

*Department of Pediatric Surgery,
Christian Medical College and Hospital,
Ludhiana 141 008, India.
E-mail: kamaleshpal@gmail.com
kamalesh_pal@yahoo.com*

REFERENCES

1. Collins DC. A study of 50,000 specimens of the human vermiform appendix. *Surg Gynecol Obstet* 1955; 101: 437-445.
2. Zaidenstein L, Freud E, Schwartz M, Zer M. Clinical presentation of rare appendico-omphalic anomalies. *J Pediatr Surg* 1995; 30: 1702-1703.
3. Crankson SJ, Ahmed GS, Palkar V. Patent omphalomesenteric duct of the vermiform appendix in a neonate: Congenital appendico-umbilical fistula. *Pediatr Surg Int* 1998; 14: 229-230.
4. Kadzombe E, Currie ABM. Neonatal fistula from the appendix to the umbilicus. *J Pediatr Surg* 1988; 23: 1059-1060.
5. Ucchedu A. Primary appendiculo-cutaneous fistula caused by appendicitis: Description of a case and review of the literature. *Ann Chir* 1984; 38: 216-219.
6. Jeffrey RB, Tolentino CS, Federle MP. Percutaneous drainage of periappendiceal abscess, review of 20 patients. *Am J Roentgenol* 1987; 149: 59-62.

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 manifestation, 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.

**Sidharth K. Sethi,
M.M.A. Faridi,**

*Department of Pediatrics,
University College of Medical Sciences and
GTB Hospital,
Shahdara, Delhi 110 095, India.*

Correspondence to:

Dr. Sidharth Kumar Sethi,

*CP-109, Pitampura, Maurya Enclave,
Delhi 110 088,
India.*

E-mail: sidsdoc@yahoo.com

REFERENCES

1. Niikawa N, Matsuura N, Fukushima Y, Ohsawa T, Kajii T. Kabuki make-up syndrome: A syndrome of mental retardation, unusual facies, large and protruding ears and postnatal growth deficiency. *J Pediatr* 1981; 99: 565-569.
2. Vaux KK, Hudgins L, Bird LM, Roeder E, Curry CJ, Jones M, *et al.* Neonatal phenotype in Kabuki syndrome. *Am J Med Genet* 2005; 132A: 244-247.
3. Digilio MC, Marino B, Toscano A, Giannotti A, Dallapiccola B. Congenital heart defects in Kabuki Syndrome. *Am J Med Genet* 2001; 100: 269-274.
4. Armstrong L, Abd El Moneim A, Aleck K, Aughton DJ, Baumann C, Braddock SR, *et al.* Further delineation of Kabuki syndrome in 48 well-defined new individuals. *Am J Med Genet* 2005; 132A: 265-272.
5. Donadio A, Garavelli L, Banchini G, Neri G. Kabuki syndrome and diaphragmatic defects: A frequent association in non-Asian patients? *Am J Med Genet* 2000; 91:164-165.

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