Bilateral lens subluxation in a patient with suspected Loeys-Dietz syndrome

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Quick Response Code:	Website: www.afrjpaedsurg.org
	DOI: 10.4103/0189-6725.79077

Sir,

We would like to highlight a case of a child seen at our out-patients clinic at P.C.E.A. Kikuyu Eye Unit, who presented with bilateral subluxation of the crystalline lenses, along with multiple dysmorphic features. We believe that this patient may have Loeys–Dietz syndrome (LDS), a rare genetic disorder similar in many ways to Marfan syndrome. Our patient may represent the first reported occurrence of ectopia lentis within LDS.

LDS was described in 2005 by Loeys and colleagues, making it a relatively recently recognized disorder caused by mutation in the genes *TGFßR1* or *TGFßR2*.[1] These genes encode two major isoforms of the transforming growth factor beta receptor. The disease is characterized by multiple craniofacial, cardiovascular, and skeletal abnormalities and also bears several similarities to the more common Marfan syndrome, such as arachnodactyly, arterial aneurysms, and palatal abnormalities. Unique findings in LDS include hypertelorism, craniosynostosis and bifid uvula (craniofacial symptoms), and fibrous contractures and bony deformity of the extremities (musculoskeletal abnormalities).

Ectopia lentis, although common in Marfan syndrome, has not been described in LDS and we report a case of a child with the LDS phenotype who presented with bilateral subluxation of the crystalline lens.

An 11-year-old Kenyan boy presented with a 2-year history of poor vision in both eyes. Ophthalmic examination revealed unaided Snellen visual acuities of 6/60 right and 4/60 left, with no relative afferent pupillary defect. Normal extraocular movements were associated with an intermittent small-angle exotropia. Bilateral inferonasal subluxation of the lens was present, with phacodonesis. The child underwent sequential bilateral lensectomy through limbal paracenteses, without complication, and achieved a corrected visual acuity of 6/6 right and 6/6 left. At all stages, the intraocular pressures were within normal range and the optic discs were normal.

Several craniofacial, skeletal, and cardiac abnormalities were also present: the child's facial shape was unusual, with hypertelorism (palpebral width 25 mm; intercanthal distance 33 mm; interpupillary distance 63 mm), low-set ears and some frontal bossing [Figure 1]. A high-arched palate was also present. Arachnodactyly and palmar muscle wasting was evident, as was marked fibrous contractures of the thumbs that could not be straightened [Figure 2]. There was also bony deformity of both feet [Figure 3] and the ratio of arm-span to height was increased (125 cm:120 cm; 104%). Although a fit young man with good exercise tolerance, a grade 2 systolic murmur – consistent with cardiac abnormality – was noted; further investigation was unavailable at the hospital.

Bilateral, symmetrical ectopia lentis is a common

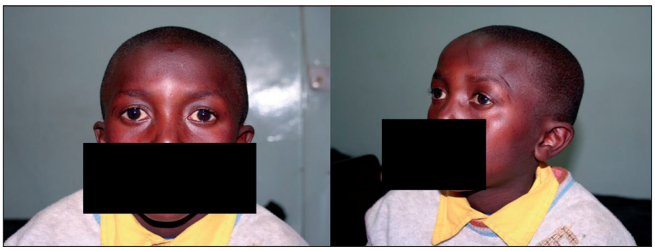


Figure 1: Craniofacial abnormalities. Hypertelorism is evident (left) as well as the cranial shape abnormality (right).



Figure 2: Abnormalities of the hands. Note muscle wasting, slight arachnodactyly, and fibrous contracture of the thumbs.



Figure 3: Extensive bony deformity of the feet.

sign of Marfan syndrome, occurring in up to 80% of cases.[2] Indeed, this sign – if present with high-arched palate, arachnodactyly, and an arm-span greater than the height – would be highly indicative of Marfan syndrome. Where additional features are present, such as craniofacial abnormalities (especially hypertelorism) or musculoskeletal abnormalities, a diagnosis of LDS should be considered. It is likely, therefore, that our patient had LDS and may represent the first reported occurrence of ectopia lentis within this rare syndrome.

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