Subluxation of Lens in Marfan Syndrome

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A 13-year old boy presented with loss of vision in both the eyes. On examination the boy had long thin limbs, arm span (175 cm) exceeded his height (165 cm), ratio being 1.06:1. The lower segment of his body (87.5 cm) exceeded the upper segment (75 cm), US: LS was 0.83. He had arachnodactyly of hands and the fingers were hyperextensible. Steinberg sign was positive i.e., the thumb could be adducted across the narrow palm. The wrist sign was also positive. The thoracic cage revealed pectus excavatum and scoliosis was present. Ocular manifestations included myopia and iridodonesis. Slit lamp examination revealed subluxation of lens in both eyes (***Fig.*1**) Fundus examination revealed bilateral retinal detachment . Systemic examination revealed no other abnormalities. Echocardiography revealed mitral regurgitation. The elder brother has almost similar abnormalities indicating the hereditary origin.

Marfan syndrome, caused by mutations in the fibrillin gene on chromosome 15, presents with abnormalities of the cardiovascular, musculoskeletal, and ocular systems. Nearsightedness and astigmatism are common, but farsightedness can also result. Subluxation of lens (ectopia lentis) in one or both eyes (in 80% of patients) also occurs. In Marfan syndrome the dislocation is usually superotemporal (in 75% of cases). Typically, the zonules that are visible are intact and unbroken, in contrast to the broken zonules seen in homocystinuria. Sometimes eye problems appear only after weakening of connective tissue has caused detachment of retina in the second and third decade of life. Early onset glaucoma can be another complication. Other systemic disorders associated with ectopia lentis include homocystinuria, where the lens is displaced inferonasally, Weill-Marchesani syndrome, where the lens is displaced downwards and forwards and the lens tends to be small and round. In ectopia lentis et pupillae, an autosomal recessive disorder, both the lens are displaced in opposite directions. Other causes of ectopia lentis are- Ehler-Danlos, Sturge-Weber, Crouzon and Klippel-Feil syndromes, oxycephaly and mandibulo-facial dysostosis.

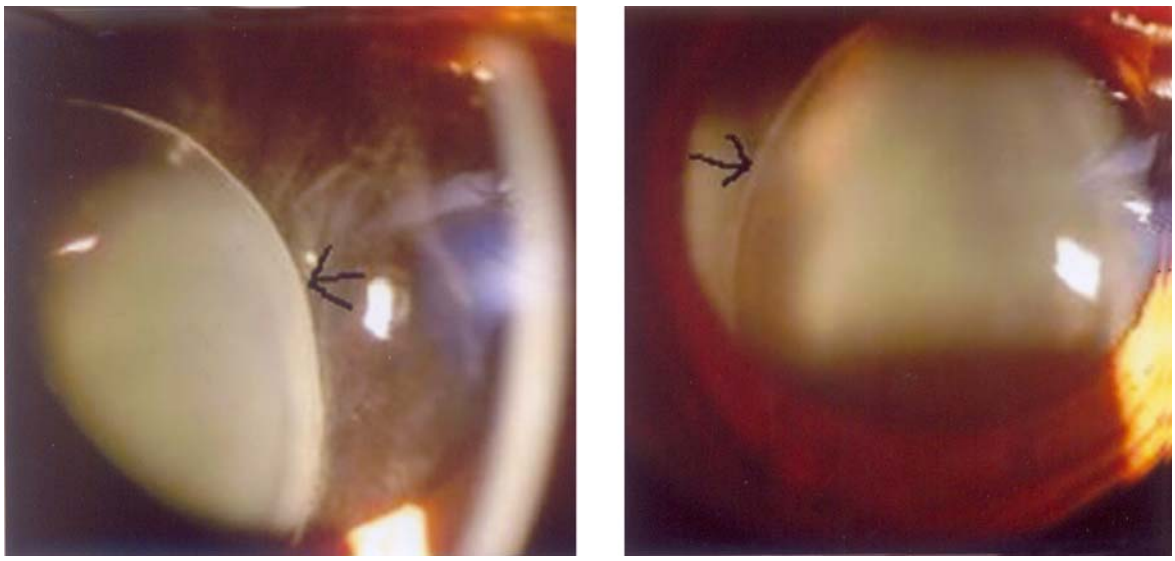

FIG. 1 *Subluxation of lens in both eyes.*