

## SHORT COMMUNICATION

### BRIEF CLINICAL REPORT: THE KABUKI MAKE-UP SYNDROME IN A BRAZILIAN BOY

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#### ABSTRACT

The Kabuki make-up Syndrome is a MCA/MR recurrent-pattern syndrome with a characteristic facial appearance. So far it is principally described in Japanese (58 out of 62 patients) (Niikawa *et al.*, *Am. J. Med. Genet.* 31: 565-589, 1988). Here we report on a rather typical case in a Brazilian boy of non-Japanese extraction.

#### INTRODUCTION

The Kabuki Make-up syndrome, is MCA/MR syndrome, originally described by Nikawa *et al.* (1981) and up to now all reported cases have been sporadic. Main signs include mental retardation, postnatal short stature, a characteristic facial appearance with long palpebral fissures, eversion of the lateral third of the lower eyelids, arched eyebrows, broad and depressed nasal tip, large prominent ears, short fifth fingers and abnormal dermatoglyphics. A number of other anomalies are also frequently found in this syndrome, and most of the patients described are Japanese. Here we report on a Brazilian child of non-Japanese extraction, presenting the clinical features of the Kabuki make-up syndrome.

## CASE REPORT

ASR (Figures 1A,B), the propositus, is the first child of a 19-year-old G2P2 mother and her 25-year-old unrelated husband. The younger sister and other relatives are normal. Parental ancestors are of Brazilian extraction for several generations. The pregnancy was uneventful. There was no exposure to toxic agents or to X-rays and no trauma was reported. Delivery was normal at 38 weeks of gestation.

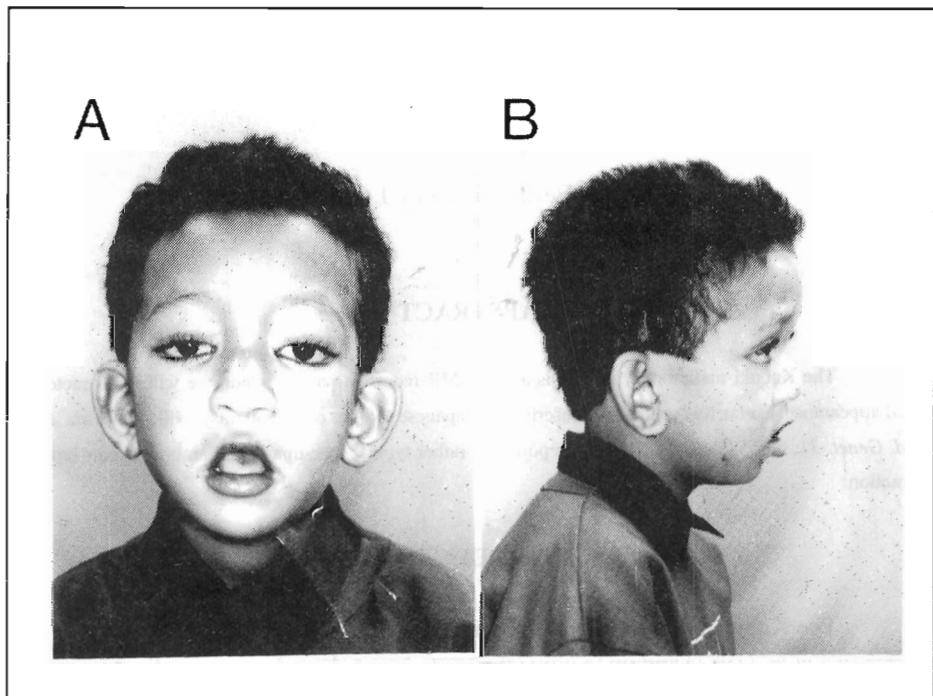


Figure 1 - A, Frontal view of the propositus; B, Lateral view of the propositus.

Birth weight was 1600 g (< 3rd centile), TBL (total body length) and OFC (occipitofrontal circumference) were not recorded. The child was anemic and hypotonic, and only seven days later cleft palate was noticed. He presented recurrent ear, throat and pulmonary infections during early infancy. Neuropsychological development was delayed.

Examination at the age of 8 1/2 years showed: weight of 17.2 kg (< 3rd centile), length of 108 cm (< 3rd centile), OFC 50.5 CM (3rd centile), ICD of 2.8 cm (50th centile),

OCD of 9.5 cm (97th centile). The boy presented a flat occiput, prominent forehead, low-set, large and prominent ears with hypoplastic anti-helix, long eyelashes, arched eyebrows, sparse laterally, large palpebral fissures with eversion of the lateral third of the inferior eyelids, bilateral epicanthus, shallow orbits, large nasal bridge, broad depressed nasal tip with a short septum, mild retrognathia, surgically repaired cleft palate, small mouth with downturned corners and eversion of the lower lip, widely spaced hypoplastic teeth, brachydactyly (Figure 2), hyperextensibility of the fingers and mental retardation. Dermatoglyphic study showed an excess of ulnar loops, but no hypothenar loops nor absence of a *c* or *d* digital triradius.

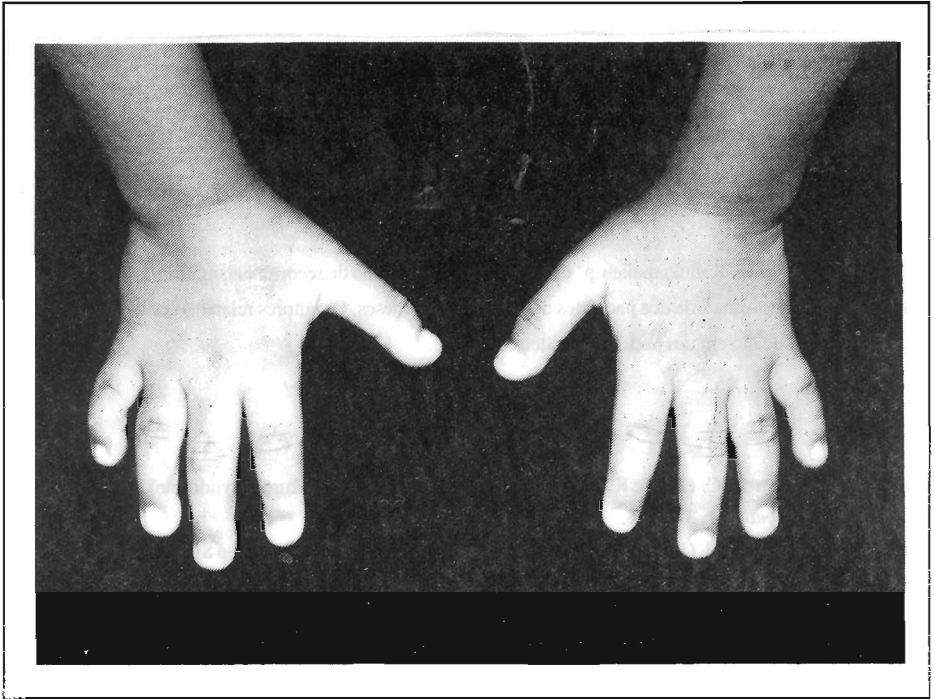


Figure 2 - Clinical aspects of the hands of the propositus.

## DISCUSSION

The Kabuki Make-Up syndrome has been so called due to the resemblance of these patients to the traditional make-up of the Kabuki actors. The cause of KMS is unknown, the sex-ratio is even, there is no correlation with birth order, all reported

patients up to now have been sporadic, and never have been found to reproduce (Niikawa *et al.*, 1988; Gorlin *et al.*, 1990). Autosomal dominant inheritance has been proposed, with every patient representing a new mutation, with a mutation rate of  $15.6 \times 10^{-6}$  per gamete per generation (Kuroki *et al.*, 1981; Niikawa *et al.*, 1988; McKusick, 1990). Similar to the patients previously described with this condition (Niikawa *et al.*, 1981; Kuroki *et al.*, 1981; Braun and Schmid, 1986; Kaiser-Kupfer *et al.*, 1986; Pagon *et al.*, 1986; Sheikh *et al.*, 1986; Niikawa *et al.*, 1988; Benito and Ferreti, 1989; Halal *et al.*, 1989; Mulvihill and Kaiser-Kupfer, 1989), our patient presented the characteristic facial appearance, the brachymesophalangy V, unusual dermatoglyphic patterns with an excess of ulnar loops, mental and growth retardation. To our knowledge this is the first report of this condition in Brazilian patients.

### ACKNOWLEDGMENTS

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### RESUMO

A síndrome "Kabuki make-up" é uma síndrome de padrão de recorrência, malformativa e que cursa com retardo mental. A maioria dos pacientes descritos são japoneses. Os autores relatam o primeiro caso desta síndrome diagnosticado em um paciente brasileiro.

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