

Auditory processing, reading and writing in the Silver-Russell syndrome: case report

Processamento auditivo, leitura e escrita na síndrome de Silver-Russell: relato de caso

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ABSTRACT

The aim of this study was to describe the speech-language pathology aspects of auditory processing, reading and writing of a male patient diagnosed with Silver-Russell syndrome. With two months of age the patient presented weight-for-height deficit; broad forehead; small, prominent and low-set ears; high palate; discrete micrognathia; blue sclera; cafe-au-lait spots; overlapping of the first and second right toes; gastroesophageal reflux; high-pitched voice and cry; mild neuropsychomotor development delay; and difficulty to gain weight, receiving the diagnosis of the syndrome. In the psychological evaluation, conducted when he was 8 years old, the patient presented normal intellectual level, with cognitive difficulties involving sustained attention, concentration, immediate verbal memory, and emotional and behavioral processes. For an assessment of reading and writing and their underlying processes, carried out at age 9, the following tests were used: Reading Comprehension of Expository Texts, Phonological Abilities Profile, Auditory Discrimination Test, spontaneous writing, Scholastic Performance Test (SPT), Rapid Automatized Naming Test (RANT), and phonological working memory. He showed difficulties in all tests, with scores below expected for his age. In the auditory processing assessment, monotic, diotic and dichotic tests were conducted. Altered results were found for sustained and selective auditory attention abilities, sequential memory for verbal and non-verbal sounds, and temporal resolution. It can be concluded that the patient presents alterations in the learning of reading and writing that might be secondary to the Silver-Russell syndrome, however, these difficulties can also be due to deficits in auditory processing abilities.

Keywords: Silver-Russell syndrome; Language; Hearing; Reading; Manual writing; Craniofacial abnormalities

INTRODUCTION

The Silver-Russell syndrome (SRS) is a genetic involvement described by Silver and Russel in the years of 1953 and 1954, respectively. The diagnostic of this syndrome is carried out based on clinical evaluation, for the etiology is considered heterogeneous by many authors⁽¹⁾.

The diagnostic criteria are the presence of three main signs and one or more secondary signs. The main signs are: low birth weight (retarded intrauterine growth); low stature (retarded

post-natal growth); small triangular face; and clinodactyly of the fifth finger. The most common secondary signs are: macrocephaly, skeletal asymmetry, syndactyly, corners of the mouth turned down, cafe-au-lait spots, precocious puberty, genital abnormalities, delayed development of oral language, feeding difficulties. The SRS may also present other manifestations, such as cognitive impairment, gastrointestinal disorders, aversion to food, cardiac anomalies⁽²⁾, extreme thinness, excessive sweating, and frequent episodes of hypoglycemia⁽³⁾.

The incidence is estimated in 1:50,000–1:100,000 born alive. Close to 19% of the cases present more than one individual affected in the family, providing evidence for a genetic cause⁽²⁾. In a case report⁽⁴⁾, the author quotes that this syndrome is genetically heterogeneous, with different patterns of heritage. For these patterns of heritage, three hypothesis were proposed: recessive autosomal heritage, dominant heritage linked to the X chromosome, and a new mutation of dominant character, located in the region 17q25. The latest is the most accepted heritage pattern⁽⁵⁾. The recent progress in the characterization of the chromosome 7 has defined two candidate regions, 7p11.2-p13 and 7q31-qter⁽⁶⁾.

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There are few studies that focus on the speech-language pathology aspects of the SRS, especially with regards to language, learning and auditory processing aspects. The most frequent manifestations reported are alterations of oral functions, triggered primordially by hypotonia; structural alterations and craniofacial disproportions, with repercussion in speech and swallowing; presence of difficulty in formal learning (reading, writing and math), with need of specialized educational tutoring; oral language alterations⁽⁷⁾ and dysphonia with tendency to high-pitched voice⁽⁸⁾.

Some studies suggest that cognitive impairment is not frequent⁽⁹⁾, while others describe that mild mental deficiency is expected in approximately 25% of the cases⁽¹⁰⁾.

The aim of this study was to describe the case of a patient with Silver-Russel syndrome diagnosis, addressing the speech-language pathology aspects of reading, writing and auditory processing.

CLINICAL CASE PRESENTATION

This study was approved by the Research Ethics Committee of the Bauru School of Dentistry – Universidade de São Paulo (FOB-USP), under protocol number 69/2010. The legal guardian of the patient signed a free and informed consent term consenting with the conduction and dissemination of this research, according to the resolution 196/96.

The patient, male gender, began treatment at the Speech-Language Pathology clinic of the institution at 7 years of age, when his mother presented complaints of literacy difficulties, noted by the school staff.

The clinical history showed that the individual was born full-term (38 weeks), weighing 3,560 kg, measuring 48.5 cm, with head circumference of 36 cm, chest circumference of 35.5 cm, Apgar eight in the first minute, and congenital clubfoot to the left. The patient's mother denies pre-natal complications, however there was a peri-natal complication: the subject presented transitory taquipnea and stayed in the intensive care unit (ICU) for seven days. On the fifth day after his birth, he was diagnosed with laryngotracheomalacia.

The individual was admitted to the institution's hospital with 2 months of age due to feeding and breathing difficulties and laryngeal stridor, when the hypothesis of SRS was purposed. In this occasion the following clinic findings were observed: weight-for-height deficit; large front; small, prominent low-set ears; high palate; mild micrognathia; blue sclera; cafe-au-lait spots; overlapping of the first and second right toes; gastroesophageal reflux; high-pitched voice and cry; mild delay in neuropsychomotor development; excessive sweating during feedings; and difficulty gaining weight.

In a new evaluation, at 3 years and 10 months of age, the patient presented low stature and weight, oblique eyelid fissure, thin upper lip with commissure slightly down, shortened fifth fingers, second toe overlapped by the first toe on his left foot, body asymmetry in the lower limbs, normal cardiac auscultation, and normal results in the neurological evolution exam. Currently, at 9 years of age, the patient is 1.26 m tall, weights 28.3 kg, and has 51 cm of head circumference.

Psychological assessment

The subject was submitted to a psychological evaluation in May 2008, at 8 years of age. He was assessed using the Wechsler Scale of Intelligence for Children (WISC-III), Color Span Test, Infant Behavior Scale A2 (ECI – Rutter), Bender and Raven.

The intellectual level was found within normal parameters, but with cognitive difficulties involving sustained attention, concentration, immediate verbal memory, as well as emotional (anxiety and immaturity) and behavioral (impulsivity) processes.

This assessment also found alterations in auditory memory span for digits and non-verbal logical-temporal sequence.

Reading and writing assessment

The subject presented oral language within normal parameters, with the syntactic, phonological, semantical and pragmatic subsystems preserved.

The reading and writing assessment was carried out in the second semester of 2009, when the child was 9 years old and in the third grade of elementary school. The tests used for this evaluation were: Reading Comprehension of Expository Texts, Phonological Abilities Profile, Auditory Discrimination Test, spontaneous writing, Scholastic Performance Test (SPT), Rapid Automatized Naming Test (RANT), and a phonological working memory task.

During the Reading Comprehension test, the patient read out loud the text "The Giraffe", indicated for children in the third grade, where he didn't respect punctuations, substituted letters and words, added, omitted and transposed syllables and words, and, finally, repeated words and sentences. As for the comprehension of the same text, the child identified the central ideas and was able to locate the comprehension errors, however he was not able to establish thematic continuity between ideas, as well as he was not able to identify alone the type of organization of the information in the text, to use his previous knowledge to infer the non-explicit information, nor to construct a vision of reality based on previous knowledge.

In the phonological awareness test, the analysis, addition, segmentation, subtraction, substitution, rhyme, and syllable reversion subtests were evaluated, obtaining a total score of 53 points, which is considered below the expected for the patient's chronological age.

The patient's performance on the Auditory Discrimination Test was of 95% of correct answers (34 out of 36 proposed pairs), which is considered satisfactory for his age.

In the Scholastic Performance Test, the reading, arithmetic and writing subtests were evaluated, obtaining a score below the expected for his educational level.

During the Rapid Automatized Naming Test the patient presented naming speed for colors, letters and digits below the expected for his educational level; on the other hand, his naming speed for objects was compatible with the expected for his educational level.

The theme chosen by the subject for spontaneous writing was a letter to the director of a television channel, regarding

a program of his interest. The patient presented maintenance of the theme and establishment of temporal links, however the text presented was a short narrative of four sentences, with absence of punctuation.

In the phonological working memory task, the subject scored 70 points for pseudowords, 20 points for digits in direct order, and four points for digits in reverse order, which is considered below the expected for his chronological age.

Auditory processing evaluation

The patient presents normal peripheral hearing and middle ear function, as verified by conventional audiological evaluation. He did not present hearing complaints and/or upper airway complications when the evaluation was performed, as well as any difficulty to understand the tests.

The tests used for auditory processing evaluation were:

- Diotic tests: sound localization test (SL), verbal sequential memory test (VSMT), non-verbal sequential memory test (NVSMT), Random Gap Detection Test (RGDT), and Sustained Auditory Attention Ability Test (SAAAT).
- Monotic tests: Pediatric Speech Intelligibility Test with ipsilateral competitive message – synthetic sentences and word recognition (PSI/ICM).
- Dichotic tests: Pediatric Speech Intelligibility Test with contralateral competitive message (PSI/CCM), Staggered Spondaic Word Test (SSW), Dichotic Digits Test (DD), and Non-verbal Dichotic Test.

The tests SL, VSMT and NVSMT were performed in free field, while the other tests were carried out within a soundproof booth with a two-channel Midimate 662 audiometer, linked to a CD player.

The sound localization test (SL) evaluates the ability to localize sounds, and its objective is to provide information about the physiological hearing mechanism of discrimination of the sound source⁽¹¹⁾. The VSMT and NVSMT tests had the aim to obtain information regarding the discrimination of sequences of sounds⁽¹¹⁾, in order to evaluate the hearing ability of simple temporal ordination.

The RGDT test is designated to measure temporal resolution by determining the auditory fusion threshold, measured in milliseconds (ms). The PSI test evaluates the ability of figure-ground for verbal sounds⁽¹¹⁾, and has the aim to provide information about the physiological hearing mechanism of recognition of verbal sounds in monotic and dichotic hearing.

Just like the DD test, the SSW test evaluates the ability of figure-ground for verbal sounds in dichotic hearing⁽¹¹⁾. The SSW test also evaluates the complex temporal ordination ability, providing information about the discrimination mechanism of sounds in sequence, when there are inversions in the execution of the task⁽¹¹⁾. The SAAAT test has the aim to evaluate sustained hearing attention, and the Non-Verbal Dichotic Test, to verify selective attention through a binaural separation task.

In the diotic tests, the patient presented results within normal standards only in the sound localization test, being

able to localize the sound source in the five requested positions. In the VSMT test, he responded adequately to two out of three sequences of syllables. In the NVSMT test, he presented great difficulties in the execution of the task. He was not able to perform the demonstration task, nor the test itself. Therefore, he was not able to memorize any of the presentation orders of non-verbal sound.

In the RGDT test, the subject presented difficulties to perform the task, however, it was possible to determine the auditory fusion threshold in 15 ms for the frequency 500 Hz, 25 ms for 2 kHz, and 20 ms for 4 kHz. It was not possible to determine the auditory fusion threshold for 1 kHz. In the SAAAT test, the patient presented a total of 39 errors, 22 being from lack of attention and 17 from impulsivity. The vigilancy decrease was six. Considering the errors analysis, the sustained attention ability is altered.

In the monotic PSI tests, in the signal/noise ratio zero and +5, as well as in the PSI – word recognition with signal/noise ratio zero, -10, -15 and -40, the patient presented scores of 90 and 95%, which is within the normal standards for his age group.

In the dichotic PSI tests – synthetic sentences in signal/noise ratio zero and +5, the child presented scores from 90 to 95%, within the normal standards for his age group. In the SSW, the items hearing effect, order effect, inversions and type A, the errors were not significant. On the other hand, the errors of right competitive and left competitive were significant, with scores of 67.5% and 65%, respectively, while the expected score for both is 90%.

In the Dichotic Digits test the punctuation obtained was 92.5% of correct answers; the minimum expected score is 90%, therefore the subject's performance was adequate for his age group. In the Non-Verbal Dichotic test he presented four errors in the of free attention subtest, while only one mistake is allowed; hence, his performance was below the expected for his age group. In the subtests of directed attention to right and left, he did not present any errors, being, therefore, adequate in this ability.

According to the auditory processing evaluation, the abilities of sustained auditory attention, selective attention in the free attention subtest, sequential memory for verbal and non-verbal sounds, and temporal resolution are altered; meanwhile, the abilities of figure-ground discrimination and binaural integration are adequate.

DISCUSSION

There are very few national or international studies that focus language, learning and auditory processing aspects in SRS, hence we will highlight the relationship between reading, writing and auditory processing in general, which may justify the difficulties presented in this syndrome, also considering the results of the psychological evaluation.

Even though the psychological evaluation is not part of the main objective in this case report, there was a need to describe this aspect due to the relationship between the abilities assessed and the speech-language pathology aspects described.

In both the psychological and the auditory processing assessments, alterations of sustained attention, auditory memory, and temporal-logical sequence for non-verbal sounds were observed, and the diminished concentration may also have interfered in the results of the other auditory processing abilities.

For the accomplishment of reading with adequate comprehension it is necessary to have integrity of the peripheral and central nerves system, as well as certain fundamental pre-requisites, such as selective and sustained attention, auditory discrimination and perception, short and long-term memory, and phonological awareness – abilities assessed in the group of special auditory processing tests⁽¹²⁾.

According to the auditory processing tests used, the child presents alterations of sustained attention, auditory discrimination and perception, short-term memory, and phonological awareness. In the psychological evaluation, the alterations found were of auditory memory with digits, immediate memory, sustained attention, and concentration. These altered abilities are some of the pre-requisites for reading.

Children with learning disorders may have neural hearing disorders. These disorders involve the central auditory nervous system and can be identified, analyzed, and quantified through auditory processing evaluation, besides some aspects of the psychological evaluation.

The brain of individuals with reading difficulties is unable to rapidly process changes and order of acoustic patterns of speech⁽¹²⁾. In view of the fact that the RGDT test is designated to measure temporal resolution, that is, the capacity to detect intervals of time between sound stimuli or to detect the shortest time in which an individual can discriminate between two audible signals, an alteration in this test may result in difficulties to identify short acoustic variations of speech or to interpret a heard message⁽¹³⁾.

In a previous study, it was found that children with reading difficulties had an inferior performance in the temporal resolution test, showing significant correlation between temporal processing and reading abilities⁽¹⁴⁾. Our findings collaborate these studies, since the patient presents alteration in the temporal resolution ability and in

reading and writing, besides an alteration in the non-verbal logical-temporal sequence, observed in the psychological evaluation.

As for the quantitative analyses of the SSW test, poor performance in the right competitive (RC) and left competitive (LC) conditions may suggest left temporal lobe dysfunction, and may be associated to poor phonemic decoding abilities, which can demonstrate poor phonetic abilities that affect reading and spelling⁽¹⁵⁾.

In order to read, the child must acquire a series of cognitive and linguistic-perceptive abilities that include: ability to focus attention and concentration; ability to comprehend and interpret spoken language in daily life; ordination and auditory memory; ordination and visual memory; word processing ability; vocabulary development; and reading fluency. From these abilities, attention, auditory memory, word processing and fluency in reading are altered in the case studied.

The findings of the present investigation suggest that the child with Silver-Russell syndrome showed important alterations in the evaluated auditory and reading and writing abilities, verified by his poor performance in some of the auditory processing tests, as well as in the reading and writing tests and in the psychological evaluation.

Hence, it is evident that reading and writing and auditory processing abilities, as well as the cognitive and emotional processes, are interconnected.

FINAL COMMENTS

In the psychological evaluation the patient presented alterations in sustained attention, concentration and immediate verbal memory. In the speech-language pathology evaluation, he manifested difficulties in phonological awareness, working memory, and RAN, as well as alterations in auditory processing tests.

Thus, the patient presented alterations in learning how to read and write that may be secondary to SRS, however, these difficulties may also be due to auditory processing alterations.

The importance of the auditory processing evaluation is emphasized for the therapeutic process and further studies in this theme.

RESUMO

O objetivo deste trabalho foi descrever os aspectos fonoaudiológicos de processamento auditivo, leitura e escrita de um paciente do gênero masculino com diagnóstico de síndrome de Silver-Russell. Aos dois meses de idade o paciente apresentava déficit pândero-estatural; frontal amplo; orelhas pequenas, proeminentes e com baixa implantação; palato ogival; discreta micrognatia; esclera azulada; manchas café-com-leite; sobreposição do primeiro e segundo artelhos à direita; refluxo gastroesofágico; voz e choro agudos; atraso leve no desenvolvimento neuropsicomotor; e dificuldade de ganhar peso, recebendo o diagnóstico da síndrome. Na avaliação psicológica, realizada aos 8 anos de idade, o paciente apresentou nível intelectual normal, com dificuldades cognitivas envolvendo atenção sustentada, concentração, memória verbal imediata e processos emocionais e comportamentais. Para avaliação da leitura e escrita e de seus processos subjacentes, realizada aos 9 anos de idade foram utilizados os testes de Compreensão Leitora de Textos Expositivos, Perfil das Habilidades Fonológicas, Teste de Discriminação Auditiva, escrita espontânea, Teste de Desempenho Escolar (TDE), teste de Nomeação Automática Rápida e prova de memória de trabalho fonológica. Apresentou dificuldades em todos os testes, estando as pontuações abaixo do esperado para sua idade. Na avaliação do processamento auditivo foram realizados testes monóticos, dióticos e dicóticos. Foram encontradas alterações nas habilidades de atenção auditiva sustentada e seletiva, memória sequencial para sons verbais e não-verbais, e resolução temporal. Conclui-se que o paciente apresenta alterações na aprendizagem da leitura e escrita que podem ser secundários a síndrome de Silver-Russell, porém tais dificuldades também podem ser decorrentes das alterações em habilidades do processamento auditivo.

Descritores: Síndrome de Silver-Russell; Linguagem; Audição; Leitura; Escrita manual; Anormalidades craniofaciais

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