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Editorial

The Kabuki Kid

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Editorial

A 5-year-old child was presented to the Outpatient Department (OPD) by his mother with complaints that her child was not gaining adequate height. She started noticing it around the age of 3 years compared to the children in Anganwadi. She also complained that he had less interest in activities compared to the children in his peer group. There is also a history of poor performance at the Anganwadi in writing or singing rhymes. There was no history of inadequate dietary intake, inadequate diet intake, pallor or pedal edema.

No history of chronic diarrhea or bowel issues. No history of polyuria, oliguria or polydipsia. No history of recurrent respiratory infections or wheezing. No history of constipation, lethargy or dry skin. No history of bony deformities or recurrent fractures. No history of headache, vomiting or visual disturbances.

There is a history of acyanotic congenital heart disease Ventricular Septal Defect (VSD) with left-to-right shunt was detected in the newborn baby and follow-up echo at 9 months showed VSD of the same size. No history of systemic illness, brain tumors, swelling, irradiation in the past, long-term steroid intake or Tuberculosis (TB) infection.

There was a history of abortion for the mother in the first pregnancy during the first trimester. She spontaneously conceived in the current pregnancy. She had a history of gestational diabetes mellitus at the 3rd month, for which she received insulin and a history of gestational hypertension at the 8th month, for which she was on some oral medication. An anomaly scan showed polyhydramnios and a Doppler scan showed Doppler abnormality, following which she was taken for emergency Lower Segment Cesarean Section (LSCS).

This is the 3rd child, preterm baby born at 32 weeks with a birth weight of 2.42 kg, cried soon after birth. There was a history of NICU admission initially for 4 days in view of respiratory distress syndrome and the baby was on Continuous Positive Airway Pressure (CPAP) for 1 day. He was on NG feeds with extra supplements for 2 days, followed by paladai feeds. He received IV antibiotics and IV fluids for 4 days. Echo showed VSD.

He was discharged on postnatal day 7 and readmitted on postnatal day 9 for 1 day in view of neonatal hyperbilirubinemia. He had language delay with a learning disability and is currently on speech and learning therapy. He is adequately nourished and immunized up to age. There is a history of mental retardation in maternal siblings child with a history of V-P shunting for hydrocephalus. History of hypothyroidism and infertility {varicocelectomy} for father.

On General Examination

Child had dolicocephaly with prominent forehead, bilateral proptosis, large eyes, depressed nasal bridge, upturned nostrils, hypertelorism, atrophic papillae, high arched papillae, dental caries, smooth philtrum, down turned lips, short hands and feet (Fig. 1).

Figure 1: Child with dolicocephaly.

No pallor, icterus, clubbing, cyanosis, lymphadenopathy and edema.
Vitals were stable
Anthropometry showed -
Weight - 16.6 kg- between 25th and 50th centile
Height - 99.5 cm -below 3rd centile
Head circumference - 49cm - between 3rd and 50th centile
MUAC -17 cm
US/ LS - 1.1 : 1
Arm span -97 cm
Chest circumference - 56 cm
MPH - between 50 and 75th centile
Bone age - 2yrs
All systems were within normal limits

He was admitted for detailed evaluation and management. On anthropometric assessment, proportionate short stature was revealed. BA< HA <CA. MRI was normal. X-ray of the left wrist (PA view) showed a bone age delay of 2 years. Ophthalmology evaluation was normal. Echocardiography showed a VSD of the same size and mild valvular pulmonary stenosis.

An endocrinology consultation was done. On the growth hormone stimulation test, the growth hormone level was found to be low. All other routine blood investigations and biochemical tests were normal. Thyroid function tests were also normal. A genetic study was done, which was suggestive of Kabuki syndrome. He was started on growth hormone replacement therapy at a dose of 15 micrograms per day, administered subcutaneously once daily at 9 pm. He was monitored for complications and side effects. His height, weight and growth velocity were charted regularly.

After one month, his height improved to 104.4 cm, placing him between the 10th and 25th percentile. He was continued on speech and learning therapy (Fig. 2,3).

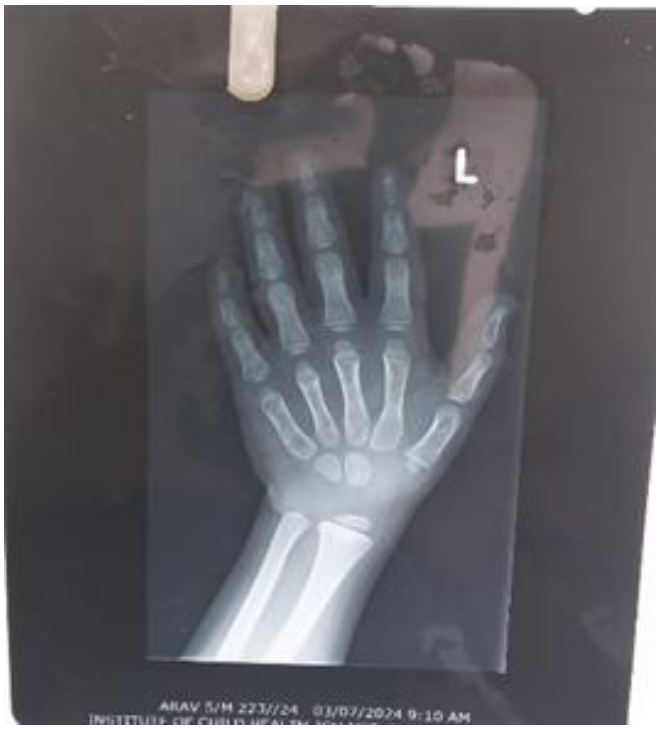

Figure 2: X-ray left wrist PA.

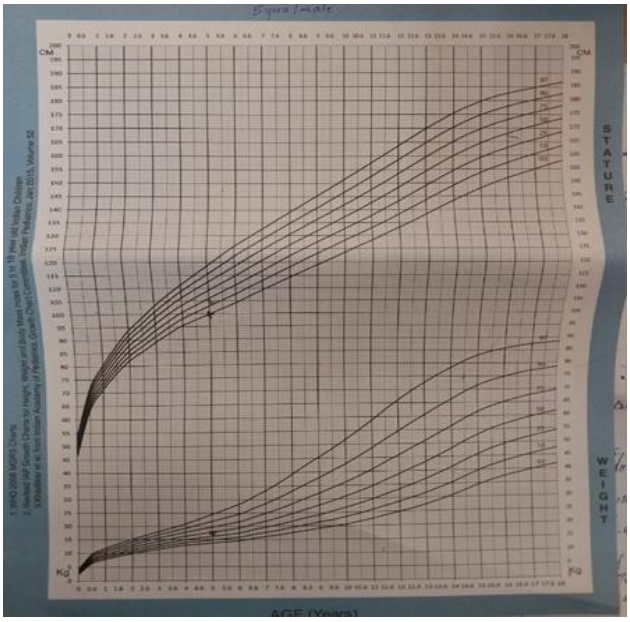

Figure 3: WHO 2006 and IAP 2015 combined height and weight growth chart.

Kabuki Syndrome (Kabuki Makeup syndrome/Nikawa Kuroki syndrome) initially reported in 1981 by Nikawa and Kuroki in 10 unrelated Japanese children, has now been reported in more than 400 patients, many of whom are non-Japanese. Because of the facial resemblance of affected individuals to the makeup of actors in Kabuki, the traditional Japanese theatre, this disorder has been referred to as Kabuki Syndrome. Approximately 60% of cases are caused by mutations in the MLL2 gene.

Natural History

Although many of the characteristic facial features are present in neonates, the features become more obvious with age. Severe feeding problems are common. Susceptibility to otitis media, upper respiratory infections and pneumonia is common and decreased levels of IgA, IgG and IgM have been documented, though not frequently. Delay in speech and language acquisition with articulation errors is also common.

Abnormalities

1. Growth -postnatal growth deficiency :onset in first year
2. Performance -development delay and low IQ
3. Craniofacial- long palpebral fissure with eversion of later portion of lower eyelid, ptosis, broad eyebrows with sparse lateral third, blue sclera, strabismus, epicanthal fold, short columella, large protuberant eyes, preauricular pit, cleft palate, tooth abnormalities, open mouth with tented upper lip
4. Skeletal - short, incurved 5th finger secondary to short 4^{th} and 5^{th} metacarpals, short middle phalanges, brachydactyly, rib anomalies, vertebral anomalies, hip dislocations, scoliosis and kyphosis
5. Cardiovascular anomalies - coarctation of the aorta, bicuspid aortic valve, mitral valve prolapse, membranous VSD, pulmonary /aortic /mitral stenosis, TOF, DORV and TGA

Additional features include joint hyperextensibility, persistent fetal pad signs, excessive ulnar loops , renal anomalies, urinary tract anomalies and hearing loss.

Conflict of Interests

The authors declare that they have no conflicts of interest.

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