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Townes-Brocks Syndrome with Hypothyroidism

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Townes-Brocks syndrome (TBS) is an autosomal dominant disorder with multiple malformations and variable expression. Major findings include external ear anomalies, hearing loss, limb deformity, imperforate anus, and renal malformations. Hypothyroidism is not a recognized feature of TBS. We are reporting a case of TBS with hypothyroidism, a rare association.

Keywords: *Chromosome 16q12.1, Hypothyroidism, SALL1, Townes-Brocks syndrome.*

Townes-Brocks syndrome is an autosomal dominant multiple malformation syndrome characterised by external ear malformations with sensoryneural hearing loss, thumb anomalies, pes planus and anorectal malformation. Intelligence is usually normal, although mild-moderate mental retardation has been reported(1,2). Townes and Brocks first described the syndrome in 1972(3). Since that time over 65 cases have been published(1-6).The gene for Townes-Brocks syndrome was mapped to 16q12.1 through identifying subjects with TBS and cytogenetic abnormalities(4-6).

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Case Report

The patient was 2½-year-old female child, born of non-consanguineous marriage to a primigravida mother with no antenatal complications. She was born vaginally at term with a birth weight 2000 g, length of 41.5 cm and head circumfrence of 32 cm. She was noted to have anal atresia with recto-vestibular fistula. Abdominal ultrasound showed absent left kidney, which was confirmed by renal scan. No other abnormality was noted at that time. At 2 years, the child was operated for the ano-rectal malformation.

At 2½ years the child presented with complaints of rapid breathing since 5 days, increasing pallor, growth retardation and delayed milestones. On examination she was conscious but irritable, pale, tachypneic, respiratory rate was 60/min and heart rate was102/min. She had facial dysmorphism with depressed nasal bridge and left sided lop ear (*Fig. 1*). Chest was clear with acidotic breathing. Liver was palpable 3 cm below costal margin, firm in consistency. Spleen was non-palpable. Others systems were normal. Child was mentally retarded.

Child's weight was 5 kg (40% of 50th centile), length was 62 cm (70% of 50th centile), head

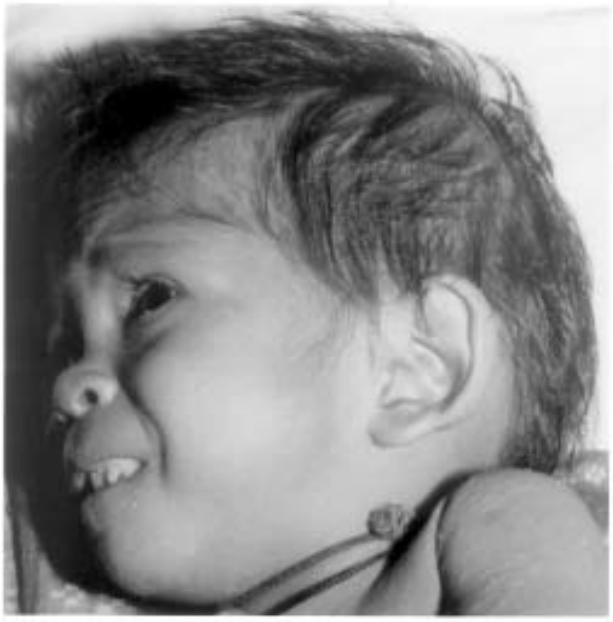

Fig. 1. Facial dysmorphism with lop ear

circumference was 42 cm (<5th centile), mid arm circumference was 9 cm and upper segment/lower segment ratio was 1.2:1.

Laboratory investigations revealed anemia (Hb 4.1 g/dL) and normal cell counts. Blood urea (240 mg/dL) and serum creatinine (6.5 mg/dL) were elevated. ABG revealed marked metabolic acidosis. Chest X-ray was normal. Ultrasound of right kidney showed increased echogenecity with splitting of renal pelvis and left kidney was not visualized. Renal scan showed absent left kidney. Skeletal survey revealed bone age of 6 months with no other abnormality. Auditory brainstem evoked response (ABER) showed bilateral sensory neural hearing loss: left 120 decibel and right 100 decibel. Echocardiography showed no cardiac abnormality. Thyroid profile showed T3 < 0.50 pg/mL, T4 < 0.15 ng/dL, TSH >100 mIU/ L. Thyroid scan was normal. Perchlorate discharge test could not be done. Karyotyping showed normal XY chromosomal pattern but detailed structural analysis of chromosome 16 could not be done.

Discussion

Townes and Brocks first described the syndrome in 1972 in a single family wherein father and 5 of the 7 offspring had a pattern of imperforate anus, sensorineural hearing loss with lop ear and bony deformities of hand and feet(4).

TBS is estimated to occur in 1:250,000 live born(7) but may be misdiagnosed because its defects overlap with those of other genetic diseases(1).

Diagnostic criteria include two or more of the following; anorectal malformation, external ear malformation with sensory neural hearing loss, hand malformation and a relative with the syndrome(1,8). Clinical features include ear anomalies and hearing loss. Ear shape includes “satyr” and “lop” and hearing loss primarily sensorineural. Limb defects include triphalangeal thumb and preaxial polydactyly. Anal anomalies include imperforate anus, rectovaginal or rectourethral fistula. Genitourinary anomalies include hypoplastic or dysplastic kidneys, renal agenesis. In eight reported patients renal failure or impaired renal function was present(2,9,10). Mental retardation is also reported (2,9,10).

Differential diagnosis includes VATER & VACTERL associations, Baller-Gerold syndrome, Oculoauriculovertebral spectrum (OAV) and Cat eye syndrome.

Our patient had renal failure, anorectal malformation, left renal agenesis, left lop ear, bilateral sensory neural hearing loss, hypothyroidism, growth retardation, mental retardation. Anemia was ascribed to chronic renal failure. Yano *et al*(11) in 1998 described a 10 yr old boy with anomalous left ear, mild sensory neural hearing loss, preaxial polydactyly, anal atresia, prominent perineal raphe, and chronic renal failure. He had congenital hypothyroidism with an organification defect. Authors suggested that this is Townes-Brocks and Pendred syndrome in the same patient(11). This is the second report of TBS associated with hypothyroidism. Extensive genetic analysis is required to prove the association.

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Volvulus Complicating Dextrogastria in an Infant

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We report eventration of right hemi-diaphragm resulting in gastric volvulus of the right-sided stomach in an infant. The diagnosis of this rare association was made with contrast CT scan. Patient was initially managed with reduction of stomach, plication of right hemi-diaphragm, anterior gastropexy and Ladd's procedure, but required re-laparotomy after two months for recurrent volvulus.

Keywords: *Dextrogastria, Infant, Volvulus.*

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Congenital anomalies in the position or attachment of proximal portion of alimentary tube are exceedingly rare and usually occur as apart of general transposition of the viscera. Isolated dextrogastria is the rarest of all visceral transpositions and usually coexists with eventration of right hemi diaphragm. Another gastric condition known to coexist with eventration of diaphragm is gastric volvulus. But eventration of right hemi diaphragm resulting in volvulus of the right-sided stomach has never been reported hitherto. We report this association in an infant here. Another peculiarity of this case lies in the fact that rotational abnormalities of both the foregut and the midgut coexisted.

Case Report

An 8-month-old male child was admitted with recurrent chest infections and non-bilious vomiting since birth, for which he was treated with antibiotics elsewhere. There was no history of cyanosis. Antenatal history was not significant. Examination revealed a chronically malnourished baby weighing 6.4 kg. Although there was no apparent distress, auscultation of the chest revealed absence of air entry in inframammary and infraaxillary region in the right side. The breath sounds on the left side and