Performance regarding the learning process in a Velocardiofacial Syndrome case

Desempenho nas habilidades subjacentes a aprendizagem em um caso de Síndrome Velocardiofacial

Desempeño en las habilidades que son la base del aprendizaje en un caso de Síndrome Velocardiofacial

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Abstract

Introduction: The Velocardiofacial Syndrome is a genetic condition involving more than 180 clinical manifestations. The presence of learning disabilities is highly reported in this syndrome, and it is considered one of the most challenging manifestations. According to literature, the genetic condition of the syndrome causes neurological disorders that harm the learning process. Therefore, it is essential to investigate which skills are altered and preserved. Objective: To describe the main difficulties and potentialities underlying the learning process in a case of VCFS. Methods: Standardized instruments were applied to assess cognitive-linguistic skills, perceptual-motor skills and executive functions of a male person, 13 years old, who was diagnosed with SVCF. Results: The results indicate deficits in phonological processing, working memory, rhythmic organization, receptive and expressive language, semantics and pragmatics, graphic-motor perception and executive functions related to sustained attention, planning, strategy and cognitive flexibility. Such lags proved detrimental to the use of reading comprehension activities, writing

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and numeric computation. Neuropsychological functions of cutaneous and synesthetic sensations, higher visual functions and numerical dexterity were preserved. **Conclusion:** This study identified impairments in neuropsycholinguistic and neurocognitive skills involved in the learning abilities. Thus, it informs the diagnostic and possibly the therapeutic process of individuals with SVCF.

**Keywords:** DiGeorge Syndrome; learning disorders; speech, language and hearing sciences; neuropsychology.

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**Resumo**

**Introdução:** A Síndrome Velocardiofacial (SVCF) é uma condição genética envolvendo mais de 180 manifestações clínicas. A presença do distúrbio de aprendizagem é altamente reportada nessa síndrome, sendo considerada uma das manifestações mais desafiadoras. De acordo com a literatura, a condição genética da síndrome acarreta em disfunções neurocorticais prejudiciais ao processo de aprendizagem. Sendo assim, torna-se essencial a investigação sobre quais competências encontram-se alteradas e quais preservadas. **Objetivo:** descrever as principais dificuldades e potencialidades em habilidades subjacentes a aprendizagem em um caso de SVCF. **Métodos:** Foram avaliadas, por meio de instrumentos fonoaudiológicos e neuropsicológicos padronizados, as habilidades: cognitivo-linguísticas, percepção-motoras e de funções executivas de um sujeito do gênero masculino, 13 anos de idade, com histórico de prematuridade e intercorrências gestacionais, diagnosticado com SVCF. **Resultados:** os testes demonstraram déficits no processamento fonológico, memória operacional, organização rítmica, linguagem receptiva e expressiva, semântica e pragmática, percepção gráfico-percepto-motora e nas funções executivas relacionadas à sustentação da atenção, planejamento, estratégia e flexibilidade cognitiva. Tais defasagens mostraram-se prejudiciais ao aproveitamento de atividades de compreensão leitora, de escrita e cálculo numérico. As funções neuropsicológicas de sensações cutâneas e sinestésicas, funções visuais superiores e destreza numérica encontram-se preservadas. **Conclusão:** O estudo de caso identificou alterações nas habilidades neurocognitivas e neuropsycholinguísticas implicadas nas competências para a aprendizagem, representando fator contribuinte no processo diagnóstico e terapêutico de indivíduos com SVCF. O sujeito apresenta em seu histórico fatores considerados de risco para os transtornos de aprendizagem, recorrentes na SVCF, e que podem contribuir com os resultados observados.

**Palavras chave:** Síndrome de DiGeorge; transtornos de aprendizagem; fonoaudiologia; neuropsicologia.

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**Resumen**

**Introducción:** El síndrome velocardiofacial (SVCF) es una condición genética que reúne más de 180 manifestaciones clínicas. La presencia de problemas de aprendizaje es muy reportada en este síndrome y es considerada una de las manifestaciones más desafiantes. Según la literatura, la condición genética del síndrome conduce a trastornos neurocorticales que son perjudiciales para el proceso de aprendizaje. Por lo tanto, es esencial investigar cuales habilidades están alteradas y cuales conservadas. **Objetivo:** Describir las principales dificultades y potencialidades en las habilidades subyacentes al aprendizaje en un caso de síndrome SVCF. **Métodos:** por medio de los instrumentos fonoaudiológicos y neuropsicológicos estandarizados, se evaluaron las habilidades: cognitivo-linguísticas, perceptor-motoras y funciones ejecutivas de un sujeto del género masculino de 13 años de edad, con histórico de prematuridad y complicaciones gestacionales, diagnosticado con SVCF. **Resultados:** Las pruebas mostraron déficits en el procesamiento fonológico, la memoria de trabajo, la organización rítmica, el lenguaje receptivo y expresivo, la semántica y pragmática, la percepción gráfico-percepto-motora y las funciones ejecutivas relacionadas con el mantenimiento de la atención, la planificación, la estrategia y la flexibilidad cognitiva. Tales retardos demostraron ser perjudiciales para las actividades de comprensión de lectura, escritura y actividades de cálculo numérico. Las funciones neuropsicológicas de sensaciones cutáneas y sinestésicas, funciones visuales superiores y destreza numérica estaban preservadas. **Conclusión:** El estudio de caso identificó cambios en las capacidades neurocognitivas y neuropsychológicas implicadas en las habilidades para el aprendizaje, lo que representa factor que contribuye en el proceso diagnóstico y terapéutico para las personas con la SVCF. El sujeto presenta en su histórico factores considerados de riesgo para los trastornos de aprendizaje, recurrentes en la SVCF y que pueden contribuir con los resultados observados.

**Palabras clave:** Síndrome de DiGeorge; trastornos del aprendizaje; fonoaudiología; neuropsicología.
Introduction

The Velocardiofacial Syndrome (VCFS), also known as DiGeorge syndrome or 22q11.2 deletion syndrome (22q11.2 DS), is a genetic condition associated with a multisystemic expression that may include: facial alterations, neurological disorders, cardiac alterations, submucous cleft palate, speech and language difficulties, and learning disorders, among others.

22q11.2 DS is the most common microdeletion identified in humans with a population prevalence of about 1:4000 born alive.

This is a syndrome that has a broad spectrum of over 180 clinical manifestations; however, the learning disorders and neuropsychiatric disorders are described as the most challenging manifestations.

This disorder has been reported at an incidence rate that can vary from 80 to 100% in individuals with VCFS.

The microdeletion contains more than thirty genes that are possible candidates for the pathogenesis of 22q11.2 DS. The disease is caused due to the haploinsufficiency (insufficient amount of the gene product, due to the presence of a single allele, rather than two) of one or more genes contained in the chromosomal region 22q11.21. The lack of adequate amounts of these proteins can impact the neurological development stages and affect operation of the mature brain. Thus, the fronto-striatal and frontoparietal neural networks seem to be particularly affected, resulting in poor performance for cognitive level on tasks requiring changes in attention, cognitive flexibility and working memory (frontal cortex and caudate nucleus) and in tasks involving visual-spatial and numerical skills (posterior parietal cortex).

Although learning disabilities are considered to be part of the syndrome symptomatology, determinants factors of diagnosis are disregarded, such as the existence of potential integrity, absence or presence of neurological signs, social history, and also the essential involvement of a multidisciplinary team to diagnose the varied manifestations.

Therefore, it is essential to research which competences are changed and maintained. Objective: To describe the main difficulties and potentialities in skills that underlie learning in a case of VCFS.

According to the literature, the genetic condition of the syndrome leads to important neuro-cortical disorders for learning. The hypothesis of the study is that the studied individual had learning problems due to linguistic and neuropsychological deficits that are part of the picture. The objective of the study is to describe the altered and preserved skills that underlie the learning process in a case of VCFS.

Case report

The study was approved by the Research Ethics Committee of the institution under protocol number 214/2010. Those responsible for the subject of the study signed the Term of Informed Consent, consenting with the release of data for scientific purposes.

The 13-year-old male subject was forwarded for speech pathology and neuropsychological assessment with complaints of poor academic performance.

It is listed in his clinical history that the subject is an only child of outbreed parents, born preterm (32 weeks) by cesarean section and at a low weight (1,850 g). During the prenatal stage, there were reports of gestational hypertension, bleeding and contractions throughout the stage. The psychomotor development of the children was abnormal. He started walking at 1 year and 8 months and the emission of the first words at 3 years of age.

Genetic diagnosis of VCFS was performed at age 7 in the Human Genome Research Center – Sao Paulo University, by means of FISH technique (fluorescence in situ hybridization) and PCR (based on genotypic analysis of polymerase chain reaction). The phenotypic characteristics of VCFS found in the subject were: cleft palate hidden submucosa, velopharyngeal dysfunction, retrognatia, straight facial profile, asymmetrical ears, slightly narrow palpebral fissures, elongated nasal bridge and scoliosis. Audiological testing showed hearing thresholds within normal limits and, by the time of evaluation, cardiac abnormalities have not been diagnosed.

At the time of speech and neuropsychological evaluation, the subject was studying in 7th grade of elementary school at a private educational institution, conducting psycho-pedagogical supervision since the 3rd grade. According to parents, despite the progress observed with the pedagogical intervention, the adolescent school performance
remained unsatisfactory. Still, according to them, there was no family history of syndromes and/or learning problems in the family.

**Speech pathology assessment**

The speech pathology assessment, directed toward aspects of the written language, investigated the phonological awareness skills, phonological working memory, lexical access, speed and reading comprehension, writing and arithmetic. The following instruments were used:

**Phonological awareness: Sequential Assessment Instrument – CONFIAS**

The CONFIAS test assesses the phonological awareness in syllabic (9 subtests) and phonemic levels (7 subtests). The performance analysis is done by comparing the child’s spelling hypothesis with the correct answers on tests. In the subtests of syllabic level, the subject demonstrated difficulty in performing medial syllable identification, rhyme production, exclusion and syllabic transposition, getting 22 hits. For the phonemic level, 14 hits were obtained; difficulties were manifested in the initial and final phoneme identification subtests, exclusion, segmentation and transposition of phonemes.

Considering that the writing of the subject transits in the alphabetical hypothesis, the average values of expected hits are approximately 35 and 20 for the test of syllables and phonemes, respectively. Considering that the right answers obtained are less than expected, there appears to be a lag in phonological awareness skills.

**Rapid Automatized Naming Test – RAN**

RAN was used to evaluate the speed of access to mental lexicon regarding the naming of colors, letters, numbers and objects. The obtained values were: 61’13”, 32’24”, 40’47” and 52’31” seconds to naming colors, letters, digits and objects, respectively. The values obtained for the colors and digits categories were equivalent to the average performance of 1st-grade students. In letters and objects categories, the results are equivalent to the performance of students in 2nd grade. Considering afore mentioned results, as well as that the subject attends the 7th grade, there is a deficit in the speed of access to mental lexicon.

**Phonological working memory - MTF**

The test assesses the phonological working memory for pseudo words (formed by 2 to 5 syllables) and digits in direct and reverse order. Comparing the reference values given by the test (expected performance for children from 6 years old) with the performance of the subject, there is impairment in phonological working memory.

**Reading Comprehension of Expository Texts Test**

In order to assess reading comprehension, the text “Mammals” was used, which is suitable for 7th-grade students. Before he starts reading, the subject has shown interest in the text, reporting facts that indicate prior knowledge of the subject. During silent reading, he showed low concentration and interest, interrupting the reading to make comments not relevant to the text. During oral reading, the subject again demonstrated that he was dispersed, interrupting the reading to manipulate objects. The reading was shaky, slow and predominantly conducted through phonological via. There was no respect to the text punctuation rules, and the child got lost in the lines reading the same paragraph again. After reading, it was found that the subject had not identified the central ideas of the text. He answered the questions asked by the evaluator; however, the answers given were related to prior knowledge of information. Thus, deficits in reading comprehension ability were observed.

**Oral and Silent Reading Speed Test**

The test provides parameters for evaluation of the oral and silent reading speed. The subject performed the reading of 33 words per minute (oral reading) and of 35 words per minute (silent reading). The values obtained are equivalent to the average reading speed of children in the 1st grade. These results show unsatisfactory performance in reading speed.

**Academic performance test – APT**

APT was used to evaluate school performance related to writing, arithmetic and reading skills. The Gross Score expected for students over 12 years in the tests of writing, arithmetic, reading and total
are, respectively: 30, 23, 68 and 119 points. The results obtained by the subject evaluation were as follows: 18 points (writing), 15 (arithmetic), and 61 (read), totaling 94 points. Thus, it appears that the performance skills in writing, reading and arithmetic are unsatisfactory. The score obtained is equivalent to the performance of a child who is 9 years old. In writing skill, spelling mistakes due to omission of letters, support in orality, voiced/unvoiced phonemes sound exchanges and the possibility of multiple representations were found.

TABLE 1. Performance of the individual with VCFs in speech pathology assessment regarding written language

<table>
<thead>
<tr>
<th>SKILLS</th>
<th>EXPECTED</th>
<th>OBTAINED</th>
</tr>
</thead>
<tbody>
<tr>
<td>PHONOLOGICAL PROCESSING</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Phonological Awareness</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(CONFIAS Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Syllables</td>
<td>22p</td>
<td>35.8p</td>
</tr>
<tr>
<td>Phonemes</td>
<td>14p</td>
<td>20.6p</td>
</tr>
<tr>
<td>Rapid Naming (RAN Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Colors</td>
<td>61.13s</td>
<td>36.42s</td>
</tr>
<tr>
<td>Numbers</td>
<td>40.47s</td>
<td>22.92s</td>
</tr>
<tr>
<td>Letters</td>
<td>32.24s</td>
<td>21.98s</td>
</tr>
<tr>
<td>Objects</td>
<td>52.31s</td>
<td>46.74s</td>
</tr>
<tr>
<td>Working Phonological Memory (WPM Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pseudo words</td>
<td>60p</td>
<td>69.4p</td>
</tr>
<tr>
<td>Numbers DO</td>
<td>7.0p</td>
<td>13.8p</td>
</tr>
<tr>
<td>Numbers RO</td>
<td>3.0p</td>
<td>6.2p</td>
</tr>
<tr>
<td>WRITING</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Word writing (APT Test)</td>
<td>18p</td>
<td>30p</td>
</tr>
<tr>
<td>READING</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Word reading (APT Test)</td>
<td>61p</td>
<td>68p</td>
</tr>
<tr>
<td>Oral reading speed</td>
<td>33ppm</td>
<td>97ppm</td>
</tr>
<tr>
<td>Silent reading speed</td>
<td>35ppm</td>
<td>107ppm</td>
</tr>
<tr>
<td>Text comprehension (Reading Comprehension Test)</td>
<td>1p</td>
<td>5p</td>
</tr>
<tr>
<td>ARITHMETIC</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mathematical operations</td>
<td>15p</td>
<td>23p</td>
</tr>
</tbody>
</table>

Legend: p=points, s= seconds, wpm= words read per minute, DO= numbers in direct order and RO= numbers in reverse order
Neuropsychological assessment

Neuropsychological assessment investigated the intellectual skills, cognitive-linguistic, perceptual-motor and executive functions. The following instruments were used:

**Wechsler Intelligence Scale for Children-WISC-III**¹¹

The tool was used to assess cognitive abilities (intellectual capabilities for learning). Classified in general scales (TIQ = 73) and verbal (VIQ = 79) with borderline weighting, executive exams were more affected (EIQ = 61), with intellectually disabled weighting. The factorials index related to verbal comprehension (IQ = 87) showed up in middle-lower levels. The attentional index and information processing indicated borderline profiles for development, and the perceptual organization pointed to deficient category. The accentuated difficulties in cognitive performance tasks, combined with changes in the organization of perceptual visual information, were suggestive of dysfunction in the processing of parietal-occipital-temporal associative areas, which are important to the mastery of skills needed for learning reading, writing and arithmetic. These performances were lowered on the used scale.

**Progressive Matrices - General Scale**¹²

The logical reasoning capabilities to solve space-time problems had classification percentile V in the intellectually deficient category, representing limitations to establish analogies and others’ space, logical and temporal relations in non-verbal activities.

**Wisconsin Card Sorting Test - WCST**¹³

This instrument assesses executive functioning. It was found that the abstract reasoning capabilities and the ability to modify the cognitive strategies in response to changing environmental contingencies, corresponding to executive functions, were impaired in error detection, in sustaining attention and in the productivity of task.

**Bender-Gestalt Test**¹⁴

This procedure evaluates the perceptual-motor maturation by analyzing tracings/drawings. The graphic perceptual-motor assessment revealed results below the age of 6 years, so the maturity of the subject to perform graphics tasks, such as writing, did not correspond to their chronological age, with losses mainly in building angles and spatial orientation (Table 3).

**Simplified Neuropsychological Examination-BANI-TS**¹⁵

The test was used in the investigation of neuropsychological functions. Satisfactory results were found to be related to motor functions of the hands, to the cutaneous and kinesthetic sensations, to the higher visual functions and numerical dexterity. The difficulties highlighted were related to visual and auditory memory short-term processes, rhythmic organization, cognitive-linguistic functions (pragmatic), receptive and expressive language, and reading and writing skills.

Considering that the cutoff point of average accomplishment occurs at 70-80 points, 60% of the tasks had poorer performance, suggestive of evolitional dysfunctions of processing, integration and regulation, which are necessary for learning.

In the sphere of social and moral understanding, the subject showed satisfactory resources, as well as intentional and collaborative behavior during evaluation.
TABLE 2. Performance of the individual with vcfs in neuropsychological assessment

<table>
<thead>
<tr>
<th>NEUROPSYCHOLOGICAL ASSESSMENT</th>
<th>COGNITIVE SKILLS</th>
<th>CLASSIFICATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intellectual capabilities</td>
<td>Total IQ =73p</td>
<td>Borderline</td>
</tr>
<tr>
<td>(WISC-III Test)</td>
<td>Executive IQ =79p</td>
<td>Borderline</td>
</tr>
<tr>
<td>Verbal IQ =61p</td>
<td></td>
<td>Borderline</td>
</tr>
<tr>
<td>Logical reasoning</td>
<td>Logical reasoning to solve</td>
<td>Percentile V (intellectually disabled)</td>
</tr>
<tr>
<td>(Progressive Matrices)</td>
<td>spatio-temporal problems</td>
<td></td>
</tr>
<tr>
<td>Executive functions</td>
<td>Abstract-reasoning capabilities and ability to modify the cognitive strategies</td>
<td>Loss on error detection, in support of the attention and task productivity.</td>
</tr>
<tr>
<td>(WCST Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Neuropsychological functions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(BANI-TS Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Motor functions of the hands</td>
<td>Satisfactory</td>
<td></td>
</tr>
<tr>
<td>Cutaneous and kinesthetic sensations</td>
<td>Satisfactory</td>
<td></td>
</tr>
<tr>
<td>Higher visual functions</td>
<td>Satisfactory</td>
<td></td>
</tr>
<tr>
<td>Numerical dexterity</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Visual short-term memory</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Short-term auditory memory</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Rhythmic organization</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Cognitive-linguistic functions (pragmatic)</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Receptive and expressive language</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Reading and writing</td>
<td>Altered</td>
<td></td>
</tr>
<tr>
<td>Perceptual-motor (Bender Test)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Angles</td>
<td>Obtained/Expected</td>
<td></td>
</tr>
<tr>
<td>Spatial orientation</td>
<td>Performance related to age 6</td>
<td></td>
</tr>
<tr>
<td>Relative position</td>
<td>5p/11p</td>
<td></td>
</tr>
<tr>
<td>Global score</td>
<td>19p/33p</td>
<td></td>
</tr>
</tbody>
</table>

Legend: IQ= intellectual quotient, p=points

Discussion

It is important to consider that the studied patient has in his history important risk factors for learning problems such as maternal hypertension, bleeding and contractions during pregnancy, prematurity and low birth weight, which can be associated with manifestations of the clinical condition. In the literature, there were several studies linking these risk factors with learning disabilities. It is worth quoting two systematic reviews that have made the analysis a large part of these studies. In the study of Rodrigues, Melo and Fonseca\textsuperscript{16, 18} studies were analyzed that found worse academic performance of low birth weight compared to children born at full term, with evidence for an association between low birth weight and cognitive impairment. Moreira, Magellan and Alves\textsuperscript{17} sought the association between prematurity with motor development, as well as behavioral and academic performance. The study included\textsuperscript{33} researches, and only four did not find any effect of prematurity on the results. Thus, the authors concluded that premature children are more susceptible to alterations in motor development, behavior and academic performance when compared with full-term infants.
Regarding maternal hypertension, Chaim, Oliveira and Kimura\textsuperscript{18} conducted a study with newborns and found that maternal diastolic blood pressure greater than 110 mmHg showed a significant association with low birth weight and prematurity. On the other hand, the longitudinal study conducted by Dias et al.\textsuperscript{19} with 30 newborns, children of mothers who had high blood pressure, found that 30% were premature and 17% were small for gestational age. All the children were reassessed between 7 and 15 months and demonstrated normal neurological examination and psychomotor development, not allowing any correlation with the risk indicators.

Regarding individuals with VCFS, in a study case performed with six North American patients, it was verified that the history of prematurity, delay on the pre and postnatal development and neuropsychomotor development delay were prevalent among patients because five of them were born premature and all of them had low birth weight. Still, all patients had restriction on postnatal growth. The global developmental delay and/or intellectual disabilities were found in four out of six patients. One patient had language-borderline development, and only one showed normal development.

The authors reported that, although the research is one cohort study with a small sample size, the probability that prematurity among patients has happened randomly is about 12.7%, i.e., much lower than that found in the study. They also argue that the distal deletion of 22q11 region, which is located in the MAPK1 gene, may be associated with the development of the placenta, leading to restriction of intrauterine growth and low birth weight. Thus, the study states that although the risk factors can be related to learning problems, many of these factors can actually be conditions of their own genetic syndrome.

This study does not intend to determine the profile of the learning disorder in VCFS because the restricted number of subjects (case report) would not allow such a finding. It sought to highlight the findings of a multidisciplinary assessment, since no studies were found associating speech and neuropsychological assessment in cases of VCFS. The searches were conducted in the Lilacs, Scielo, Pubmed and Web of Knowledge databases until March 2014.

Alterations in phonological awareness have been described in a Brazilian study carried out with individuals with clinical signs of VCFS. Such individuals showed alterations in both syllabic phonemic levels in phonological awareness tests\textsuperscript{21}.

The investigation of verbal working memory (phonological) in children and adults diagnosed with VCFS demonstrated that there is impairment in information processing involving short-term memory. The study that investigated the relationship between VCFS and working memory reveals that the performance of these individuals is significantly worse compared to the control group\textsuperscript{22}.

These results corroborate the findings of this study, as in speech pathology assessment. Deficits were also observed in the phonological awareness skills and phonological working memory.

A single publication was found\textsuperscript{23} describing the performance speed of access to mental lexicon in VCFS. The authors of the above-quoted article also criticized the lack of studies addressing the performance of access to mental lexicon in this syndrome. In the above-mentioned publication, children with VCFS had very low error rates (<1%) in the RAN test tasks. Thus, there was no statistically significant difference between children without alterations and with VCFS. These data differ from those found in the current study, which pointed to performances below average in both correct answers and in the speed of access to mental lexicon.

International studies report that children of school age with VCFS show remarkable deficits in mathematics when compared to reading and writing skills that usually are preserved \textsuperscript{23,25}.

However, in a Brazilian study\textsuperscript{24} conducted with a sample of 35 individuals ages 6 to 24 years, the written language was significantly compromised, since all of them showed alterations, according to the used criteria analysis. The investigation of the incidence of learning problems in VCFS reveals the presence of learning disorders in 77.1% of the subjects and reading and/or writing difficulty in 22.9%\textsuperscript{24}.

The arithmetic difficulties in children with VCFS are noted in numbers comparison tasks, performing calculations and solving problems\textsuperscript{25}. The difficulties in this type of activity were also found in the subject of this study.

In another case study carried out with an individual who was 11 years old and diagnosed with VCFS, the tests of reading, writing and arithmetic (APT) were also applied. The results were similar
to those observed in the present study, in which underperformance was found in arithmetic, reading and writing skills when compared to educational attainment.

One of the challenging features of 22q11.2 DS is the cognitive profile, which shows great variability in the impairment level. Studies show that cognitive skills (intellectual capabilities for learning) can be preserved or even indicate intellectual disabilities.

However, there seems to be a consensus between the VIQ discrepancy (verbal intelligence quotient) and EQI (executive intelligence quotient), with higher losses in the EIQ.

The superiority of VIQ over EIQ's performance is common in children and adolescents with the syndrome, with a recorded difference of 11.3 points (cutoff point for clinical significance) between the coefficients.

In the present study, in the tests of cognitive skills, the subject showed better results in verbal activities (VIQ = 79) with borderline weighting, compared to executive tests (EIQ = 61) whose weighting was intellectually deficient. Such findings, regarding the intellectual quotient, are also reported in the literature.

Some neuropsychological characteristics have been identified as impaired in this population, such as the verbal quotients, visual-spatial and motor skills, visual-spatial short- and long-term memory, and executive functions.

The neuropsychological functions most affected in the subject were related to the rhythmic organization, short-term memory, receptive and expressive cognitive-linguistic functions, executive functions and reading-writing skills, corroborating the findings in the literature.

As previously discussed, the microdeletion in the region q11.2 of one allele on chromosome 22 affects the development and functioning of cortical brain regions. It is important to consider that the patient studied here presents in his history important risk factors for learning problems (prematurity, maternal hypertension, bleeding and contractions during pregnancy), which can be aggravating and causing manifestations of this condition; however, such risk factors do not justify all the difficulties encountered because, primarily, there is a genetic mutation that leads to alterations in the development of the cortical areas of the brain, which are important for learning.

Considering the aforementioned results, disturbances in neuropsycholinguistic aspects, which are important to the learning process, were found.

Recent research carried out with 60 Norwegian subjects with VCFS revealed that only one individual did not need academic support during the school years. The others needed or still need special teachers or attend support classrooms to deal with difficulties in math, reading and writing.

Given this background, the evaluation of the skills related to learning in individuals with VCFS is essential, because the diagnosis and early intervention are key factors to ensure better prognosis. The interdisciplinary study (phonoaudiological and neuropsychological) of neurogenetic syndromes can enable an increase in understanding the brain-behavior relationship and also the development of more specific interventional proposals.

**Conclusion**

The results of speech pathology and neuropsychological assessment showed deficits in the aspects related to phonological processing, speed and comprehension of reading, writing and arithmetic, resulting from alterations in the development of perceptual-motor skills, psycholinguistic and executive functions in sustenance of selective, and alternating attention. Some neuropsychological functions are preserved, such as the motor functions of hands, cutaneous and kinesthetic sensations, visual functions and numerical dexterity. These skills are important ways of learning, and the multi-sensory teaching may be valued and used as a pedagogical and therapeutic resource. One should take into account that the subject of the study presents in his history significant risk factors for learning disorders that are recurrent in VCFS and may contribute to the observed results.

**References**

Performance regarding the learning process in a Velocardiofacial Syndrome case

ARTICLES


