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CASE REPORT

# Kabuki make-up syndrome with genitourinary anomalies, ophthalmologic features and hyperpigmentation in an Egyptian child

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**KEYWORDS**

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**Abstract** We report a 3.5 year old male child, first in order of birth of healthy consanguineous Egyptian parents with typical characteristics of Kabuki make-up syndrome. The patient had microcephaly, high arched sparse eyebrows, hypertelorism, long palpebral fissures with eversion of the lateral third of the lower eyelids, bilateral ptosis, long eyelashes, blue sclera, depressed nasal bridge, broad nose with everted nares, and low set small deformed ears, thin lips, low post hair line, short neck, persistent fingertip pads, dysplastic nails, hypermobile joints, pigmented nevus on the back, lateral side of right foot and right leg and mild hypertrichosis over the lower back. Our patient had also a non-functioning left kidney, multiple chalazions in upper eyelids, enlargement of the glans penis, which were not reported previously, and moderate mental retardation.


## 1. Introduction

Kabuki make-up syndrome (KMS) is a congenital mental retardation syndrome characterized by typical facial features including: long palpebral fissures with eversion of the lateral third of the lower eyelids, arched eyebrows with sparse outer half, prominent eye lashes, broad and depressed nasal tip, large prominent earlobes, a cleft high-arched palate, micrognathia, low posterior hair line, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy [1].

Additional features include short stature, internal malformations (involving the heart, genitourinary and gastrointestinal systems) and immunological defects [2].

Kabuki make-up syndrome was first described in Japanese children in 1981 [3,4]. The estimated prevalence of KMS in Japan was 1/32,000 people with almost equal sex distribution. Although it was initially considered as a disease affecting exclusively the Japanese population, several reports support a widespread ethnic distribution of KMS [5].

We report a case with the typical features of Kabuki make-up syndrome who has in addition some unreported features after taking consent of the parents.

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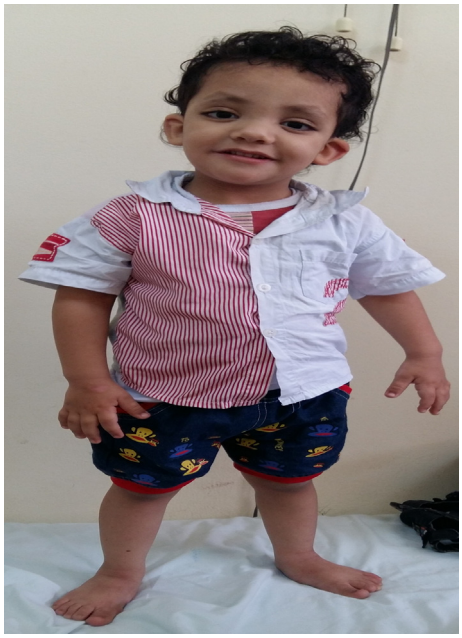

**Figure 1** Photo of the patient.

## 2. Case report

A 3.5 year old male child, first in order of birth of healthy consanguineous Egyptian parents. The patient was delivered at full term by cesarean section. His birth weight was 3 kg. No problems were noted during pregnancy. The patient was referred to the Genetics Clinic, Pediatric Hospital, Ain Shams University complaining of developmental delay and abnormal features.

The patient was born with undescended testes and had repair surgery at the age of 1 year. At the age of 1.5 year, he had multiple chalazions in upper lids of both eyes which resolved spontaneously leaving small scars. Family history was unremarkable. Both parents were normal.

On examination, the patient had mental retardation, his weight was 13.5 kg (5th percentile), his height was 90 cm (5th percentile), and his skull circumference was 47 cm (3rd percentile).

The patient had microcephaly, high arched eyebrows with sparse outer half, hypertelorism, long palpebral fissures with eversion of the lateral third of the lower eyelids, bilateral ptosis, long and prominent eyelashes, blue sclera, depressed nasal bridge, broad nose with everted nares, low set small deformed ears, thin lips, high-arched palate, micrognathia, low post hair line and short neck (Figs. 1 and 2).

The patient had also persistent fingertip pads, dysplastic nails (Fig. 3), hypermobile joints (Fig. 4), overlapping of the second toes over third toes, short second toe of the left foot, pigmented nevus on the back, lateral side of the right foot and front of the right leg (Fig. 5). He also had mild hypertrichosis over the lower back.

Abdominal, cardiac and neurologic examinations were normal. The genitals demonstrated enlargement of the glans penis (Fig. 6). IQ was 48.

Extended metabolic screen, serum lactate and serum ammonium were normal.

Karyotype was also normal. Fundes examination, audiometry and EEG were normal.

ECHO cardiography was normal. Abdomino-pelvic ultrasonography revealed non-visualized left kidney. Renal isotope

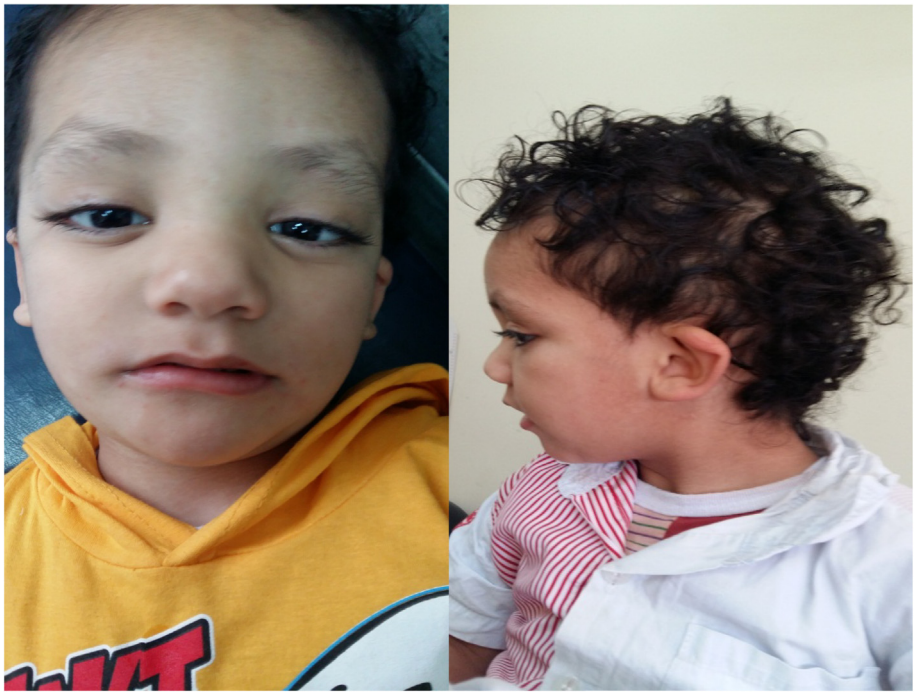

**Figure 2** Microcephaly, high arched sparse eyebrows, hypertelorism, long palpebral fissures, bilateral ptosis, long eyelashes, blue sclera, depressed nasal bridge, broad nose with everted nares, low set small deformed ears, thin lips, low post hair line and short neck.