
OPTIZ SYNDROME: AUDIOLOGICAL, SPEECH AND NEUROPSYCHOLOGICAL ASSESSMENT

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ABSTRACT

Optiz Syndrome, also named Hypertelorism-Hypospadias Syndrome, is determined by a dominant autosomic genetic condition linked to X chromosome. This syndrome causes anatomic and neurophysiological malformations and secondary impairments in different levels of development. The objective of this study was to analyze the speech, audiologic and neuropsychological aspects of a patient carrier of Optiz Syndrome, comparing the clinical and instrumental findings to the ones in the literature known. The subject focused in the research was R.A.C., a 16 year-old male attending the fourth grade of primary school. The results obtained highlight satisfactory performances concerning remote memory, opticum-spatial organization of the motor act, as well as complex forms of praxias. Maturational deficits were observed in the cognitive tasks related to the motorial and perceptual organization of the intellectual operations in space, pragmatic language and semantics. Handicaps in specific areas of reading, writing and numeric were significantly determining for the diagnosis of learning disorders.

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INTRODUCTION

The Opitz Syndrome or Hypertelorism-Hypospadias Syndrome, was described in 1969 by J.M. Opitz and associated (OPITZ, 1987).

The syndrome includes two entities, the BBB Syndrome and the BBB/G Syndrome, which are the initials of the families that first described the syndrome, also known as BBBG Opitz Syndrome (OPITZ, 1987).

Initially, it was considered as a separate syndrome described by members of a same family. The Opitz Oculogenitolaryngeal Syndrome is an autosomal dominant congenital and rare syndrome (CHRISTOULOU, 1990).

The clinical characteristics are hypertelorism and hypospadias, strabismus, prominent nose tip (flat in the G variety), anteverted nostrils, flat philtrum, cleft lip and palate or submucous cleft, ogival palate, micrognathia, rotated ears with helix malformation (MACDONALD, 1995).

The syndrome also includes cranial asymmetry, epicanthus, mongoloid or anti-mongoloid lid cleft, multiple teeth anomalies, shortened lingual frenum, bifid tongue, bifid uvula and anomalies of larynx, trachea and esophagus. Other features are mental retardation, rectus muscle diastasis, lipomas. Mild joint hyperlaxity and visceral anomalies such as urethral stenosis, inguinal hernia, bifid scrotum, cryptorchidism, imperforate anus, ectopic anus, retro-urethral fistula, duodenal stenosis, mesocardia, interatrial communication. Less frequent are multiple cerebral malformations: agenesis or hypoplasia of the corpus callosum, cerebellar agenesis, Dandy Walker agenesis, hydrocephalia, dilatation of the cerebral ventricles, dysplastic ears, bone anomalies such as clinodactyly, syndactyly in feet and other organs anomalies as lobulated lungs, lung hypoplasia, Meckel diverticulum, umbilical hernia, absence of gall bladder and bifid urethras. Platelet dysfunction, anosmia and osteopenia, bowel and kidney malformation (LEICHTMAN, 1991; MACDONALD, 1995).

The craniofacial malformation present in the Opitz Syndrome makes difficult the air flow favoring the occurrence of night apnea and predominance of an oral breathing pattern (MACDONALD, 1995).

In 1991, Leichtman et al suggested that the gene responsible for the Opitz Syndrome is located in the duplication of the region between 5p12 and 5p13, since children with duplication in the region p12 showed more alteration than that without duplication. Later, molecular genetic studies showed that the syndrome has a heterogenic disorder

TABAQUIM, Maria de Lourdes Merighi et al. Opitz Syndrome: audiological, speech and neuropsychological assessment. *Salusvita*, Bauru, v. 25, n. 3, p. 413-421, 2006.

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located at the chromosome 22 in the region q 11.2 9,10 and in the chromosome X in the region p 22 11,12 (CHRISTODOULOU, 1990; MACDONALD, 1995).

Light mental retard is common and severe retardation is quite rare (EINFELD, 1987; GUION, 1992).

Richieri and Guion (1992) reported five male children with delay in the neuro-psycho-motor development and mild mental retard and one case of a male infant with no delay in the neuro-psycho-motor development but showing behavior problems, hyperactivity. However, he was able to read short sentences.

A study by MacDonald et al. (1995) describes the development of a female that seated by 16 months, walked at two years (with motor discordination) and spoke short sentences as 2 years old. She went to special school since the 1st grade and at age 17th she made and evaluation that revealed a IQ of 76.

The process of language acquisition and development depend on neurological, genetic, emotional, social and familial factors. Alterations in these factors can compromise the neurological development leading to disturb in the oral and writing language (MYSAK, 1988).

Frequently, phono audiologic alterations (language/learning, speech, hearing and swallowing) are part of the clinical spectrum of many genetic syndromes with abnormal structures in the Central Nervous System.

The main challenge to health and education professionals has been the search for the recognition of physical, behavioral, cognitive and language characteristics that allows the diagnose, the adequate intervention and the determination of prognosis in genetic syndromes. Therefore, interdisciplinary investigations should be encouraged to detect individual differences in what regards cognitive and linguistic performance in children with this syndrome.

CASE REPORT

a) Relevant information on the anamnesis

It is reported the case of R.A.C, male, 16 years and 3 months old with a dignosis of Opitz Syndrome. The mother reports pre-natal follow-up with used of drugs to blood hypertension during the pregnancy. The birth was at term by cesarian. The child showed icteritia, generalized hypotonia and delayed reflexes.

Physical examination revealed malformation related to peno-scrotal hypospadias, anteriorized anus and biphid scrotum, micrognatia,

ogival palate, lower implantation of ears and mielomeningolece at the thoracic collum.

He showed cardiopaty, compromise of the upper airways and lungs, bronchitis and repetition pneumonia, miopaty and hypospadias. There were no references for familial antecedents of pathologies related to epilepsia, mental retard or pshychiatric disease.

b) Relevant information on the evaluations

Neuro-psychological evaluation: it was investigated the cognitive, perceptive- motor and behavior levels. It was used the Wechsler Intelligence Scale for Children (WISC) (WESCHLEY, 1945) and the Raven Test for Progressive Matrixes (1979) in order to obtain the quoscient for mental performance. The results were below the expected for the age in the test that required abstraction capacity, logical-spatial reasoning and analogic relation involving social critical judgment. There was also hazard in the visual identification of familiar objects, capacity to isolate and identify essential from non-essential characteristics and in tasks that required attention and concentration. The best cognitive performance were related to numeric abilities, auditory short run memory and visual perception for absent detail (FIGURE 1).

TABAQUIM, Maria de Lourdes Merighi et al. Optiz Syndrome: audio-logical, speech and neuropsychological assessment. *Salusvita*, Bauru, v. 25, n. 3, p. 413-421, 2006.

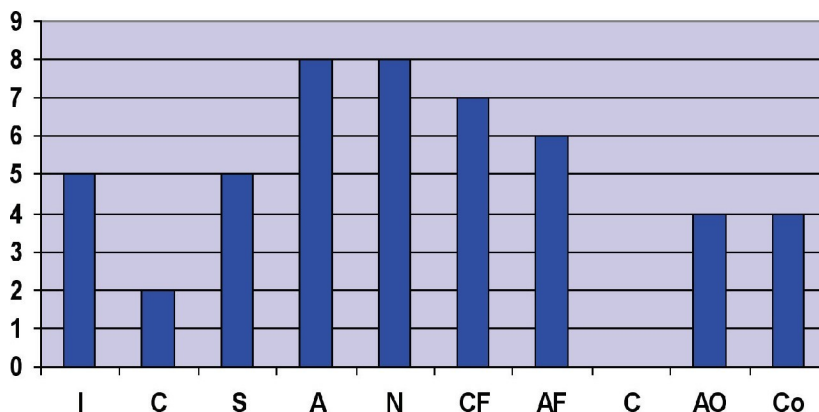


FIGURE 1 - Performance in the WISC test.

In the evaluation of the performance in the visual-motor perception it was used the Bender Santucci (BENDER, 1964), showing difficulties in the activities that required coordination and perception of form, with deficit of prox. 5 years between the graphic-perception-motor age and the chronologic age. For the evaluation of the cortical function it was used the Luria-Nebraska

TABAQUIM, Maria de Lourdes Merighi et al. *Optiz Syndrome: audiological, speech and neuropsychological assessment. Salusvita*, Bauru, v. 25, n. 3, p. 413-421, 2006.

Battery (LNB) (CIASCA, 1994) and the Neuro-psychological Exam (TABAQUIM, CIASCA, 2001), which result were related to the perceptive difficulties, to the expressive and receptive language, to the short run memory, motor difficulties in the left unilateral hand and abstract reasoning (FIGURE 2).

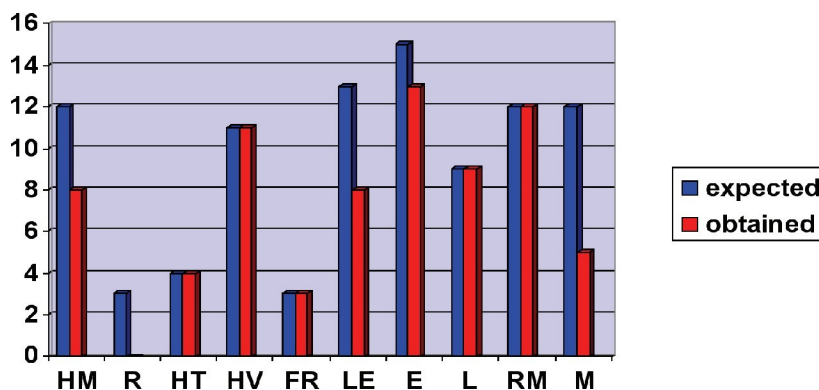


FIGURE 2 - Comparizon among the expected performance and the obtained performance in the Luria Nebraska Battery.

Significant difficulties were observed in the performance of motor tasks, such as the use of associated and dissociated movements, in the acoustic driving organization of the rhythm structures involving more than one stimulus and of the receptive language in the understanding of the logic and thematic grammatical structures.

Phono-audiologic evaluation: the clinical phono-audiological evaluation was made in three parts: oral language evaluation, written language (reading and writing) and speech evaluation.

In oral language the communication ability was evaluated in what concerns the semantic and pragmatic aspects through spontaneous and directed speech, without alteration. For the phonologic aspects it was used the Phonologic Evaluation proposed by Yavas et al. (1992), which verified the adequation of the phonologic and syllabic procedures in the oral language.

For the writing language it was used the Reading and Writing Test (PINHEIRO, 1994) showing greater mistake prevalence in created words during he oral reading. In the writing dictation, mistakes occurred with more frequency not only with created words but also with low frequency words (FIGURE 3)

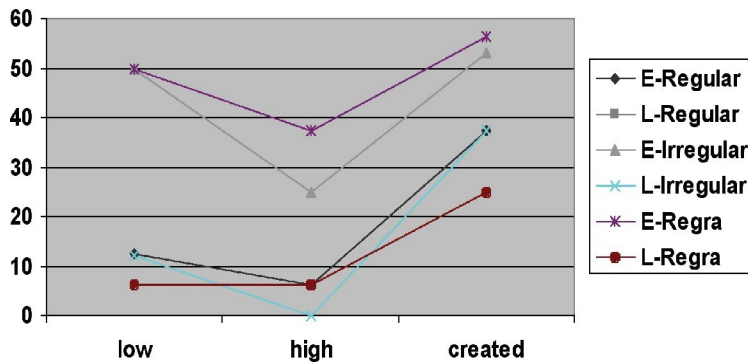


FIGURE 3 – Performance in the Writing under Dictation and Oral Reading.

It was used the Level of Reading Test (CAPELLINI, 2001) and it was verified that individual is in the orthographic step, with knowledge of the orthographic conventions of the Portuguese language. In what regards the reading speed it was used the Silent Reading Velocity Evaluation (CONDEMARIM, BLOMQUIST, 1989) and the Oral Reading Velocity Evaluation (CAPELLINI e CAVALHEIRO, 2000). In the silent reading the patient read average 64 words per minute in the oral and silent, showing, therefore, an oral and silent reading velocity adequate to its age and schooling, although with partial understanding of the texts.

In spontaneous Writing it was observed the aspects related to the elaboration of the text, lexical level, textual coherence and cohesion. In the patient it was observed lexical restriction, absence of punctuation rules and textual cohesion incompatible with its age and schooling.

In the Stomatognathic Functions it was made an evaluation of the phono-articulatory organs and its functions, such as phono-articulation, breathing, swallowing and mastication. The patient showed buccal breathing, adapted swallowing with tongue interposition, topo/topo bite, presence of differentiated pigmentation in the dorsum of the tongue, possible absence of uvula, tonsillar hypertrophy and generalized hypotonia.

Phonologic Conscience Test:

The Test for Phonologic Conscience (TPC) (CAPOVILLA e CAPOVILLA, 1998), consist of 10 sub-tests, each with four items related to the abilities of synthesis, syllabic and phonemic segmentation and transposition, rhyme and alliteration (FIGURE 4).

TABAQUIM, Maria de Lourdes Merighi et al. *Optiz Syndrome: audiological, speech and neuropsychological assessment. Salusvita, Bauru, v. 25, n. 3, p. 413-421, 2006.*

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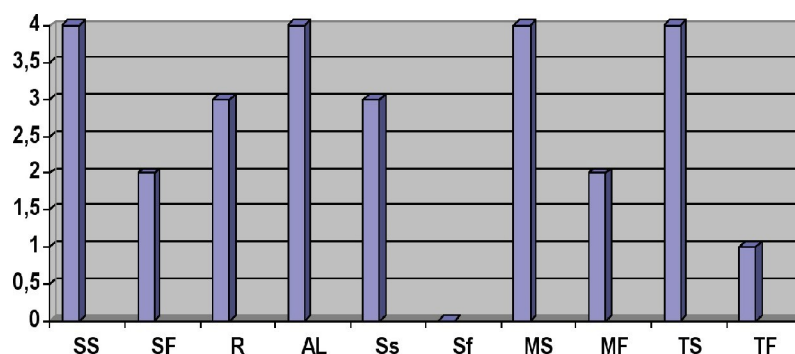


FIGURE 4 – Performance in the Test of Phonologic Conscience.

In this test the patient showed difficulties involving phonemic abilities such as synthesis, segmentation, manipulation and transposition, making evident, thus, the difficulties in the use of the phonologic system to the analysis and synthesis of words and, therefore, formation of new words. In this connection, the patient showed a score below the expected for its age and schooling.

DIAGNOSTIC CONCLUSIONS

In the evaluation of the superior cortical functions the patients showed adequate space-time orientation for similar stereognostic perceptions, being insufficient in the test that evaluate the intellectual operations in the tri-dimensional space (secondary associative cortical areas). In what regards the capacity to use numeric structures, the patient had recognized them but his performance was below the expected for his age and schooling, the same applying for spontaneous and directed writing, which were classified as primary. The maturation of the motor perception organization below the expected for his age contributed to his non satisfactory school performance.

In the phono-audiologic evaluation it was made evident the presence of language alteration in the reading and writing activities, characterizing a learning disturb due to language problems due to compromise in the use of phonologic and syntactic abilities. These characteristics were made evident by difficulty in the use of the rhythm (intonation) during reading, the partial understanding of the read text, the sole recognition of words frequent in his lexicon, the difficulty in the use of the mechanism of phonema-graphema

conversion for codification and recognition of low frequency and created words, besides a restricted vocabulary in terms of his age schooling.

CONCLUDING REMARKS

The present study is pertinent taking into consideration the incipient literature available on neuro-psychological and phonologic profile, particularly, of individuals with a diagnosis of OPTIZ G/BBB Syndrome.

TABAQUIM, Maria de Lourdes Merighi et al. Optiz Syndrome: audiological, speech and neuropsychological assessment. *Salusvita*, Bauru, v. 25, n. 3, p. 413-421, 2006.

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TABAQUIM, Maria de Lourdes Merighi et al. Optiz Syndrome: audio-logical, speech and neuropsychological assessment. *Salusvita*, Bauru, v. 25, n. 3, p. 413-421, 2006.

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