ectopia lentis, , marfans syndrome, arachnodactyly, tall stature .

INTRODUCTION

Ectopia lentis is the dislocation or displacement of the natural crystalline lens. Ectopia lentis may occur after trauma or may be associated with ocular or systemic disease. Simple ectopia lentis can occur as a congenital disorder or as a spontaneous disorder later in life1.Marfans syndrome is the most common cause of heritable ectopia lentis ,and ectopia lentis is the most frequent ocular manifestation of Marfans syndrome, occurring in approximately 75 % of patients. . It has worldwide prevalence rate of approximately 1 per 5,000 live births[2].

CASE REPORT

An 8 year old male child born out of non-consanguineous marriage presented to OPD of Ophthalmology with progressive painless decreased vision in both eyes since childhood. Detailed family and medical history was taken which revealed mother had normal hospital based vaginal delivery. In family, sibling and near relatives are normal. On general examination he is tall statured (FIGURE 1)with pectus excavatum, kyphoscoliosis, hypermobile joints , arachnodactyly(FIGURE 2) and pes panus . On local examination best corrected visual acuity is OD - CF 1/2metres not improving further ,OS – 6/36 not improving further, OU - Near vision reads N8 without glasses. Anterior segment – OU - superotemporal subluxation of lens (FIGURE 3) , pupils are brisk . Fundus (FGURE 4)showed clear media with normal disc, physiological cup with normal foveal reflex. IOP is within normal limits. Gonioscopy showed open angles . A-scan & B – scan(FIGURE 5)were normal .Patient was referred to department of General Medicine & Cardiology for systemic evaluation.

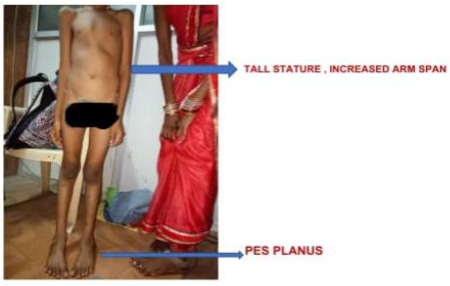

FIGURE 1: GENERAL PHYSICAL EXAMINATION

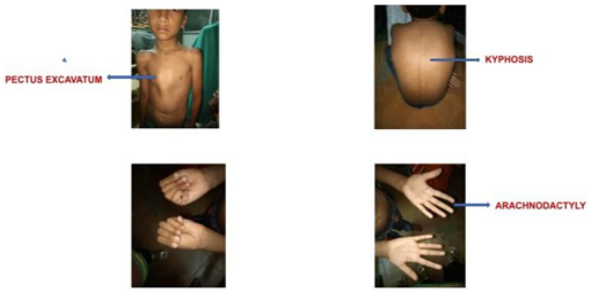

FIGURE 2: SYSTEMIC FEATURES

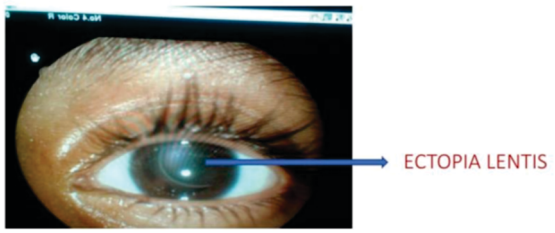

FIGURE 3 : ECTOPIA LENTIS

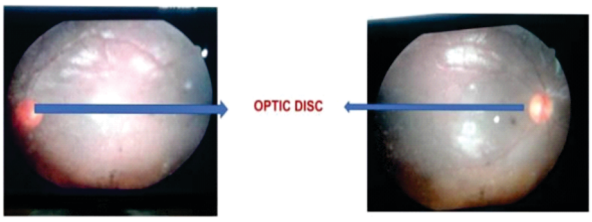

FIGURE 4 : COLOUR FUNDUS PHOTOGRAPH

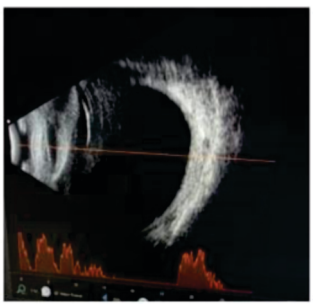

FIGURE 5 : NORMAL B-SCAN

DISCUSSION

Marfans syndrome is a systemic disease with ocular abnormalities occurring in 80 % of patients.It is caused by mutation in Fibrillin-1 (FBN1) gene on chromosome 15q21.1. Ectopia lentis (50%) being most common ocular abnormality followed by myopia , astigmatism ,iridodonesis / phacodonesis,smal, poorly dilating pupils retinal detachment .Investigations are made on basis of clinical systemic features, complete ophthalmic(slit- lamp, retinoscopy, refraction , detailed fundus) examination, and echocardiogram. Diagnosis of Marfans is made through **Revised Ghent Nosolgy**. Diagnosis can be confirmed by genotyping .Medical management involves refractive correction as well as co -management of any associated systemic disease. As this is hereditary, genetic counselling may be indicated. Multiple surgical techniques exist for exist for surgical correction of ectopia lentis. Few of them are lensectomy /vitrectomy with aphakic contact lens , iris fixated IOL etc. Indications for lens extraction were delineated by Nemet et al . In order to prevent complications secondary to aortic root dilatation, beta blockers are most often used.

CONCLUSION

Management of patients with Marfans syndrome requires multidisciplinary approach and early diagnosis is important. From the above findings taking aortic criterion , systemic score >= 7, ectopia lentis , he is diagnosed as case of Marfans Syndrome.

REFERENCES

1. Cross HE & Jensen A.D: Ocular manifestations in Marfans syndrome & Homocystinuria. AM J Opthalmol. 75: 405, 1973.
2. Robinson P, Godfrey M . The molecular genetics of Marfans syndrome and related microfibrillopathies. J Med Genet 2000; 37:9 -25
3. Nelson LB , Maumenee IH. Ectopia lentis .Surv Opthalmol 1982; 27: 143-60
4. De paepe A , Devereux RB , Dietz HC , Hennekam RC , Pyeritz RE . Revised diagnostic criteria for the Marfans syndrome. AM J Med Genet 1996; 62: 497 – 26.
5. Sridhar J , Chang JS . Marfans syndrome with ectopia lentis. N Engl J Med 2017; 377: 1076.