

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, lumbosacral 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 (*myopia, mitral valve prolapse, aortic dilatation, skin involvement,*

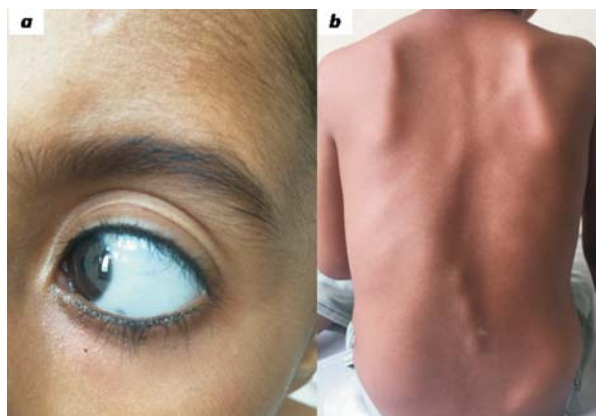


FIG. 1 Clinical photographs of the child showing blue sclera, downslanting palpebral fissure, hypoplastic maxilla and supraorbital ridge (a), and thoraco-lumbar scoliosis (b).

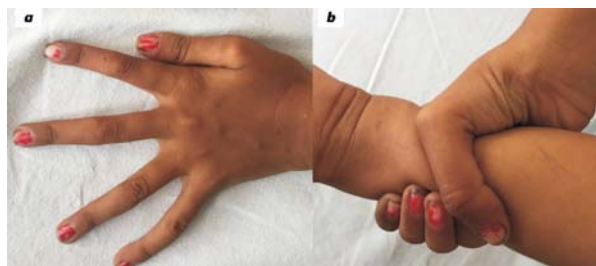


FIG. 2 Photographs of the hand showing arachnodactyly (a), and thumb sign (b).

skeletal involvement) syndrome. Management of the patients with MS is complex and requires a multidisciplinary approach with regular cardiac and ocular surveillance.

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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

(hemoglobin 8.9 g/dL), thrombocytopenia (platelet count, $90 \times 10^9/L$) and raised liver enzymes. Antinuclear antibody (ANA) test was positive (2+); speckled pattern and anti-Ro La was present in high titers in both mother and infant. VDRL of mother and baby was non-reactive. Ultrasound abdomen, electrocardiogram and echocardiography were normal. We diagnosed neonatal lupus erythematosus, and advised tacrolimus 0.03% ointment once daily with strict photo-protection. The patient showed marked improvement in the lesions in the next three months.

Neonatal lupus erythematosus (NLE) is a rare, often misdiagnosed immune-mediated disease. It is usually associated with maternal antibodies to proteins Ro/ La (SSA/SSB) which are transferred by the placenta. Differential diagnoses of NLE include tinea corporis (centrifugal skin lesions with more inflammation at periphery), seborrheic dermatitis (scaly and yellowish round or annular pattern of lesions), congenital syphilis (positive VDRL test), annular erythema of infancy, and aplasia cutis (absence of skin since birth). Congenital heart block and cardiomyopathy are cardiac manifestations of NLE. Management of cutaneous lesions include photo-protection and low-potency topical corticosteroids. Systemic corticosteroids, intravenous immunoglobulins, and immunosuppressive agents have also been tried.



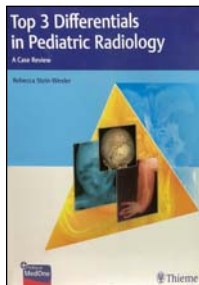
Fig. 1 Multiple depressed, atrophic and hypopigmented plaques on forehead, cheeks, periorbital area and scalp.

Majority of patients with NLE of the skin, liver or blood have transient disease that spontaneously resolves after 4-6 months, but can have substantial morbidity and mortality if there is congenital heart block.

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BOOK REVIEW



Top 3 Differentials in Pediatric Radiology

REBECCA STEIN-WEXLER

Thieme Publisher, Germany.

Pages: 410; Price: Not mentioned.

Top 3 Differentials in Pediatric Radiology

by Rebecca Stein-Wexler is a comprehensive compilation of common and clinically relevant cases encountered in Pediatric Radiology practice. The book is divided into six sections based on imaging of different body systems. A short clinical history is provided with each radiological image to generate an appropriate clinical perspective, followed by description of the salient imaging findings.

The top three imaging differential diagnoses are well discussed, and along with imaging features that justify the diagnosis, a brief description of the classical clinical features and suggested management is also provided. This is followed by the final diagnosis of the case and the

important take away points from the discussion. The description of each case is concise, yet provides all essential and relevant information about the topic under consideration. One caveat is that 'arrows' or other appropriate pointers are often missing from the images and legends, which makes it difficult for reader to spot the radiological findings accurately.

Overall, this book is very well-written, informative, and includes all aspects of Pediatric Radiology in terms of the organ systems and radiological modalities (*i.e.* conventional X-rays, special X-ray investigations, ultrasound, doppler, Computed tomography and Magnetic resonance imaging), simultaneously incorporating cases of varying levels of difficulty. The book will be very beneficial for both Pediatricians and Pediatric Radiologists – residents, teachers as well as practitioners.

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