

## A case of Kabuki syndrome with immunodeficiency and symphalangism

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### Introduction

A new style of dance drama prevalent in the Kyoto province in Japan in the 16<sup>th</sup> century was called "Kabuki"<sup>1</sup>. The facial make up used by the artist to draw the attention of the audience was unique and resembles a genetic disorder called Kabuki syndrome. We report a case of similar nature in a 10 years old female with immunodeficiency and symphalangism.

### Case report

A 10 year old girl (Figure 1) was admitted to hospital with three episodes of urinary tract infection (UTI) over the last two years. She had recurrent chestiness and otitis media from birth.



**Figure 1: 10 year old girl**

\*Permission given by parents to publish photograph

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Detailed evaluation of the urinary tract with ultrasonography of kidney, ureter and bladder failed to identify any pathology. Each time she had neutrophil leucocytosis with high acute phase reactants, but during her last admission two months back serum immunoglobulin (IgA) was 15mg/dl which was significantly low (normal range 30-60 mg/dl).

Her body weight was less than the 3<sup>rd</sup> centile and the height was less than the 25<sup>th</sup> centile at 10 years of age. Examination of head and face revealed microcephaly, high arched eyebrow, broad nasal bridge, squint of right eye, mild eversion of left lower eye lid and large low set ears. Other significant finding in this case were clinodactyly of both little fingers (Figures 2), persistence of fetal finger pad with nail hypoplasia of all fingers more prominent in both thumbs (Figures 3), generalised hypotonia, progressive kyphoscoliosis and moderate specific learning disorder.



**Figure 2: Bilateral clinodactyly**



**Figure 3: Nail hypoplasia of thumbs**

X-rays of both hands (Figure 4) showed atretic middle phalanx of little fingers of both sides with fusion of proximal and middle phalanx (figures 5 and 6). This fusion of proximal interphalangeal joint is called symphalangism. Echocardiography did not show any structural heart problem apart from moderate pulmonary arterial hypertension.



**Figure 4: Atretic middle phalanx of little fingers of both sides**



**Figure 5: Symphalangism of left proximal interphalangeal joint**



**Figure 6: Symphalangism of right proximal interphalangeal joint**

## Discussion

Facial resemblance of 16<sup>th</sup> century Japanese drama actors plus other features in different body parts is referred as Kabuki syndrome. Not all features are universally present in a particular case, some features evolving with time<sup>2</sup>. Persistence of fetal finger pad is present in 96% of reported cases<sup>2</sup>. Contrary to standard case described in Smith's recognisable pattern of human malformations our case does not have cardiac or genitourinary abnormality. Recurrent sino-pulmonary infection and UTI without any risk factors plus IgA deficiency proves humoral immunodeficiency which is a common feature of this syndrome.

The fusion of middle and proximal phalanx of fifth finger is not found in any of the reported cases and unique to our case. This symphalangism of the fifth toe 3 is commonly seen in 40 to 70% of general population at a later age as old-age ankylosis. But here occurrence in upper limb and its pathogenesis in Kabuki is unknown.

In most of the cases, the gene for kabuki syndrome KMT2D 4,5 is located on chromosome number 12 and a few cases another gene KDM6A 4,5 was isolated on X chromosome. The gene KMT2D runs as autosomal dominant manner. The variable expression of autosomal dominant genes could explain variable features as in our case.

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