LETTERS TO THE EDITOR

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Kabuki Syndrome and
Diaphragmatic Defect

Kabuki syndrome is a rare genetic disorder
with characteristic facial features. Other
common findings are mental retardation,
postnatal progressive growth retardation,
skeletal, cardiac and dermatoglyphics
abnormalities(1).

A child, 13 months old, sixth in birth order,
product of non consanguineous marriage,
presented with failure to thrive and
developmental delay. He 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.
(Fig.1). A detailed dermatoglyphic study
showed an absent digital triradius ‘c’ and ‘d’.
Detailed cardiovascular examination revealed
an ostium secundum atrial septal defect. The
CECT upper abdomen showed diaphragmatic
eventration on the right side with a mediastinal
shift. The X-ray of bilateral hands showed
clinodactyly. The abdominal ultrasound
examination was normal. Karyotype done on
peripheral lymphocytes was normal. A

Fig.1. Figure showing the distinctive facial features
of Kabuki syndrome.
diagnosis of Kabuki syndrome was made, based on the characteristic clinical features.

Most of the patients of Kabuki syndrome have five cardinal manifestations, namely, the characteristic facial features (100%), skeletal abnormalities (over 90%), postnatal growth deficiency (over 70%), mild mental retardation (over 90%) and dermatoglyphic abnormalities (over 95%). Our patient had all of the cardinal manifestations(2).

Digilio, et al. reported at least 58% of patients with Kabuki syndrome had congenital heart diseases and considered them as cardinal features(3). Renal, hepatobiliary(4) and diaphragmatic(5) anomalies are also reported in this syndrome. This is the first Indian report of Kabuki syndrome with a diaphragmatic abnormality.

Because Kabuki syndrome is not associated with severe medical complications, it is presumed that the prognosis for survival into adulthood is good.

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Snakebite Envenomation in India: A Rural Medical Emergency

Snakebite is a common medical emergency encountered among Indian population(1). According to World Health Organisation, 15,000 people of 2,00,000 bitten by snakes die every year in India(2). The number may be more owing to the lack of proper documentation and the uncounted deaths that occur before reaching the hospital. Most of the affected, including children are from rural areas. Data on snakebite envenomation among Indian children are limited.

A retrospective study was carried out in the Department of Pediatrics, Jawaharlal Institute of Post Graduate Medical Education and