Kabuki Make-Up Syndrome with Bilateral Dislocation of the Hip

Aghoutane Elmouhtadi*, MD; Fezzazi Redouane, MD
Department of Orthopedic Pediatric Surgery, Medical University Hospital Mohammed VI, Faculty of Medicine and Pharmacy, Marrakech, Morocco

Received: Oct 19, 2011; Accepted: Apr 08, 2012; First Online Available: Jan 17, 2013

Kabuki make-up syndrome (KMS) is a rare multiple congenital anomalies/mental retardation syndrome of unknown cause, first described independently by Niikawa and Kuroki. It is characterized by distinctive facial features, skeletal anomalies, dermatoglyphic abnormalities, short stature, and mental retardation. A group of abnormalities with involvement of other organs can lead to diagnosis of KMS.

The authors report a 4-and-half-year old girl with KMS, admitted for bilateral dislocation of the hip. On this case, we try to explain the syndrome and to allow pediatricians and other physicians to become familiar with this syndrome.

KMS (OMIM#147920) is a rare dysmorphic syndrome that was first described in 1981 by Niikawa et al[1] and Kuroki et al[2]. The name of the syndrome, Kabuki make-up, refers to resemblance of the facial traits of KMS patients to the make-up of actors in Kabuki, the traditional Japanese theatre[1,2].

KMS is more common in Japan, although it has been reported from different geographic regions in the world[3]. Only few hundred cases have been described, and it is expressed as 1/32000 incidence in Japan[4]. We recently observed a 4 and half year old girl with typical KMS presented with bilateral dislocation of the hip.

The patient was referred to pediatric orthopedic department for bilateral dislocation of the hip. Her parents were healthy with no consanguinity.

Psychomotor development was delayed with speech defects. On physical examination at the age of 4 years, the patient’s height was 92 cm, weight 15 kg. She had characteristic facial dysmorphism with high arched eyebrows that were sparse in the lateral part, long palpebral fissures, prominent and protruding ears and lip pits on the lower lip, depressed nasal tip (Fig. 1A). She also had joint laxity, brachydactyly V (Fig. 1B) and finger tip pads (Fig. 1C).

Other findings were normal. Chest radiography showed kyphotic deformity of the spine. Echocardiography was normal. Abdomen and pelvic sonography showed pelvic situation of the right kidney. Pelvic radiography showed bilateral dislocation of the hip (Fig. 2). She was diagnosed as Kabuki make-up syndrome due to clinical findings.

She was operated for bilateral dislocation of the hip with, in the first, the right hip by open reduction, iliopsoas and adductor releases, capsuloraphy and Salter innominate osteotomy, and the same technique in the left hip with good results.

KMS occurs mostly sporadic, although some familial cases have been reported[6]. Inheritance

![Fig. 1: A) Characteristic facial dysmorphism with high arched eyebrows that were sparse in the lateral part, long palpebral fissures, prominent and protruding ears and lip pits on the lower lip, depressed nasal tip. B) Brachydactyly V and C) finger tip pads in patient with Kabuki make-up syndrome.](image-url)
Structural heart defects are encountered in 32%-58% of children, and non specific congenital heart defect predominates[7,9].

Neonatal hypotonia and feeding difficulties, recurrent infections, reno-urinary malformations, diaphragmatic anomalies, neurological symptoms, abnormalities of the central nervous system, obesity, and precocious puberty are also reported in this syndrome[9,10].

**Key words:** Kabuki Make-Up Syndrome; Children; Dislocation of the hip

The high risk of dislocation of the hip has been attributed to the generalized joint laxity that was observed in 10 of 16 cases[8,9].

References

1. Nikawa N, Matsuura N, Fukushima Y, et al. Kabuki make up syndrome: a syndrome of mental retardation, unusual facies, large and protruding ears, and post natal growth deficiency. *J Pediatr* 1981; 99(4):565-9.

2. Kuroki Y, Suzuki Y, Chyo H, et al. A new malformation syndrome of long palpebral fissures, large ears, depressed nasal tip, and skeletal anomalies associated with postnatal dwarfism and mental retardation. *J Pediatr* 1981; 99(4):570-3.

3. Muluk NB, Yalcinkaya F, Budak B, et al. Evaluation for language and speech development in Kabuki make-up syndrome. A case report. *Int J Pediatr Otorhinolaryngol* 2009; 73(12): 1837-40.

4. Tekin M, Fitoz S, Arici S, et al. Niikawa Kuroki (Kabuki) syndrome with congenital sensorineural deafness: Evidence for a wide spectrum of inner ear abnormalities. *Int J Pediatr Otorhinolaryngol* 2006; 70(Suppl 5): 885-9.

5. Vaccaro M, Salpietro DC, Briuglia S, et al. Cutis laxa in Kabuki make-up syndrome. *J Am Acad Dermatol* 2005; 53(Suppl 1): 24:0-51.

6. Courtens W, Rassart A, Stene JJ, et al. Further evidence for autosomal dominant inheritance and ectodermal abnormalities in Kabuki syndrome. *Am J Med Genet* 2000; 93(3): 244-49.

7. Niikawa N, Kuroki Y, Kajii T, et al. Kabuki make-up syndrome: a study of 62 patients. *Am J Med Genet* 1988; 31(3): 565-89.

8. Kawame H, Hannibal C, Hudgins L, et al. Phenotypic spectrum and management issues in Kabuki syndrome. *J Pediatr* 1999; 134(4): 480-8.

9. Santiago J, Muszlak M, Goulois E, et al. A case of Kabuki syndrome admitted for acute diarrhea and growth retardation in a French hospital in tropical area. *Arch Pédiatrie* 2010; 17(5): 588-93. [In French]

10. Matsumoto N, Niikawa N. Kabuki make-up syndrome: a review. *Am J Med Genet* 2003; 117C(1): 57-65.