Pierre-Robin Syndrome: A Romanian Psycho-Pedagogical Case Report

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Abstract

In this paper, it is presented a psycho-pedagogical report for a Pierre-Robin syndrome diagnosed participant. The novelty aspect is ensured by the fact that the case study reflects specific learning difficulties regarding this syndrome; this syndrome is more often analyzed from a medical perspective. A psycholinguist profile of the participant in the study was developed on the basis of dual path model, aspect that provides differential diagnosis on learning difficulties, especially in dyslexia filed. Moreover the assessing instruments used are adapted and elaborated based on Romanian language specificity.

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Selection and/or peer-review under responsibility of PSIWORLD 2012

Keywords: Pierre Robin syndrome, dual path model, phonological path, lexical path, dyslexia, rhinolalia, learning difficulties;

1. Pierre Robin Syndrome-background

One of the bibliographic records belongs to Pierre Robin (1926), a physician who describes the case of a newborn with an abnormally small lower jaw, macroglossia and breathing difficulties. Scientific literature does not provide a precise circumscription of this syndrome; there are at least 15 different opinions on symptoms and about the same number of definitions for this syndrome [1-2]. Some sources call this syndrome Pierre Robin complex and others Pierre Robin sequence due to succession of sequences that occur in intrauterine period and lead to abnormal placement of the tongue tip and inappropriate palate development [3].

Epidemiological studies show that Pierre Robin syndrome is a rare, complex syndrome, affecting one in 8,500 to 14,000 births [3] (there is higher incidence for twins; there are no gender differences identified). Multiple birth defects are frequently described correlated with insufficient growth of the lower jaw. Pierre Robin Complex

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presents three distinct anomalies located on the face: micrognathia, being present in 91.7% (underdeveloped lower jaw, chin very small chin), glossoptosis present in 70-85% of cases (posterior lingual position), macroglossia and anchiloglossia occure in 10-15% of cases. The arched palate, the hard palate completely closed, U, in association with breathing problems, heart defects, malformations of the ear (otitis media is present in 80% of cases, hearing loss transmission in 60% of cases) and skeletal malformations (syndactyly, oligodactyly, coxa varus, scoliosis, kyphosis, etc.) are also presented in some cases. Pronunciation disorders are common in velopharyngeal failure, more severe in Pierre Robin patients than in those with cleft palate [1], [4].

There are several theories explaining this syndrome, the most popular is the mechanical theory. This theory considers the impact of various external factors on the fetus in intrauterine period. The mechanical theory is completed by the neurological theory and the genetic theory. The neurological theory refers to a delay in neurological maturation observed in the electromyography for the tip of the tongue. The genetic theory postulates the fact that this syndrome can be associated with chromosomal deletions such as: 2q24.1-33.3, 4q32-qter, 11q21-23.1 and 17q21-24.3 [2], [5].

The most important medical problems in this syndrome are: breathing difficulties (dyspnea) and eating disorders [6]. Babies with cleft palate need a special feeding system; an example is Haberman feeding system [1]. Also, a nasogastric tube for feeding it is necessary to gain weight, in some cases. In breathing disorders, surgical intervention can lock the front of the tongue by Beverly Douglas procedure [4].

2. Case study

The student, a 14 year old girl currently an 8th grader in a regular school from Cluj-Napoca was diagnosed with Pierre Robin syndrome during the first month of life. The student was diagnosed with learning disabilities in fifth grade and she has been included in the resource program receiving special education support for Romanian language and mathematics. The student received speech services in Elementary School, the progress in pronunciation abilities is obvious, and the student progressed from a complex polymorphic dyslalia panel, to hyper nasal speech. The student’s speech is still strongly influenced by nasal resonance, leading to extremely low intelligibility, affecting communications with peers and adults.

2.1. Objectives

We propose an investigative approach with the following goals:
• to underline developmental features of a person with Pierre Robin syndrome;
• to establish a comprehensive evaluation approach using "the dual path model" to diagnose dyslexia;
• to examine auditory-oral skills, in order to diagnose speech and language difficulties features;

To achieve these goals, the student has been examined from the following perspectives: psycho-pedagogical, speech and language therapy and educational audiology. The following examples will illustrate some sample tasks and results.

2.2. Research framework- Dual Path Model

The dual path approach is the base for the proposed assessment of this study. This model is proposed by Coltheart, 1987 [7].

The model postulates that there are two paths of treating information in reading [8-9]:
• The first system, represented by the lexical path, adopts a global comprehensive treatment of the input stimulus. The word’s spelling representation in reading is activated in the vocabulary spelling and provides access to the corresponding phonological word form with or without prior access to the semantic content.
The second system, represented by the phonological path or non-lexical path, is characterized by sequential analytical treatment of the stimulus. This system activates a converting rule system based on spelling/phonological relationship when the known word was stored independent of lexical information.

Lexical path integrity is assessed using a given lists of irregular words. A poor performance in reading irregular words compared with regular words or pseudo-words shows an interference of the reading lexical path and the analytical system. Phonological path integrity assessed is used in reading and writing in a list of pseudo-words for dyslexic and dysortographic children evaluation. In order to diagnose a specific type of learning difficulties, not only the scores but also the nature of errors and the time needed to perform the task are relevant.

2.2.1. Aspects of the procedure and working methods regarding phonological abilities

To assess complex phonological skills, the following types of tasks were used: reading a 40 words list characterized by phonetic writing; reading a 40 words list with phonological writing.

The two lists of words are approximately equivalent, both following specific phonological Romanian language rules: presence of diphthongs and triphthongs; presence of words with hiatus; short-vowel "i" in post-consonant position; words written with two "i" as the middle word and the final word; doubling consonant "n" in compound words; consonant "m" before consonant "p" or "b"; explosive consonant-final aspiration; sound-groups "che", "chi", "ghe", "ghi", "ce", "ci", "ge", "gi"; words containing graphemes "x", "k"; omission of the glottal stop by labial and palatal appendix; doubling the vowel "o"; words ending in sound sequence "iu"; words that are written both "â" and "i"; compound words written with "i".

The two lists contained monosyllabic words and four syllables words.

2.2.2. Results and discussions

The student records most errors while reading pseudo-words. Thus, the student has difficulty reading 8 pseudo-words out of the 40 pseudo-words from the given list.

The following errors are frequent:

- Omission of the first "o" in the pseudo-word "nealocon". Thus, the child pronounced "nealcon" because pronunciation is much easier.
- Replacing group sounds "gi", in "ghi"; the pseudo-word "pogi" was transformed to "poghi". This can be explained also through minimizing co-articulator effort.
- Shifting stress from vowel "u", to vowel "i" in the word "túti", resulting "tutíi". In this case a possible hearing deficit may be an explanatory hypothesis.
- Omission of consonant "n" from pseudo-word "cantamuc", resulting "catamuc", in this case auditory perception could be the problem.
- Transforming the hiatus "ia" from the pseudo-word "chetian" resulting "chetan". In this case we identify difficulty handling "i" vowel. The same difficulty in "i" phoneme perception seems to be present in pseudo-word "jíridin" which is pronounced "jírdin".
- There are only two situations where the child pronunciation differs greatly from given items, like "cozdromuláč" and "tialutip" resulting "cozdromuale" and "tialupi". In these situations the pseudo-words phonological complexity, their syllabic length exceeded student's lexical skills. These two pseudo-words are the most evident proof of the student's lexical deficit.

When the student read the 40 words list she recorded three errors. This demonstrates and argues that phonological processing deficit is to be considered in combination with the lexical nature, in order to explain learning difficulties in this case. The word "mioara" is pronounced "mioarä", the tendency is to neutralize the final vowel "a". Delayed trend can be explained by accessing lexical path, decoding is limited to the first sound in the word. The same situation occurs when the word "înotă" is read "înotăță". Thus, we are entitled to affirm that the lexical component part is affected, in order to explain phonological learning difficulties that the student records.
2.2.3. L’Alouette test

L’Alouette test "Skylark" (Lefravais, 1967, reviewed by Debray, 1972) is a commonly used test in language therapy and neuro-pediatrics [10-11]. L’Alouette test contains a text designed to show the structure of an apparent text, containing meaningful words, taking advantage typical sentence structures of French, but where juxtaposition is meaningless. The task is to read orally and fluently. Theoretical hypothesis - the author's intention is to examine the process of decoding and to highlight the discrepancy between the phonological decoding speed and lexical processing. Romanian version of the text is experimental.

From the total of 265 words presented, the student read correctly 245 words. In the table 1 teh English equivalent word is presented in the first column. The correct form of the words to be read is presented on the second column. The third column three presents how these words were read by the student. The elision of several sounds target lexical item is observed, some items are replaced with other Romanian lexical items, for example the word "arteră" is read "arcă"; the addition of some sounds, for example the word "parbrizul" is read "parbrizului" and replace sounds to obtain known words, for example the word "impozanții" is read "importanții".

Tabl 1. Errors l’Alouette-Romanian variant

<table>
<thead>
<tr>
<th>English equivalent word</th>
<th>Correct word</th>
<th>Word read</th>
</tr>
</thead>
<tbody>
<tr>
<td>devoted</td>
<td>devotat</td>
<td>dezvoltat</td>
</tr>
<tr>
<td>cellular</td>
<td>celularului</td>
<td>celularor</td>
</tr>
<tr>
<td>cut out</td>
<td>decupa</td>
<td>deasupra</td>
</tr>
<tr>
<td>precipitated</td>
<td>precipitată</td>
<td>precipată</td>
</tr>
<tr>
<td>brush</td>
<td>penel</td>
<td>penele</td>
</tr>
<tr>
<td>monitor</td>
<td>monitorul</td>
<td>monitorului</td>
</tr>
</tbody>
</table>

2.2.4. Pure tone audiometric examination

During the audiometric examination, in addition to recording responses to pure tones, the student’s behaviour was also recorded and the data were analyzed on the following coordinates: the nature of behavioural responses; responses latency to hearing stimuli; the presence of a response even when the sound was absent. Since the testing period was extended by the need, to resume the demonstration, as well as due to the numerous hesitations and at random responses, during the first session only three frequencies were tested. The procedure was resumed and it was completed in other assessment sessions.

The air conduction thresholds are higher in the right ear than left ear: right ear (RE) 500Hz – 35 dB; 1000 Hz – 30 dB; 6000 Hz – 10 dB; left ear (LE) 500Hz – 35 dB; 1000 Hz – 20 dB; 6000 Hz – 25 dB.

The bone conduction thresholds were recorded as following: 500Hz - 5 dB, 1000 Hz - 10 dB, 6000 Hz - 5 dB, indicating a higher auditory sensitivity in high frequencies than in low frequencies, where hesitations, the attempts and excessive latencies were more numerous than in the case of 6000 Hz frequency.

Clinical interview revealed the presence of intermittent sensations by the participant, as followling: popping, sometimes the feeling of sunk head (ear), feeling that disappears when swallowing or yawning, indicating a link to Eustachian tube. Under these circumstances, it can be underlined the presence of a low hearing loss in low frequencies. In order to verify the assumption of a conductive hearing loss, examination will be resumed and completed with other investigations of the auditory system. These data will be corroborated with data obtained from speech and language evaluations and from psycho-pedagogical assessments.
3. Conclusions

The dual path model provides the possibility to elaborate a complex assessment procedure, in this case, revealing specific learning difficulties due both to lexical and phonological paths in accessing language. In order to clarify the results obtained using the meaningful words lists, pseudo-words list and the text from Romanian variant L’Alouette a pure tone audiometric examination was developed in order to eliminate the suspicion of hearing loss. In this way, student’s linguistic abilities are more adequately revealed in order to properly diagnose the specific type of dyslexia, the student presents. This research results will be further used in developing a psycho-pedagogical assistance program for this student. The assessment procedure is considered a good practices model and it can be further implemented to other situations to diagnose learning difficulties.

References


