Picture Story

Williams syndrome

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(Key words: Williams syndrome)

A 5 year old girl was brought to our paediatric clinic with a history of delayed development. She was born to non-consanguineous healthy parents, at term, following an uneventful pregnancy, with a birth weight of 2.75 kg. No birth insult was recorded. She is the youngest in the family of three children. Elder siblings are healthy.

During the first few years of her life feeding has been extremely difficult as she used to regurgitate. She was able to sit with support at age of 11 months and sat unaided at 13 months. She started walking alone at two and half years. Still she finds it difficult to run, stand on one foot and climb upstairs. Speech was initially delayed but is now garrulous. Though she attends a pre-school, she is significantly retarded intellectually, when compared with her peers. She can't identify letters, numbers or colours but has a remarkable ability in singing and dancing. Her attention span is significantly short.

On examination, she was found to have characteristic facies of Williams syndrome comprising a retrousse nose with flat nasal bridge and bulbous upturned tip, long philtrum, wide mouth with full lower lip, small chin, flat malar region with full cheeks, periorbital fullness, medial eyebrow flare, epicantic folds and long neck (Figure 1). Her height was 94 cm and weight 10 kg. Both were well below the third centile. She was very friendly in nature and loquacious. Sudden loud noises always made her uncomfortable suggesting that she is having hyperacusis.

A significant systolic murmur was heard over left lower sternal border and also over the pulmonary area. 2D Echo revealed it as a sub-aortic ventricular septal defect and excluded the presence of supravalvar aortic stenosis and peripheral pulmonary artery stenosis. Blood pressure was within normal range for age.

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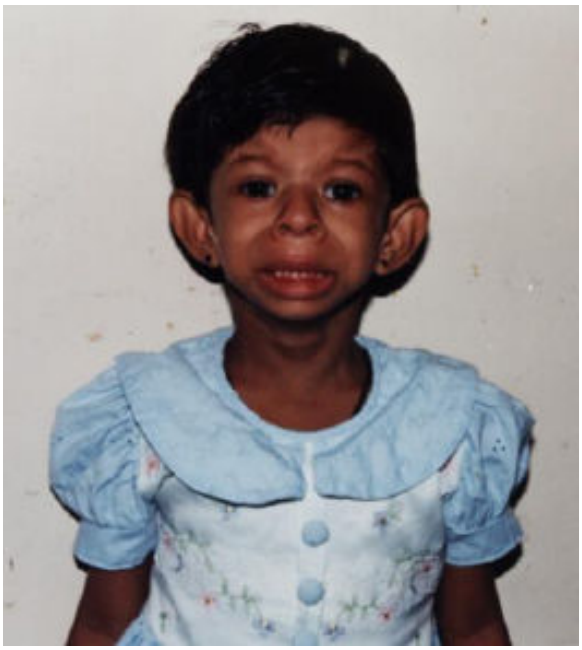

Figure 1

Femoral pulses were normal in volume. Serum ionized calcium level was 1.17mmol/L (normal range 1.12 – 1.32mmol/L). Ultrasound scan of abdomen did not reveal any abnormality in the genitourinary system.

Williams syndrome (WS) was described in 1961 by Williams et al who recognized a group of children with supravalvar aortic stenosis, mental retardation and dysmorphic facial features[1]. WS is caused by a micro deletion of number 7 chromosome at 7q11.23[2]. Routine chromosome analysis is usually normal and the deletion is detected by fluorescent in situ hybridization (FISH) using a probe for the elastin gene[3].

Infants with WS are usually born following an uneventful pregnancy with an average birth weight of 2760 g[4]. In the neonatal period feeding problems are common and often accompanied by vomiting and poor weight gain4. There is postnatal growth

retardation with most children below the 10th centile for height[4].

A proportion of infants are found to have idiopathic hypercalcaemia, which is treated with a low calcium and vitamin D restricted diet[4]. The underlying hormonal basis for the hypercalcaemia is not known. Resolution of hypercalcaemia occurs spontaneously, usually at around 18-24 months of age[4].

Developmental milestones are delayed with sitting unsupported achieved at an average of 13 months and walking at 28 months[4]. After an initial period of speech delay most children with WS tend to be overtalkative with a characteristically hoarse voice. This, with a notable overfriendliness towards strangers, has been termed a "cocktail party manner"[4]. Hypersensitivity to certain sounds or hyperacusis affects over 90% of individuals and appears to be centrally mediated[4]. Intelligence quotient (IQ) is usually in the range 40-85[4]. People with WS have relatively good verbal abilities and auditory rote memory with deficient visuospatial abilities and this has been termed the WS cognitive profile (WSCP)[5]. While many develop quite good reading ability, few have more than elementary numeracy[4]. Poor concentation and distractibility are almost universal[4].

Cardiovascular abnormalities occur in around 75% of cases with WS. Characteristic abnormalities are supravalvar aortic stenosis and peripheral pulmonary artery stenosis, although valvar and septal defects occur less commonly[4]. Hypertension occurs in around one third of patients with WS[4]. Renal artery, cerebral and left coronary artery stenoses have been described[4]. Therefore periodic cardiac and blood pressure monitoring is mandatory.

Both structural and functional urological abnormalities are seen with increased frequency in WS[4]. Hernias, most commonly inguinal occur in over a third of cases[4]. Scoliosis is seen in around 17%, large joint contractures in around 15%, radio-ulnar synostosis in 10% and recurrent patellar dislocations in 5% of cases[4]. Physiotherapy is very helpful in improving muscle tone, strength and joint range of motion. Most acquire independence in toileting, dressing and washing although supervision is often needed[4].

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