

Townes-Brocks syndrome or hemifacial microsomia-VACTERL phenotype?

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ABSTRACT

We report a male with imperforate anus, pedunculated triphalangeal thumbs, hemifacial microsomia, microtia, preauricular tags and cardiac anomalies. This is the first individual with Townes-Brocks syndrome to have hydrocephalus.

INTRODUCTION

In 1972, Townes and Brocks described an association of imperforate anus, supernumerary or triphalangeal thumbs, satyr ears, preauricular tags and sensorineural hearing loss in a father and five of his seven offspring. Afterwards, more families with dominant transmission of an analogous combination of anal, renal, limb and ear anomalies were described by Reid and Turner (1976), Kurnit *et al.* (1978) and Walpole and Hockey (1982). Isolated but very similar cases were reported by Monteiro de Pina-Neto (1984) and Hersh *et al.* (1986). This has been recognized as a dominant inherited syndrome, the Townes-Brocks syndrome (De Vries-Van der Weerd *et al.*, 1988) (McKusick, 1983, catalog number *10748).

Bivalent associations between anorectal malformations, limb defects, cardiovascular anomalies, vertebral defects, tracheoesophageal fistula and urogenital malformations are not uncommon, as reviewed by Khoury *et al.* (1983). A broader association between imperforate anus, polydactyly, vertebral

anomalies and tracheoesophageal fistula has been described by Say and Gerald (1968).

A number of clinical features in the Townes-Brocks syndrome may also be seen in VACTERL association, and hemifacial microsomia (HFM). However, differentiation of the two conditions should be possible (Hersh *et al.*, 1986).

CLINICAL REPORT

The proband was a male born at term after an uncomplicated pregnancy. Cesarean section was performed because of prolonged labor. He was evaluated for multiple congenital anomalies by the Malformations Surveillance project in Pelotas, RS, Brazil.

He was the second child of nonconsanguineous, healthy young parents. There was no family history of congenital malformations or hearing loss. At birth, his weight was 3200 g, length 48 cm and head circumference 36 cm.

Significant findings included a high imperforate anus (Figure 1), rectourethral fistula, hemifacial microsomia (Figure 2), microtia (Figure 3) grade III (Kaye *et al.*, 1992) with preauricular skin tags on the

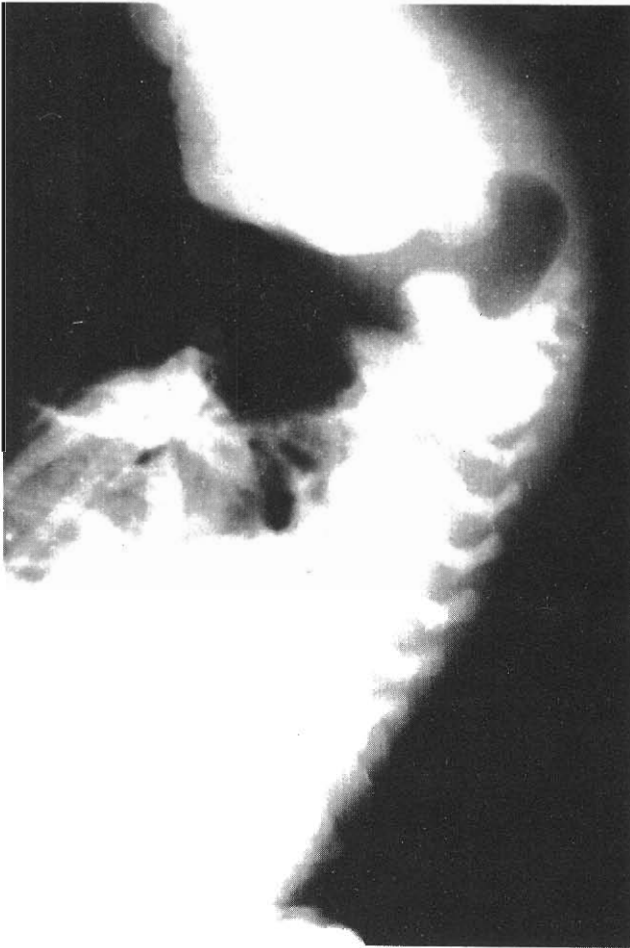


Figure 1 - X-ray of the abdomen showing rectal obstruction.

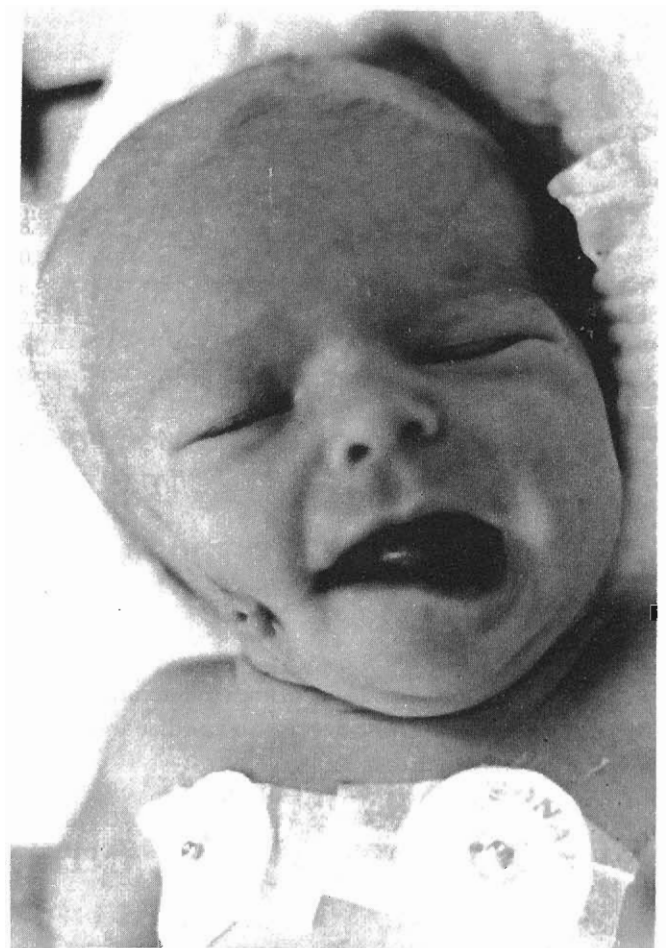


Figure 2 - Frontal view of the propositus showing hemifacial microsomia.

right side, pedunculated triphalangeal right thumb (Figure 4), hydrocephalus, and cardiac anomalies.

Radiographs of the abdomen showed acute obstructive abdomen. Computerized tomography of the brain demonstrated dilated lateral and third ventricles, and noncommunicating hydrocephalus. Echocardiography, performed for investigation of heart murmur, showed complete blockage of the right bundle, and right ventricular overload. Chromosomal analysis revealed normal karyotypes.

Surgery performed: Colostomy and ventricular-peritoneal shunts.

DISCUSSION AND CONCLUSION

Townes-Brocks syndrome is characterized by ear anomalies (preauricular skin tags, auricular pits, pinna anomalies, satyr lop ears and sensorineural hearing loss), anal anomalies (imperforate anus type 1 or 3, anal stenosis, covered anus, rectovaginal fistula, perianal skin redundancy, and varying degrees of anterior

placement of the anus), radial anomalies (triphalangeal, bifid, supernumerary thumbs, coneshaped epiphyses and pseudoepiphyses of the second metacarpals), other anomalies of the hands and feet (pes planus and clinodactyly of the 5th toes), renal anomalies (hypoplastic kidney, meatal stenosis), cardiac anomalies (atrial or ventricular septal defects, tricuspid atresia and patent ductus arteriosus), and genital anomalies (hypospadias) (Ferraz *et al.*, 1989).

In view of its dominant mode of inheritance and its clinical spectrum, Townes-Brocks syndrome can be differentiated from the VACTERL association. Vertebral, tracheoesophageal and radius anomalies are absent in Townes-Brocks syndrome, while they are frequent in the VACTERL associations. On the other hand, hearing loss, satyr ears and preauricular tags are uncommon in the VACTERL association, but common in Townes-Brocks syndrome (De Vries-Van der Weerd *et al.*, 1988).

Townes-Brocks syndrome complements a series of specific syndromes, such as hemifacial microsomia and Goldenhar syndrome (or oculoauricu-

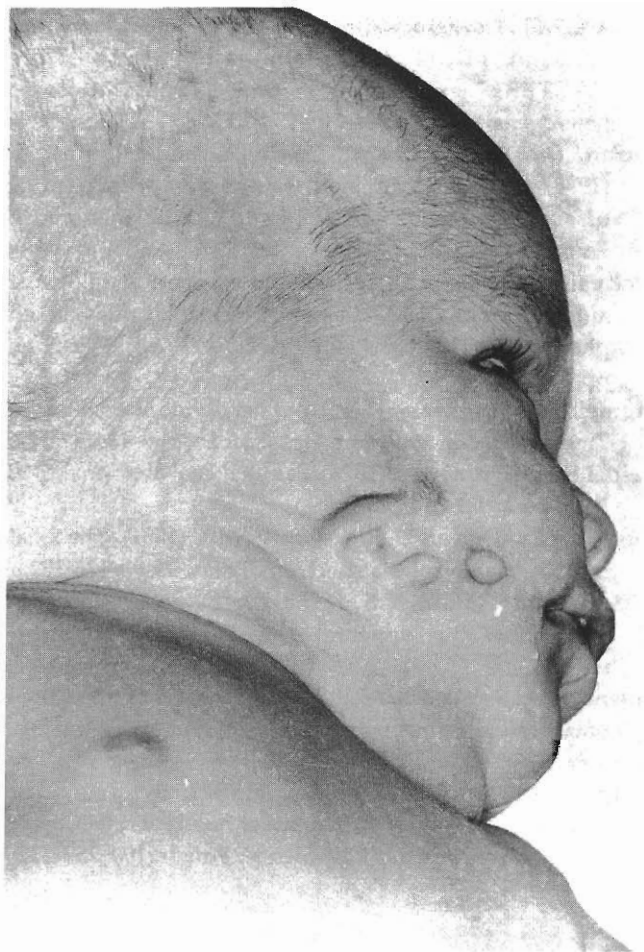


Figure 3 - Lateral view of the propositus showing microtia and preauricular tags.

lovertebral dysplasia). Hemifacial microsomia is a condition affecting primarily aural, oral, and mandibular development. This disorder varies in severity, and facial involvement is limited to one side in many cases. Goldenhar syndrome, which is characterized additionally by vertebral anomalies and epibulbar dermoids, is considered a variant of this complex (Cohen Jr. *et al.*, 1989). In an isolated case the diagnosis can be difficult, and confusion with VACTERL association may lead to an inaccurate estimate of genetical risk.

The VATER and hemifacial microsomia phenotypes are well documented in clinical genetics but, despite many studies, there are no definitive diagnostic markers and each phenotype appears heterogeneous. The most distinctive characteristic of each phenotype is the body region in which the major identifying malformations are located. In hemifacial microsomia it is the cephalic region and in VATER, it is the truncal region; but in numerous patients, these areas overlap (Duncan and Shapiro, 1992).

Duncan and Shapiro (1993) analyzed the malformations of 247 patients reported in the literature to



Figure 4 - Pedunculated triphalangeal thumb of the propositus.

have HFM. They observed the VATER association in 29 of the HFM patients, and 33 patients were designated to have the hemifacial microsomia-VATER (HFM-VATER) phenotype. They were compared to the malformations of 255 VATER and 101 sirenomelia patients studied in a similar fashion. Their data support the inter-relationships of the three phenotypes as a sequence which can explain a common embryological pathogenetic mechanism.

The patient studied by us is an isolated case, since no family members had features suggestive of a diagnosis of Townes-Brocks syndrome. He does not present all characteristics of Townes-Brocks syndrome, such as polydactyly. On the other hand, he has hydrocephalus (which also may be present in the VATER or VACTERL association); thus, he is the first individual with Townes-Brocks syndrome having hydrocephalus, or represents an overlapping phenotype (hemifacial microsomia-VACTERL phenotype).

RESUMO

Estamos relatando o caso de um menino com ânus imperfurado, polegar pedunculado e trifalangeal, microsomia hemifacial, microtia, apêndice pré-auricular e anomalia cardíaca. Este é o primeiro caso de síndrome de Townes-Brocks com hidrocefalia, permanecendo a dúvida se a hidrocefalia poderia ser incluída no espectro clínico da síndrome de Townes-Brocks.

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(Received February 13, 1995)