A Blue Eyed Girl with Hyperactivity: Look in to the Eyes and Heart!

A 3-year-old girl presented with difficulty in running and slowly progressive deformity of the back. On examination, she was hyperactive, had mild gross motor delay, normal head circumference, increased arm-span to height ratio (1.10), blue sclera, lenticular dislocation, downslanting palpebral fissure, malar hypoplasia (**Fig. 1**), pectus carinatum, pansystolic murmur, scoliosis, arachnodactyly (**Fig. 2**), joint hypermobility and pes planus. *X*-ray spine showed thoraco-lumbar scoliosis and 2D-echo was suggestive of aortic root dilatation and aortic regurgitation. Based on revised Ghent nosology, a diagnosis of Marfan syndrome was concluded.

Marfan syndrome (MS) is an autosomal dominant connective tissue disorder, due to mutation in gene encoding for Fibrillin-1. MS is multisystem disorder affecting eyes, skeleton, lung, skin, cardiovascular and nervous system. Every individual with MS do not have all the clinical manifestations of the condition. According to the revised Ghent criteria, aortic root aneurysm and ectopia lentis are the cardinal features and is sufficient for the diagnosis of MS even in the absence of family history. Central nervous system features are dural ectasia, lumbo-sacral meningocele, enlarged cisterna magna, hyperactivity and specific learning disability. The differential diagnosis of blue sclera includes Ehler Danlos syndrome, Van der Heave syndrome, Osteogenesis imperfecta, Hallerman Strieff syndrome, Incontinentia pigmenti, brittle cornea syndrome, and rarely with iron deficiency anaemia. Differential diagnosis of marfanoid phenotype is Lujan-Fryns syndrome, Shprintzen-Goldberg syndrome, Homocystinuria, Beals syndrome and MASS (*m*yopia, *m*itral valve prolapse, *a*ortic dilatation, *s*kin involvement, *s*keletal involvement) syndrome. Management of the patients with MS is complex and requires a multidisciplinary approach with regular cardiac and ocular surveillance.

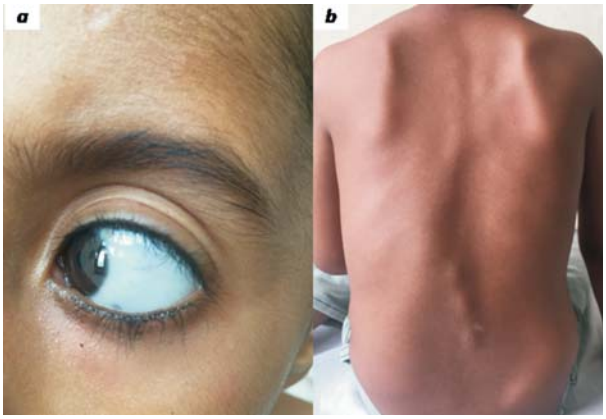

FIG. 1 *Clinical photographs of the child showing blue sclera, downslanting palpebral fissure, hypoplastic maxilla and supraorbital ridge (a), and thoraco-lumbar scoliosis (b).*

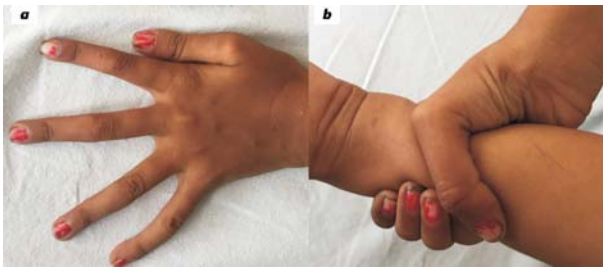

FIG. 2 *Photographs of the hand showing arachnodactyly (a), and thumb sign (b).*

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Neonatal Lupus with Extensive Cutaneous Involvement

A 2-month-old girl presented with multiple hypo-pigmented and depressed lesions over face, scalp and erythematous lesions over forehead, cheeks and around eyes, since birth. The mother was asymptomatic at the time of presentation with no history of drug intake during pregnancy, preeclampsia, photosensitivity or recurrent oral ulcers. On cutaneous examination, multiple depressed, atrophic and hypopigmented plaques were present on face involving forehead, cheeks, periorbital area and scalp (***Fig.* 1**), anterior abdomen and back. Investigations revealed anemia