

## SHORT COMMUNICATION

### SHORT SYNDROME: REPORT ON A BRAZILIAN GIRL WITH DEAFNESS, GASTROESOPHAGEAL REFLUX AND POST-NATAL GROWTH RETARDATION

Simone V. Vitti, Simone G. Ghedini and A. Richieri-Costa

#### ABSTRACT

We report on a Brazilian girl with the clinical signs of SHORT syndrome. She also presented gastroesophageal reflux and evident post-natal growth retardation. The normal neuropsychological development and severe sensorineural deafness found are clearly signs of the SHORT syndrome. Clinical and genetic aspects are discussed.

#### INTRODUCTION

The SHORT syndrome is a rare condition characterized by lipoatrophy, slow weight gain and frequent illness during infancy, delayed dental eruption, delayed speech, short stature, Rieger anomaly, wide nasal bridge, hypoplastic alar cartilage, micrognathia, and joint hyperextensibility (Gorlin *et al.*, 1975; Sensenbrenner *et al.*, 1975). Sensorineural deafness and post-natal growth retardation were reported in one instance (Toriello *et al.*, 1985) and sensorineural deafness in two (Toriello *et al.*, 1985; Schwingshandl *et al.*, 1993). Here we report on a Brazilian girl with SHORT syndrome, presenting normal neuropsychological development, post-natal growth retardation, severe sensorineural deafness, and gastroesophageal reflux.

#### CLINICAL REPORT

JCN (Figures 1 and 2) the proposita, born in 1990, was the second child of a normal 32-year-old G2P2 white woman and her normal 32-year-old unrelated husband. Pregnancy was normal and there were no toxic, infectious, or traumatic incidents or X-ray exposure. Birth weight was

3,800 g. (> 50th centile), TBL was 51 cm (50th centile), and OFC was not recorded. Neuropsychological development was normal. She presented gastroesophageal reflux, and initial weight gain was slow. Severe sensorineural deafness was diagnosed during the first year of life. There was recurrent otitis and tonsillitis. Dental eruption was delayed, with the first tooth erupting after 18 months of age.

Clinical examination at age 45 months showed: height 90 cm (< 3rd centile), weight: 12.8 kg (< 3rd centile), OFC: 48 cm (3rd centile). She had a short stature, "triangular-shaped" face, deeply set eyes, micrognathia, mild anteverted ears, clinodactyly, lack of subcutaneous fat, joint hyperextensibility, sensorineural deafness, delayed bone age, delayed speech, and normal intellect.

G-banded chromosomes in peripheral lymphocytes were normal. Routine laboratory blood tests including HgH, and blood glucose measurement were normal. Bone age was delayed (Chronological age: 45 months/Bone age: 30 months). Axial and coronal CT images of the temporal bones were normal. Audiologic examination showed bilateral sensorineural deafness.

#### DISCUSSION

Most of the patients with SHORT syndrome present intrauterine growth retardation (IUGR), short stature, slow weight gain, "triangular face", deeply set eyes, Rieger anomaly, wide nasal bridge, hypoplastic alar cartilage, delayed dental eruption, lack of subcutaneous



Figure 1 - Frontal view of the proposita.



Figure 2 - Lateral view of the proposita.

Table I - Clinical findings in patients with the SHORT syndrome.

	I	II	III	IV	V	VI	Present case
	1	2					
IUGR	+	+	+	-	+	+	+
Slow weight gain	+	+	+	+	+	+	+
Frequent illness	+	+	+	+	+	-	+
"Triangular" face	+	+	+	+	+	+	+
Telecanthus	?	?	+	+	-	-	-
Deeply set eyes	+	+	+	+	-	+	+
Rieger anomaly	+	+	+	-	+	+	-
Wide nasal bridge	+	+	+	+	+	-	+
Hypoplastic alae	+	+	+	+	+	+	+
Micrognathia	+	+	+	+	+	+	+
Anteverted ears	+	+	+	+	+	+	+
Clinodactyly	+	?	+	+	+	-	+
Delayed dental eruption	+	+	+	+	+	+	+
Lack of subcutaneous fat	+	+	+	+	+	+	+
Joint hyperextensibility	+	+	+	+	+	-	+
Short stature	+	+	+	+	+	+	+
Deafness	-	-	-	+	-	-	+
Functional heart murmur	+	?	+	-	-	+	-
Inguinal hernia	+	+	-	-	-	+	-
Delayed bone age	+	+	+	+	+	-	+
Delayed speech	+	+	+	+	+	+	+
Normal intellect	+/-	?	+	+	+	+	+

I - Gorlin *et al.*, 1975; II - Sensenbrenner *et al.*, 1975; III - Torriello *et al.*, 1985; IV - Lipson, 1989; V - Aarskog *et al.*, 1983; VI - Schwingshandl *et al.*, 1993.

fat, delayed bone age, delayed speech, and joint hyperextensibility (Table I). Insulinopenic diabetes (Aarskog *et al.*, 1985), and diabetes mellitus secondary to severe insulin resistance (Schwingshandl *et al.*, 1993) have been described.

The patient here reported differed from the patient of Toriello *et al.* (1985) since she did not have IUGR, Rieger anomaly, hernia, or heart murmur, and she presented sensorineural deafness. The report on a third patient with deafness (Schwingshandl *et al.*, 1993) strongly suggests that this is a consistent finding within the clinical spectrum of the SHORT syndrome.

Most of the reported patients are isolated cases (Sensenbrenner *et al.*, 1975; Torriello *et al.*, 1985; Lipson *et al.*, 1989; Schwingshandl *et al.*, 1993; present case), however, autosomal dominant (Aarskog *et al.*, 1983), and recessive inheritance (Gorlin *et al.*, 1975) have been suggested.

In spite of the small number of reported patients (five male/three female) there is evidence of clinical and genetic heterogeneity in the SHORT syndrome. Only the report of new patients with these signs could determine if gastroesophageal reflux is part of the clinical spectrum.

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## RESUMO

Nós relatamos sobre uma paciente com a síndrome SHORT. Trata-se da primeira descrição na literatura brasileira. Como sinais adicionais a paciente apresenta refluxo gastro esofágico e evidente retardo do crescimento pós natal. Surdez neurosensorial assim como desenvolvimento neuropsicológico normal são partes integrantes desta síndrome.

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