

Make up your knowledge: Kabuki make-up syndrome and pes planovalgus

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Kabuki make-up syndrome (KMS) is a rare syndrome with multiple congenital anomalies first described by Japanese researchers Kuroki and Niikawa in 1981.^[1] It is characterized by a typical facial appearance (reminiscent of the traditional Japanese Kabuki theater face make-up), musculoskeletal anomalies, cardiac malformation, mild-to-moderate mental retardation, and postnatal growth deficiency. The most frequently reported musculoskeletal findings are hip joint dislocation, fifth finger clinodactyly, absent palmar transverse crease, fingertip pads, and kyphoscoliosis.^[1] Although few, foot deformities such

as congenital talipes calcaneovalgus^[2] and congenital talipes equinovarus^[3] have been also reported in the literature. Drawing attention of Physical Medicine and Rehabilitation (PMR) physicians to this syndrome; we, herein, report an additional foot deformity, i.e., pes planovalgus in KMS.

A 13-year-old girl with known KMS presented to our PMR outpatient clinic for gait/posture problems. She had intellectual disability, bilateral hearing loss and dental problems, as well. Physical examination revealed typical facial appearance, hirsutism, spine asymmetry, and feet deformities (Figure 1). When

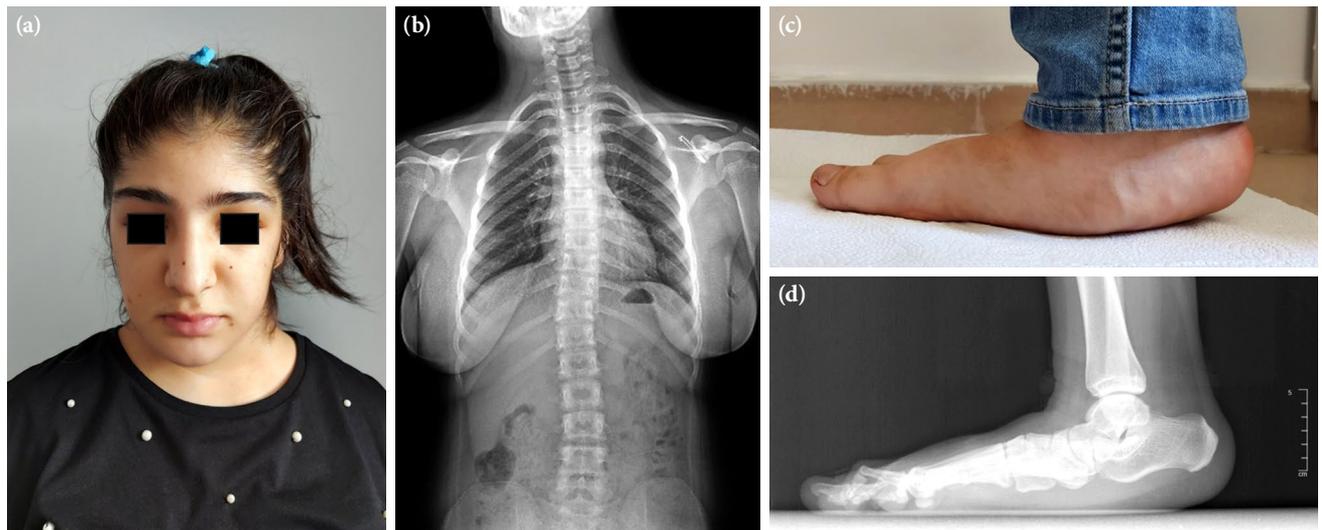


Figure 1. (a) Photograph of the patient showing typical facial appearance of Kabuki make-up syndrome. (b) Mild left cervicothoracic, right thoracolumbar, and left lumbar scoliosis seen in anteroposterior radiograph. (c) Photograph and (d) lateral radiograph showing pes planovalgus deformity in right foot.

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the patient was asked to walk on the toes, feet deformities were observed to improve, suggesting a flexible deformity. Radiographs were confirmatory without additional problems (e.g., tarsal coalition) either (Figure 1). As there were significant, but flexible bilateral pes planovalgus deformities, the patient was prescribed personalized insoles with medial arch support in addition to home-based exercises and close follow-up for scoliosis.

Kabuki make-up syndrome is a recognized clinical problem with various musculoskeletal abnormalities such as hip dislocation and scoliosis. Although various associated foot deformities have been reported in the literature, to the best of our knowledge, pes planovalgus has not been reported before. In this context, the purpose of our report was two-fold. First, we aimed to call attention of PMR physicians to this syndrome which is not commonplace in routine/daily practice. Second, highlighting the relevant musculoskeletal findings of the syndrome, we also attempted to demonstrate that

additional involvement patterns might be likely. As such, prompt assessment is noteworthy and the management needs to be tailored accordingly.

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