Auditory processing and language evaluation in patients with Cat Eye Syndrome - case study

Processamento auditivo e avaliação de linguagem em indivíduos com a síndrome do Olho de Gato - estudo de caso

Procesamiento auditivo y evaluación del lenguaje en indivíduos con Síndrome de Ojo de Gato - estudio de caso

Aline Priscila Cibian[*]
Larissa Mendonça Agessi[*]
Thiliê Palácios[*]
Juliana Silveira Ferreira Medeiros[*]
Ellen Osborn[*]
Liliane Desgualdo Pereira[*]

Study presented as posters at the 22nd Brazilian Congress of Speech Language Pathology and Audiology, on 8 October 2014, in Joinville-SC, Brazil.

* *Universidade Federal de São Paulo (UNIFESP), São Paulo,SP,Brasil.*
Conflito de interesses: *Não.*
Authors' Contribution: APC: participation in data collection and analysis and composing of article; and submission; LDP: chief supervisor, analysis and composing of article; TP: participation in data collection and analysis and composing of article; JSFM: participation in data collection and analysis and composing of article, and poster presentation in scientific congress; EO: supervisor LMA: participation in data collection and analysis and composing of article..
Correspondence Adress: Aline Priscila Cibian -
E-mail: aline.cibian@gmail.com
Received: 11/ 07/ 2015 **Approved:**31/ 08/ 2015

Abstract

The Cat Eye Syndrome is a rare syndrome characterized by coloboma of iris and pre-auricular malformation. The objective of this study was to evaluate the speech, comprehension of speech and auditory processing of two brothers with a diagnosis of cat eye syndrome. The evaluations were performed in a neuroaudiologic clinic and speech screening clinic of the Speech Therapy Department, Federal University of São Paulo. The tests were: pure tone audiometry, research of the speech reception threshold, tympanometry and acoustic reflex, index of speech recognition percentage, speech in noise with figures, digit dichotic test Random Gap Detection Test, vocabulary test and phonology and an Item which assesses speech comprehension of Luria Nebraska Neuropsychological Assessment. In the evaluation the children showed bilateral sensorineural hearing loss of minimum level, changes in hearing closure and figure-ground in auditory processing, and in the skills of listening and spoken language issue in phonology and vocabulary tests.

Keywords: Speech Language Pathology and Audiology; Hearing; Hearing Disorders; Auditory Perception.

Resumo

A síndrome do Olho de Gato é uma síndrome rara, caracterizada por coloboma de íris e malformação pré-auricular. O objetivo deste estudo foi relatar a avaliação fonoaudiológica e de processamento auditivo de dois irmãos com diagnóstico da síndrome do olho de gato. As avaliações foram realizadas no ambulatório de neuroaudiologia e no ambulatório de triagem fonoaudiológica do Departamento de Fonoaudiologia da Universidade Federal de São Paulo. Os testes avaliados foram: audiometria tonal liminar, pesquisa do limiar de recepção de fala, imitanciometria e pesquisa do reflexo acústico, Índice percentual de reconhecimento de fala, fala no ruído com figuras, dicótico de dígitos, detecção de intervalos aleatórios, prova de vocabulário e fonologia do teste de linguagem infantil e item que avalia a compreensão oral da Avaliação Neuropsicológica Luria Nebrasca. Na avaliação as crianças apresentaram perda auditiva neurossensorial de grau mínimo bilateral, alteração nas habilidades auditivas de fechamento e figura-fundo na avaliação do processamento auditivo e nas habilidades de linguagem de compreensão oral e emissão oral nos testes de vocabulário e fonologia.

Palavras-chave: *Fonoaudiologia; Audição; Transtornos da Audição; Percepção Auditiva*.

Resumen

Síndrome del Ojo de Gato es un síndrome poco frecuente, caracterizada por coloboma de iris y malformación pre-auricular. El objetivo de este trabajo es presentarla evaluación fonoaudiológica y de procesamiento auditivo de dos hermanos con un diagnóstico de síndrome de Ojo de Gato. Las evaluaciones se realizaron en el ambulatorio de neuroaudiologia y e nel ambulatorio de tamizaje fonoaudiológica del Departamento de Fonoaudiología de la Universidad Federal de São Paulo. Las pruebas fueron: audiometría de tono, investigación del umbral de recepción del habla, imitaciometria e investigación del reflejo acústico, índice promedio de reconocimiento de voz, habla en ruido con figuras, dicótica de dígitos, detección de intervalos aleatorios, prueba de vocabulario y fonología de la prueba de lenguaje infantil y una prueba que evalúa la comprensión oral de la Evaluación Neuropsicológica Luria Nebraska. En la evaluación, los niños presentaron una pérdida auditiva neurosensorial de grado mínimo bilateral, cambio en las habilidades auditivas de cierre y figura-fondo en el procesamiento auditivo y en las habilidades de lenguaje de comprensión oral y emisión oral en las pruebas de vocabulario y fonológicas.

Palabras clave: *Fonoaudiología; Audición; Trastornos de la Audición; Percepción Auditiva.*

Introducion

The Cat Eye Syndrome[1] (Cat Eye Syndrome - CES; OMIM #115470) was first described in 1965 by Schachenmann et al. The name "cat's eye" was adopted because the iris coloboma is similar in shape to the pupil of cats. CES is a rare syndrome; its diagnosis is based on the presence of a supernumerary marker derivative chromosome from chromosome 22. The extra chromosome usually emerges from a new mutation from parents. It is a chromosomal disorder in which transmission is not related to sex, examination of karyotype should be performed if a parent presents the syndrome, pre-auricular malformation or downward palpebral fissures .

This supernumerary marker chromosome is isodicentric with a single active centromere, and occurrence of satellites at queue presenting partial tetrasomy 22-pter to 22q11.2. An estimated incidence of 1: 50,000 to 150,000 live births [2] .

The clinical status is characterized by downward oblique palpebral fissures , ocular coloboma , unilateral or bilateral corneal opacity, cataract, anorectal and kidney malformations (absence of one or both kidneys, hydronephrosis, supernumerary kidney or renal hypoplasia), heart malformation (especially the totally anomalous pulmonary venous return- TAPVR and tetralogy of Fallot - TOF), preauricular depressions or folds, sometimes in combination with atresia of the external auditory canal and mental retardation. Only 41% of CES patients have a classic combination of iris coloboma, anal anomalies and pre-auricular anomalies [2,3].

Other abnormalities may occur and are listed in decreasing frequency of occurrence: anal atresia with rectal fistula to the bladder, vagina or perineum, microphthalmia (often unilateral), craniofacial malformation including choanal atresia, skin patches, growth hormone deficiency, cleft lip and palate, vertebral fusions, biliary atresia, among others that are less common [3].

During early childhood it is common for individuals to die of multiple malformations, life expectancy of the remaining is not significantly reduced [1,4].

The CES is associated with wide phenotype variability due to possible mutations arising from the presence of the marker chromosome and can have variations to almost normal phenotype with severe abnormalities. However, none of the features is consistently present which affects the diagnosis and leads to failure in identifying patients with CES [2,3].

The literature reports that individuals with this syndrome may have a cognitive delay [4], therefore, it is considered important to investigate individuals with Cat Eye Syndrome, aspects related to peripheral hearing, to language and to auditory processing in behavioral tests, for a better prognosis.

The objective of this study was to report the speech, language comprehension and auditory processing of two siblings diagnosed with Cat Eye Syndrome

Description of the case

This retrospective study was submitted to the Research Ethics Committee and approved. The evaluations of two siblings with CES were performed in a neuroaudiological clinic and a language assessment clinic of the Universidade Federal de São Paulo.

The mother of the patients sought a speech therapy service with the complaint that R.L.S., her 8 year-old son, and A.B.S., her 6 year-old daughter had learning difficulties. Both were diagnosed in a genetic clinic by means of a karyotype showing supernumerary marker identified as chromosome 22. The children were then referred to sectors specialized in Auditory Processing Evaluation and Language Assessment.

The subjects underwent pure tone audiometry, speech reception threshold, imittance and acoustic reflex in a soundproof booth, with an audiometer and an impedance meter, calibrated according to the technical standards ANSI, S3 1, 1991 (soundproof booth): ANSI. S3 21, 1978 (pure tone audiometry): ANSI, S3.39-1997 (impedance meter) [5].

The selected tests were: speech recognition percentage index (SRPI) [6], Speech in Noise with pictures (SNT) [6] to assess auditory closure, Dichotic Digit (DDT) [6] for figure-ground, Random Gap Detection Test (RGDT)[7] to assess temporal resolution and a Protocol of Language assessment [8,9,10].

Language Assessment involved: Oral Expressive vocabulary and phonology with ABFW [8] and oral story comprehension and reproduction

"The crow and the doves" extracted from Luria-Nebraska Neuropsychological Battery [9,10].

The boy presented phenotypical signs such as malformation of the outer ear and dyspraxia and history of meconium. He has also been diagnosed with a light form of Autism Spectrum Disorder. In the audiological evaluation, the boy presented bilateral minimal sensorineural hearing loss, type A tympanometric curve and bilateral absence of acoustic reflexes. In the auditory processing evaluation, the results were indicative of impairment of closure abilities, figure-ground and temporal resolution (table 1). His communication was predominantly verbal. As to oral story comprehension of "The crow and the doves ", he answered 06/07 questions including inference. He under-performed in the expressive vocabulary task correctly naming only 48% (57/118) of the pictures of different categories: 4/15 animals, 9/15 food, 6/10 clothing, 8/11 means of transport; 15/24 furniture and tools; 3/10 professions; 1/12 locations; 7/10 shapes and colors; 4/11 toys and musical instruments. As to phonological aspects the following processes were observed: simplification of consonant cluster and simplification of final consonant. In addition, there was assytematic liquid simplification. In imitation test the same processes were observed. He is able to read a little but does not always associate phonemes to graphemes. He only writes his own name and that of different buses

TABLE 1. INDIVIDUAL PERFORMANCE IN BEHAVIORAL TESTING

Behavioral Tests	Male child	Female child
Speech in noise (figures)	Right ear: 90% Left ear: 70% Altered	Right ear: 70% Left ear: 80% Altered
Digit Dichotic	Right ear: 75% Left ear : 60% Altered	Right ear: 92,5% Left ear : 47,5% Altered
Random Gap Detection Test	Did not understand the test (data were inconclusive)	15 milisseconds Normal

The girl is eutrophic, showed phenotypic alteration such as hand, outer ear malformation, and anal atresia, and a history of motor and speech development delay. In the audiological evaluation, the girl presented bilateral minimal sensorineural hearing loss, type A tympanometric curve and bilateral absence of contralateral acoustic reflexes. The auditory processing evaluation showed poor performance on two tests, a sign of impaired closure ability and figure-ground (Table 1). As to oral reception, the girl was able to answer 4 out of 7 questions related to "the crow and the doves" showing difficulty in attention and auditory memory. The evaluation of productive vocabulary (ABFW) resulted in 64% (76/118) correct naming of pictures of different categories: 12/15 animals; 18/24 furniture and tools; 09/10 shapes and colors and 8/11 toys and musical instruments; 6/10 clothing; 11/15 food; 07/11 means of transportation; 3/10 professions; 2/12 locations. In addition, as to phonological aspects, plosive and fricative devoicing was observed, no longer expected at her age. She was able to read roughly 20 words per minute

Discussion

The results obtained in auditory processing and language assessments were critically analyzed, emphasizing the aspects observed.

The speech in noise test, which evaluates the closing ability, is impaired in both individuals, so there is a difficulty in their ability to use extrinsic redundancy, which relates to the speech signal, to fill in the missing parts or distorted audio signal and recognize the full message. It interferes with their

daily activities because often the everyday environment cannot be considered ideal. Impairment of this ability can cause speech understanding deficit in noisy environments, and difficulty reading and writing [11,12].

The results of the dichotic listening test that evaluates the ability of figure-ground were altered in both individuals, so there may be difficulty in understanding speech in an environment like home or classroom with other competitive auditory stimuli which can cause learning disorders. Children in the literacy stage are the most affected as they are in the process of acquiring oral vocabulary and learning to read and write [11,12,13]. The mother of the children reported that the teachers at school often complain about her children's difficulty to learn new information. A gap in Random Detection Test that evaluates the ability of resolution is impaired only in the boy who had great difficulty understanding the instructions. Temporal resolution is important for speech perception, understanding language and learning to read and write [14,15]. Some consonants are characterized by brief time intervals and when there is impairment in temporal resolution the speech of the individual may be affected as is the case of the boy in our study who had consonant cluster and final consonant simplification plus assystematic simplification of liquids.

Neither of the children answered all the questions related to "The crow and the doves" and the difference can be justified by their age. Both were able to provide the resumitive inference, a sign of macrostructural adequacy[16].

As to vocabulary and phonology, it was observed that the performance of the two was below what was expected. According to some authors, children with speech disorders present a deficit of phonological memory and vocabulary [17]. Therefore, our findings are consistent with the literature.

The literature [18] shows that there is a strong correlation between listening, auditory processing and vocabulary. Individuals with auditory processing disorders may present deficit of grammatical morphology, metalinguistic deficit and difficulty in the production of / r / and / l /. Such alterations can be observed in writing, reading comprehension.

The speech therapy for these individuals should include aspects of auditory processing and the use of strategies that involve speech and language. We recommend the counseling of the teachers and of the colleagues to help them cope with the children and their difficulties and to boost their self-esteem. This should result in a more comfortable, humane and inclusive environment for the child.

We would like to stress the importance of a multidisciplinary approach that includes the study of auditory, speech and language aspects, with the contribution of the family, language pathologists, audiologists, psychologists, pedagogues and different clinicians. The focus must be the child and his general well-being and social insertion.

Final considerations

The Individuals with Cat Eye Syndrome of this study had bilateral minimal sensorineural hearing loss, impaired hearing closure and figure-ground and difficulty listening, speaking, and reading.

References

1. Chellapandian D, Schneider A. Anatomical Asplenia na Síndrome do olho de gato: uma expansão do espectro da doença. Caso Rep Pediatr; 2013. 218.124

2.Belangero, S I N et al. Interrupted aortic arch type B in A patient with cat eye syndrome. Arq. Bras. Cardiol. [online]. 2009, vol.92, n.5, pp. e56-e8.

3.Rosa, R F M et al. Clinical characteristics of a sample of patients with cat eye syndrome. Rev. Assoc. Med. Bras. [online]. 2010, vol.56, n.4, pp. 462-6.

4.McKusick V.A, Kniffin C.L. CAT EYE SÍNDROME – CES - # 115470. Associação OMIM. 2006

5.Russo, ICP. & Santos, TMM – A prática da audiologia clínica. São Paulo: Cortez, 1993. 226p.

6.Pereira LD, Schochat E. Processamento auditivo central: manual de avaliação. São Paulo: Lovise; 1997. 231 p.

7.Keith, RW. Random gap detection test. Missouri (USA): Auditec of Saint Louis, 2000.

8.Wertzner HF. Fonologia. In: Andrade CRF, Befi-Lopes DM, Fernandes FDM, Wertzner HF. ABFW: teste de linguagem infantil nas áreas de fonologia, vocabulário, fluência e pragmática. Carapicuíba: J Soc Bras Fonoaudiol; 2000. p.5-40.

9.Golden CJ. Luria Nebraska neuropsychological battery: children's revision. New York. Administration and Scoring Booklet; 1987.

10.Moraes Z, Chiari BM, Perissinoto J. Estilos de linguagem como facilitadores de memória. J Soc Bras Fonoaudiol. 2001; 13(1): 54-61.

11. Jacob LCB, Alvarenga KF, Zeigelboim BS. Avaliação audiológica do sistema nervoso auditivo central. Arq otorrinolaringol. 2000 Out/Dez; 4(4): 144-51.

12. Machado CSS, Valle HLBS, Paula KM, Lima SS. Rev. CEFAC. 2011 Mai-Jun; 13(3): 504-12

13. Lacerda A, Marasca C. Níveis de pressão sonora de escolas municipais de Itapiranga - Santa Catarina. J Soc Bras Fonoaudiol. 2001; 13(2):277-80.

14.Frota S, Pereira LD. Processamento auditivo: estudo em crianças com distúrbios da leitura e da escrita. Rev. Psicopedagogia. 2010; 27(83): 214-22.

15.Santos JLF, Parreira LMMV, Leite RCD. Habilidades de ordenação e resolução temporal em crianças com desvio fonológico. Rev. CEFAC. 2010; 12(3): 371-6.

16.Lira JO, Tamanaha AC, Perissinoto J, Osborn E. O reconto de histórias em crianças do espectro autístico: um estudo preliminar. Rev. CEFAC 2009; 11(3): 417-17.Athayde ML, Carvalho Q, Mota HB. Vocabulário expressivo de crianças com diferentes níveis de gravidade de desvio fonológico. Rev. CEFAC 2009; 11(2): 161-8

18.Chermak GD, Musiek FE. Auditory training: principles and approaches for remediation and managing auditory processing disorders. Seminars in Hearing. 2002; 23(4): 297-30

.