Case Report :

MARFAN SYNDROME IN SIBLING

Dr. Sudhanshu Kumar, Prof. Dr. Suchitra Dash, Prof. Dr. B.N.R. Subudhi,
Asso. Prof. Dr. P. Panda, Asst. Prof. Dr. S. Panigrahi

Marfan syndrome is AD disorder which is associated with fibrillin1 gene mutation on chromosome 15 characterised by triad of eye ,heart and musculoskeletal involvement. Ocular manifestation of marfan syndrome are supero temporal subluxation of lens usually but may be in any meridian , angle anomaly, retinal degeneration ,cornea plana, strabismus. We report two cases (SIBLINGS) 10yrs and 12 yrs female who presented with diminution of vision in both eye since 2 yrs. On examination of first patient VA 6/36 with pin hole no improvement in both eye , intraocular pressure 27.2mm of Hg in RE and 17.3 mm of Hg in LE , slit lamp examination of both eye shows supero nasal subluxation with infero temporal intact zonules ,. On examination of 2nd patient VA 6/24 with pinhole no improvement in both eye , intraocular pressure in 17.3 mm of Hg both eye, slit lamp examination shows superotemporal subluxation with inferonasal intact zonules ,intraocular pressure 17.3 mm of Hg in both eye, . Both patient presented with long and slender finger, arm span greater than height , high arch palate which were consistent with that of marfan syndrome. 1st patient treated trabeculectomy and correction of refractive error .And 2nd patient treated as correction of refractive error.

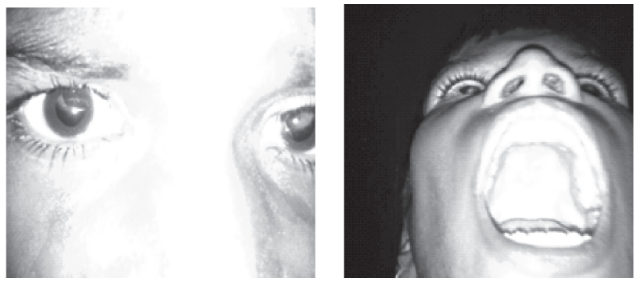
KEY WORDS; MARFAN SYNDROME, SECONDRY GLAUCOMA AND LENS SUBLUXATION

INTRODUCTION : Marfan syndrome is AD disorder which is associated with fibrillin1 gene mutation on chromosome 15 characterised by triad of eye ,heart and musculoskeletal involvement. Ocular manifestation of marfan syndrome usually include supero temporal sublaxtion of lens and rarely posterior dislocation of lens which causes uveitis and glaucoma. Other ocular manifestation are vitreous liquefaction, disruption of anterior hyaloids feces, angle anomaly, retinal degeneration ,cornea plana, strabismus

We hereby reporting two cases of marfan syndrome in same family

CASE REPORT 1

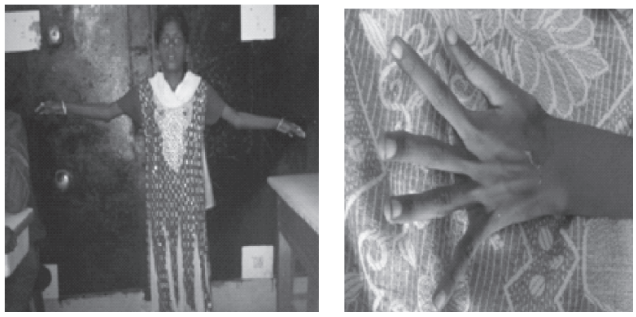
A 12 year old female presented with diminution of vision in both eye since 2 years. VA is 6/36 with pin hole no improvement in both eye and intraocular pressure in Right and Left is 27.2 mm of Hg and 17.3 mm of Hg respectively .On examination under slit lamp there was supero-nasal sublaxtion of lens in both eye with intact zonules in infero-temporally. Fundoscopy revealed normal fundus and there was no vitreous degeneration in both eye.



SUBLUXATION (SUPERO-NASAL) HIGH ARCH PALATE (BOTH EYE)

Gonioscopic examination showed trabeculo-dysgenesis in right eye. Other manifestation in favour of Marfan syndrome were long and slender fingers ,arm span greater than height and high arch palate in this patient.

Patient's parent were not willing for any type of operative procedure but agreed to

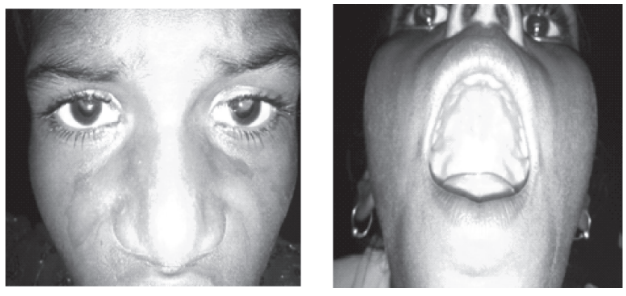


ARM SPAN > HEIGHT
LONG & SLENDER FINGERS

underwent trabeculectomy only after explaning the consequences of increase intraocular pressure. During discharge patient was asked to come after 2 weeks. Intraocular pressure on follow up was 17.3 mm of Hg in both eye. Correction of refractive error with -3.5 sph -1.75 cyl at 180p degree was given. VA after correction was 6/18. Patient was refer to cardiology and paediatrics department for evaluation and is being kept on regular follow up.

CASE REPORT 2

A 10 Years old female patient presents with diminution of vision in since 1 years . On examination VA was 6/24 both eye with pin hole no improvement. On slit lamp examination



SUBLUXATION (SUPERO-NASAL) HIGH ARCH PALATE BOTH EYE

there was supero-nasal subluxation of lens with intact zonules infero-temporally. Intraocular pressure in both eye was 17.3 mm of Hg. On gonioscopy angle were normal. Fundus examination was within normal limit. Other manifestations favouring Marfan Syndrome were long and slender finger, arm span greater than height and high arch palate were present

This patient was given refractive correction with -1.75 sph -1.5 cyl at 180 degree. After correction of refractive error VA was 6/12

LONG & SLENDER FINGERS ARM SPAN > HEIGHT

. Patient refer to cardiology and paediatrics department for evaluation. Patient is kept on regular follow up

DISCUSSION

Ocular complication are commonly found in marfan syndrome. These are sublaxtion of lens usually in supero-temporal but may be in any meridian ,angle anomaly as dysgenesis ,elongation of eye ball which leads to myopia ,astigmatism which is irregular,glaucoma,retinal detechment ,lattice degeneration ,cornea plana and strabismus. Patient need regular follow up to minimize the degree of visual loss due to lens dislocation and retinal detechment. Our patients showed satisfactory improvement with refractive correction but cases not improving with refraction patient can be taken up for lensectomy with anterior vitrectomy and scleral fixation IOL.In few cases CAPSULAR TENSION RING use for stabilization of capsular bag. Lastly patient are referred to cardiologic evaluation in order to decreae morbidity and mortality related to cardiovascular complication(aortic aneurysm an mitral insufficiency).

CONCLUSION

Marfan syndrome occur in 8-10 per 100,000 population per year.Ectopia lentis is the most comman ocular manifestations in about 70%-80% of cases. As many patients present first to the Ophthalmologist with visual impairment before presenting to physicians, Ophthalmologist can play an important role in diagnosis of Marfan Syndrome.

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