



Universidade de São Paulo

Biblioteca Digital da Produção Intelectual - BDPI

Hospital de Reabilitação de Anomalias Craniofaciais - HRAC

Comunicações em Eventos - HRAC

2013-11-08

Speech, hearing and swallowing in Richieri-Costa Pereira syndrome clinical report

Simpósio Internacional de Fissuras Orofaciais e Anomalias Relacionadas, III, 2013, Bauru.
<http://www.producao.usp.br/handle/BDPI/46691>

Downloaded from: Biblioteca Digital da Produção Intelectual - BDPI, Universidade de São Paulo

SPEECH, HEARING AND SWALLOWING IN RICHIERI-COSTA PEREIRA SYNDROME: CLINICAL REPORT

OLIVEIRA ACASF***, Miguel HC, Oliveira RP, Yamashita RP, Antoneli MZ

Setor de Fonoaudiologia, Hospital de Reabilitação de Anomalias Craniofaciais - HRAC-USP, Bauru/SP

BACKGROUND: Richieri-Costa and Pereira (1992) described a autosomal recessive condition involving acrofacial dysostosis, often associated with Robin sequence. Phenotypic findings include facial, skeletal, genetic, laryngeal and auditory anomalies, with no specific reports regarding hearing, speech or swallowing. **PURPOSE:** To describe speech, hearing and swallowing in a patient with Richieri-Costa Pereira syndrome (RCPS). **CLINICAL REPORT:** 8-year-old boy with cleft palate and mandible, supported by the institution since 2 months, requiring tracheotomy and gastrostomy. Palatoplasty was performed at 2 years. At age 4, mandibular osteogenic distraction was performed and tracheotomy removed. Epiglottis agenesis was confirmed at 6 years. Gastrostomy was removed when he was 7 years old and is currently with exclusive oral intake. Speech was assessed since age 2, with articulatory mistakes due to anatomical limitations and later dysphonia was also observed. He underwent some periods of speech therapy and in the last assessment no impairment of speech intelligibility was observed, despite the presence of nasal rustle in some phonemes. The first hearing evaluation (tonal audiometry and immittance measures) was performed when the patient was 5 years old, and repeated one year later with similar results: mild conductive hearing loss in the left ear. Two years later, hearing evaluation revealed normal hearing with a type A tympanometric curve bilaterally. **CONCLUSION:** Speech and hearing follow-ups showed initial impairment in speech intelligibility and hearing sensitivity, which improved with speech therapy, surgical reconstructive procedures and structural growing. Swallowing was impaired due to larynx malformations, but a compensatory mechanism was developed for airway protection.