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Ectopia Lentis

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CLINICAL HISTORY:

52 year old male presented with complaints of decreased vision and glare in both eyes since childhood which has worsened over past 6 months

No h/o pain, redness, watering, photophobia or haloes around light. He has no complaints of trauma to eye and no complaints of hearing difficulty.

Past Ocular History : Evaluated in other centers and was made aware of the disease and was using glasses since college days.

- History of using spectacles since 30 yrs:-
- Glasses last changed 6 months back
- Both eyes – +3.00 spherical – 6/18 add +2.50 N-12

General Medicine and surgical history:

- Marfan's syndrome with aortic regurgitation
- Oral beta blockers – Toprol XL 50mg BD daily (Metaprolol)

Family history:

- No known family members with Marfan's syndrome
- His 21 yr. old son has Marfanoid features

EXAMINATION AND INVESTIGATIONS:

OCULAR EXAMINATION:

1)Visual acuity	*OD*	*OS*
Unaided – Distant Near	6/60 N-36	6/60 N-36
With Aid	6/24	6/18
With Pinhole	Improvement 6/12p	Improvement 6/12p
2)Colour vision	Normal	Normal
3)Field of vision	Normal	Normal
4)Hirschberg's Reflex	15 degree on to nasal side (mild exotropia)	Central
5)Pupillary reaction	Brisk	Brisk
6)Ocular motility	Full in all gazes	Full in all gazes

SLIT LAMP Examination:

	OD	*OS*
1)Eye lids and Eye lashes	Normal	Normal
2)Conjunctiva	Normal	Normal
3) Cornea	Normal	Normal

4)Anterior Chamber	Deep	Deep
5)Iris	Iridodonesis	Iridodonesis
6) Pupil	3 mm , Briskly reacting	3 mm , Briskly reacting
7)Crystalline lens	Lenticualar opacification and infero-temporal subluxation seen	Lenticualar opacification and infero-temporal subluxation seen
8) IOP	16 mm Hg	16 mm Hg

POSTERIOR SEGMENT EXAMINATION :

	OD	*OS*
1)Media	Clear	Clear
2) Optic disc	Normal with C:D- 0.3	Normal with C:D- 0.3
3) Vascular arcades	Normal	Normal
4)Peripheral retina	Normal	Normal
5)Macula	Foveal reflex present Myopic fundus seen	Foveal reflex present Myopic fundus seen

MUSCULO-SKELETAL EXAMINATION :

Tall & thin stature

Disproportionately long limbs.

Arm span > height : 190.5 cm > 189.5 cm

Cardio-vascular system

Pulse: 76/min.

BP: 130/80 mmHg

S_1 & S_2 – Audible

Murmur – EDM in left 3rd ICS

Apex beat – palpable in left 6th ICS

ECG : Tall QRS Complex in leads V4, V5 , V6

Urine for Sodium Nitroprusside test : Negative

Urine for Homocysteine level : 6 µmol/g creatine

Blood for Homocysteine level : 10µmol/L

RBS : 104 mg/dL

Hbs Ag (Card method) : Negative

HIV (Card method) : Negative

FINAL DIAGNOSIS:

DIFFERENTIAL DIAGNOSIS :

Homocystinuria

Weill-Marchesani syndrome

Final Diagnosis: ECTOPIA LENTIS DUE TO MARFAN SYNDROME

DISCUSSION:

A proper estimation of the refractive error through both the phakic and aphakic portions has to be done.

Check the acceptance in dim light and daylight conditions to yield a better visual outcome

Await Vitreoretinal Surgeon opinion for Barrage laser before planned cataract surgery

Planned surgery :- Pars plana lensectomy with anterior vitrectomy and Retro-fixation IOL

Marfan syndrome (MFS) is a genetic disorder of the connective tissue . MFS is an autosomal dominant disorder. About 75% of the time, the condition is inherited from a parent, while 25% of the time it is a de novo mutation. About 1 in 5,000 to 10,000 individuals have Marfan syndrome. It occurs equally in males and females and equally present across different regions and different races.4

Marfan syndrome is caused by mutations in the FBN1 gene on chromosome15 which encodes fibrillin-1, a glycoprotein component of the extracellular matrix. Fibrillin-1 is essential for the proper formation of the extracellular matrix, including the biogenesis and maintenance of elastic fibers.4

The extracellular matrix is critical for both the structural integrity of connective tissue, but also serves as a reservoir for growth factors. Elastic fibers are found throughout the body, but are particularly abundant in the aorta, ligaments and the ciliary zonules of the eye; consequently, these areas are among the worst affected.

Diagnostic criteria of MFS were agreed upon internationally in 1996. A diagnosis is based on family history and a combination of major and minor indicators of the disorder, rare in the general population, that occur in one individual – for example: four skeletal signs with one or

more signs in another body system such as ocular and cardiovascular in one individual – REVISED GHENT CRITERIA (2010)4

Marfan Syndrome can involve skeletal system , eyes , cardiovascualar system , lungs , nervous system .

In eyes usually there is weakness in the ciliary zonules, the connective tissue strands which suspend the lens within the eye. The mutations responsible for Marfan syndrome weaken the zonules and cause them to stretch. The inferior zonules are most frequently stretched resulting in the lens shifting upwards and outwards .1

Management of Ectopia lentis :- Pars plana lensectomy with anterior vitrectomy and Retro-fixation IOL1

There is no cure for Marfan syndrome . Management often includes the use of beta blockers such as propranolol or if not tolerated calcium channel blockers or ACE inhibitors4 . If the dilation of the aorta progresses to a significant-diameter aneurysm , causes a dissection or a rupture, or leads to failure of the aortic or other valve, then surgery (possibly a composite aortic valve graft or valve-sparing aortic root replacement) becomes necessary4.However the life expectancy has increased significantly over the last few decades and is now similar to that of the average person.

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