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Atypical unilateral subluxation of lens in Marfan Syndrome- A case report

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Abstract : A 12 year old male presented with complaints of visual impairment in the right eye for the past 2 years. Features suggestive of Marfan Syndrome such as high archd palate, funnel shaped chest, scoliosis, wrist sign, thumbs sign, arachnodactyly and pes planus were noted. Urine was negative for metabolic disorders. Cardiac examination was normal. Right eye showed infero-temporal lens subluxation with zonular dehiscence. The left eye lens was normal.
Keyword : Marfan Syndrome, lens subluxation, unilateral

A 12 year old male, first born of non-consanguineous marriage was referred for visual impairment in his right eye for the past 2 years. He was a full term infant with low birth weight. His postnatal and developmental history was normal.His father was 28 years and his mother was 22 years of age at the time of his birth. His father had features suggestive of Marfan Syndrome. On examination his vitals were stable. Features suggestive of Marfan Syndrome such as high arched palate, funnel shaped chest, scoliosis, wrist sign, thumbs sign, arachnodactyly and pes planus were noted .He weighed 27kg. His height was 148cm, with the upper segment measuring 61cm and his lower segment measuring 87cm. His arm span was 154cm. His arm-span was more than his height and he had decreased upper segment to lower segment ratio. Cardiovascular system examination was normal. Apical impulse was noted in the left 5th inter-costal space in the midclavicular line. No murmurs

were noted.
Investigations were done. ECG and Echo were within normal limits.Urine for metabolic screening was negative. Examination of right eye revealed zonular dehiscence between the 6 -7`O clock position with infero-temporal subluxation of the ens. The left eye lens was in the normal position with intact zonules.

Treatment: He was advised removal of lens in the right eye with scleral fixation of the intra ocular lens. He was advised regular follow up since many complications in Marfan Syndrome are age and maturation dependant.
Discussion: Marfan Syndrome is an autosomal dominant connective tissue disorder caused by mutations the gene FBN-1. The gene that encodes fibrillin-1 is present in the long arm of chromosome 15. Fibrillin a glycoprotein is the main constituent of the micro-fibrills of the extracellular matrix. Marfan syndrome is associated with ocular, cardiovascular, skeletal, pulmonary, cutaneous and neurological abnormalities.
Diagnostic criteria according to Ghent Nosology

Criterion	Major	Minor
Skin Involvement	None indicated	Striae atrophicae Rec. or incisional hernias 1 of 2 minor
Dura Involvement	Lumbosacral dural ectasia (CT/MRI) Dural ectasia present	None indicated
Family Involvement	First degree family member independently fulfilling diagnostic criteria Mutation FBN1 known to cause Marfan Syndrome	None indicated

The Ghent criteria are used to recognize Marfan syndrome. At least 2 major criteria and 1 minor criteria or 1major and 1minor in addition to a positive family history are needed to diagnose Marfan syndrome. The diagnosis of Marfan syndrome is dependent on clinical criteria, ECHO, MRI, slit-lamp examination and a negative urine cyanide nitroprusside test. Progressive cardiovascular defects such as aortic root dilatation and mitral valve prolapse contribute to the morbidity suffered by children with Marfan Syndrome. The connective tissue defect contributes to increased distensibility of the lung parenchyma and dura causing spontaneous pneumothorax and dural ectasia. Ectopia lentis is the most common ocular abnormality noted in Marfan syndrome and is seen in 50-80% of affected individuals. The ectopia lentis is usually bilateral and symmetrical. Subluxation is most frequently in the supero temporal quadrant. Abnormal production, distribution and attachment of the fibrillin rich zonules as well as their increased susceptibility to proteolytic cleavage result in the development of ectopia lentis. Therapy aims at prevention of complications. Annual evaluation is essential to monitor for potential problems such as cardiovascular disease, scoliosis and ophthalmological problems.

Conclusion: This case is being reported for the atypical type of lens subluxation viz, unilateral inferotemporal subluxation. Follow up is needed of the second eye for subluxation.

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