RELATO DE CASO

Audiological findings in patients with the Richieri-Costa-Pereira syndrome

Achados audiológicos em pacientes com síndrome Richieri-Costa-Pereira

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Abstract We described the audiological findings in Brazilian patients with the Richieri-Costa-Pereira syndrome. The main signs observed in this condition are short stature, Robin sequence, cleft mandible and limb defects. Nine patients with ages varying between 1-23 years old were submitted to audiological assessment consisting of inquiries directed to hearing function, pure tone threshold audiometric exam, sound fields audiometry and impedance testing. To date audiological assessments in patients with the Richieri-Costa-Pereira syndrome have not been described in the literature.

Keywords Richieri-Costa-Pereira syndrome, cleft mandible, Robin sequence, audiological assessment.

Resumo Nós descrevemos os achados audiológicos de nove pacientes brasileiros com a síndrome Richieri-Costa Pereira. Os principais sinais clínicos observados nesta condição são baixa estatura, seqüência de Robin, fissura mandibular e anomalias de membros. Avaliação audiológica, incluindo audiometria tonal liminar, audiometria tonal condicionada, audiometria em campo livre e impedanciometria foram realizadas em nove pacientes com idades variando entre 1 e 23 anos. Até o momento, avaliação audiológica em pacientes com este quadro não tem sido descrita na literatura.

Palavras-chave Síndrome de Richieri-Costa-Pereira, fissura mandibular, Seqüência de Robin, avaliação audiológica

Introduction

The Richieri-Costa-Pereira syndrome is an autosomal recessive condition, which was first described in Brazil in 1992[1,2]. The main clinical signs observed are short stature, Robin sequence (Fig. 1A-C), cleft mandible or cleft lower alveolar ridge (Fig. 3), pre/postaxial hand anomalies and clubfoot (Fig. 2A-B). Other cases, Brazilian[3,4] and non-Brazilian[5], have been reported. "Laryngeal malformation and characteristics of the voice have been reported in this condition, by the first time by Tabith and Bento-Gonçalves[6] and posteriorly in other two patients[7]."

Audiological assessments were performed in nine Brazilian patients (1 male and 8 female), including pure-tone air and bone conduction audiometry, sound field audiometry, speech audiometry, and middle ear immittance testing, which consists of tympanometry and acoustic reflex testing. The degree of hearing loss was defined according to BIAP[8], and the tympanometric results were classified according to Jerger[9]. These finds have been described and discussed.

Clinical Reports

All patients described have compatible features with diagnosis of the Richieri-Costa-Pereira syndrome. Some patients were previously reported by Richieri-Costa and Pereira[2,3], and by Richieri-Costa and Brandão-Almeida[4]. Laryngeal malformations described in these patients were considered an additional clinical sign in the Richieri-Costa-Pereira syndrome[6,7]. Tables 1 and 2 summarize the main clinical features and audiological assessments.

Discussion

Hearing loss was observed in 6 out of 9 patients studied (Table 2), and the type of hearing loss in 5 of out 6 patients was conductive, whereas one patient (Patient 4) presented unilateral severe sensorioneural hearing loss. "Patient 4 had submucous cleft (not surgically corrected) and at the age of 2 years, she had mumps infection. Hearing loss was noted at the age of 6 years. Mumps infection usually causes a total unilateral hearing loss[10].

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The hearing loss observed in Patient 4 is different from those seen in patients with submucous cleft, and in patients with a history of mumps infection. Thus, the etiology of this hearing loss could not be assured.

Patients 5, 6, and 9 had cleft palate (surgical repair was performed in patients 5 and 6). The conductive hearing loss present in these patients can be attributed to cleft palate, since there is a high incidence of conductive hearing loss in patients with cleft palate[11-13]. According to Bzoch[13], patients with cleft palate show considerable alterations of middle ear caused by auditory tube dysfunction.

In patients 7 and 8, the hearing loss was of a conductive type, and they had no cleft palate or a history otitis media.

Conclusion

In conclusion, the conductive hearing loss was frequent in the Richieri-Costa-Pereira syndrome (66.7%). Considering that, 2 patients out of 5 with conductive hearing loss had no cleft palate or a history of otitis media a more detailed auditory investigation in patients with this syndrome so as to verify whether this kind of auditory impairment belongs to the phenotypic spectrum of this condition.

Figure 1A-C: Clinical aspect of the patient.

Figure 2A-B: Note anomalies of hands and feets.

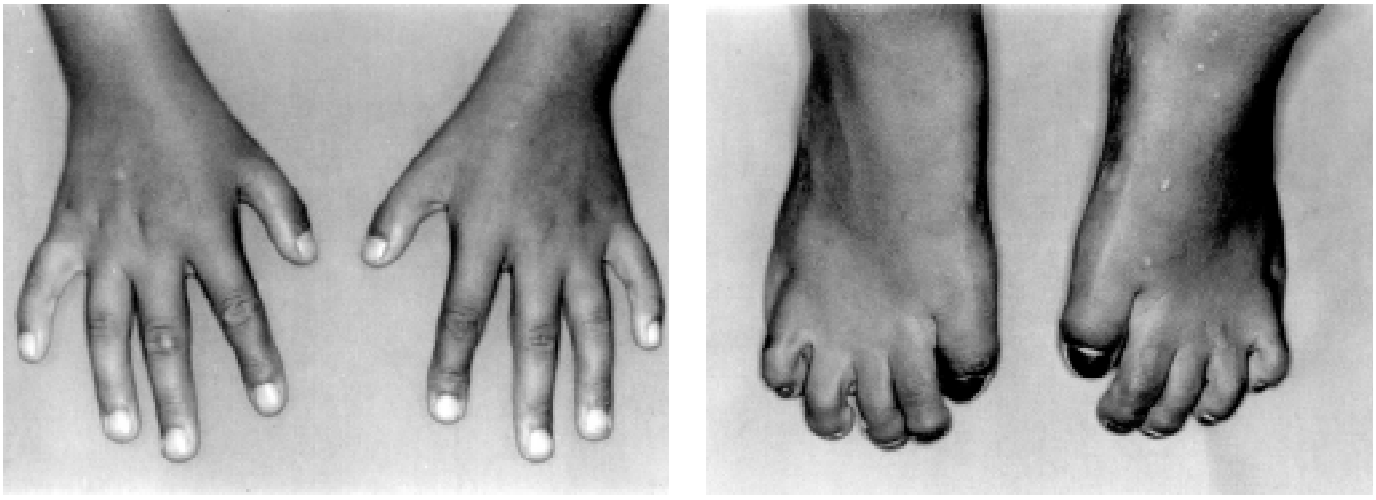

Figure 3 – Note mandible cleft.

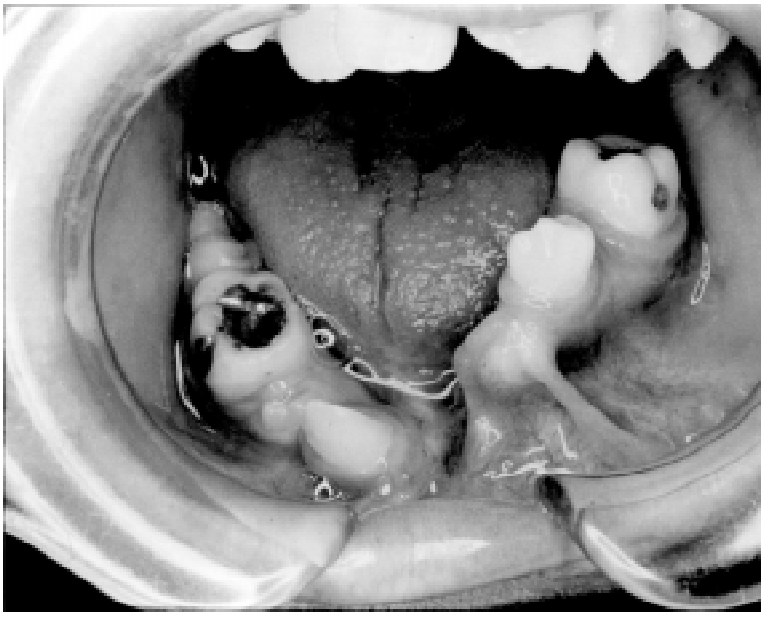

Table 1. Clinical features of nine patients with the Richieri-Costa-Pereira syndrome

	Patients								
Clinical feature	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9
Gender	F	F	M	F	F	F	F	F	F
Parental consanguinity	-	+	+	+	-	+	+	-	-
Short stature	+	+	+	+	+	+	+	+	+
Microstomia	+	+	+	+	+	+	+	+	+
Micrognatia	+	+	+	+	+	-	+	+	+
Cleft palate/uvula	-/-	-/-	-/-	+/+	+/-	+/-	-/-	-/+	+/-
Cleft mandible	-	-	+	-	+	+	+	+	-
Robin sequence	+	-	-	+	+	+	+	-	+
Laryngeal malformation	+	+	+	+	-	+	-	-	-
Low-set and prominent ears	+/+	+/+	+/+	+/+	+/-	+/+	+/+	+/+	+/+
Upper limb anomalies	+	+	+	+	+	+	+	+	+

(+) present; (-) absent; (nl) normal

Table 2. Audiological findings of nine patients with the Richieri-Costa-Pereira syndrome

	Patients								
Clinical audiological	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9
Evaluation age	12 years	16 years	23 years	14 years	13 years	16 years	13 years	13 years	23 months
Normal hearing in the right ear	+	+	+	-	-	-	-	-	-
Normal hearing in the left ear	+	+	+	+	-	-	-	-	-
Bilateral conductive hearing loss	-	-	-	-	+	+	+	+	+
Sensorioneural hearing loss in the right ear	-	-	-	+	-	-	-	-	-

(+) present; (-) absent

REFERENCES

1. OMIM Online Mendelian Inheritance in Man. Johns Hopkins University, Baltimore, [citado 2004 Mar 02]. World Wide Web Disponível em: http:/www.ncbi.nlm.nih.gov/omim/
2. Richieri-Costa A, Pereira SC. Short stature, Robin sequence, cleft mandible, pre/postaxial hand anomalies, and clubfoot: a new autosomal recessive syndrome. Am J Med Genet 1992;42(5):681-7.
3. Richieri-Costa A, Pereira SC. Autosomal recessive short stature, Robin sequence, cleft mandible, pre/postaxial hand anomalies, and clubfeet in male patients. Am J Med Genet 1993; 47(5):707-9.
4. Richieri-Costa A, Brandão-Almeida IL. Short stature, Robin sequence, cleft mandible, pre/postaxial hand anomalies, and clubfoot another affected Brazilian patient born to consanguineous parents. Am J Med Genet 1997;71(2):233-5.
5. Walter-Nicolet E, Coeslier A, Joriot S, Kacet N, Moerman A, Manouvrier-Hanu S. The Richieri-Costa and Pereira form of acrofacial dysostosis: first case in a non-Brazilian infant. Am J Med Genet 1999;87(5):430-3.
6. Tabith Jr A, Gonçalves CG. Laryngeal malformations in the Richieri-Costa and Pereira form of acrofacial dysostosis. Am J Med Genet 1996;66(4):399-402.
7. Tabith Jr A, Bento-Gonçalves CG. Laryngeal malformation in the Richieri-Costa-Pereira acrofacial dysostosis: description of two new patients. Am J Med Genet 2003;122A(2):133-8 International Bureau for Audiophonology - BIAP (BIAP recomendation no. 02/01: Audiometric classification of hearing impairments; October 26th,1996): http://www.biap.org/biapanglais/biaprecomangl/htm.
8. International Bureau for Audiophonology - BIAP (BIAP recomendation no. 02/01: Audiometric classification of hearing impairments; October 26th,1996). [citado 2004 Nov 18]. Disponível em: http://www.biap.org/biapanglais/rec021eng.htm
9. Jerger J. Clinical experience with impedance audiometry. Arch Otolaryngol 1970; 92(4):311-24.
10. Katz J. Tratado de audiologia clínica. 4ª ed. São Paulo: Manole; 1999.
11. Stool SE, Randal P. Unexpected ear disease in infants with cleft palate. Cleft Palate J 1967;4:99-103.
12. Walton WK. Audiometrically "normal" conductive hearing losses among the cleft palate. Cleft Palate J 1973;10:99-103.
13. Bzoch KR. Communicative disorders related to cleft lip and palate. 4th ed. Texas: PRO-ED; 1997.

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