Brittle Cornea Syndrome

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Clinical Image

A 6-year-old yellow Chinese girl was referred to People's hospital of Leshan, Department of Ophthalmology, in August 2017. The parents presented with progressive loss of vision and a bluish discoloration of the child's sclera. Family history was negative for known conspicuous eye disorders, no infections, or abnormalities in pregnancy or birth, and show no genetic disorders were known. General pediatric physical examination was normal for all systems. On ophthalmological examination showed that the corneas were obviously prominent, with a significant bluish discoloration of the sclera in both eyes (Figure 1). Pentacam HR anterior segment tomography revealed that keratoconus with a paracentral steepening in both eye with a maximum keratometric power of 54.10 D in the right eye and a maximum keratometric power of 54.40 D in the left eye. The thickness at the thinnest point evaluated by Pentacam was 324 μm with corneal astigmatism in topography (-2.6 D at 163 degrees) in the right eye and 313 μm with corneal astigmatism in topography (-2.7 D at 172 degrees) in the left eye. Unfortunately such molecular analyses were not performed in our case. However, she did not present bone fractures or deafness as in ontogenesis imperfect, nor did present skin or ligament hyper elastic changes as in Ehlers-Danlos syndrome or changes in stature similar to the Marfan syndrome. As far as we know, this is the first case of BCS reported in China or Asia.

Figure 1: Brittle cornea syndrome.