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NEUROPSYCHOLOGICAL AND PHONOLOGICAL EVALUATION IN THE APERT'S SYNDROME

Study of two cases

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ABSTRACT - This study evaluated two cases of Apert's syndrome, through phonological, cognitive, and neuropsychological instruments and correlated the results to complementary exams. In short, this study reveals the necessity of application of neuropsychological, cognitive and phonological evaluation and correlation of the results with complementary testings because significant differences can be present in the Apert's syndrome.

KEY WORDS: Apert syndrome, neuropsychological evaluation, phonological evaluation.

Avaliação neuropsicológica e fonológica na síndrome de Apert: estudo de dois casos

RESUMO - O objetivo deste estudo é apresentar a avaliação fonológica, cognitiva e neuropsicológica de dois casos com síndrome de Apert e correlacionar os achados destas avaliações com o resultado de exames complementares. Este estudo nos possibilitou verificar a necessidade da realização dessas avaliações em decorrência de diferenças significativas presentes nos casos com síndrome de Apert .

PALAVRAS-CHAVE: síndrome de Apert, avaliação neuropsicológica, avaliação fonológica.

Apert's syndrome was described by Wheaton[1], in 1894. In 1906, Apert[2] published a summary on nine cases, and in 1920 Park and Powers[3] wrote an excellent monography on this disease. In 1960, Blank[4] registered a total of 150 published cases. Apert's syndrome is a genetic pathology of dominant autosomal inheritance and it has as main characteristics: the acrocephalia due to synostosis of the coronary suture and the syndactilism which most of the time is symmetrical involving the four extremities.

The cranium is shortened in the antero-posterior diameter and prolonged vertically, taking the characteristic acrocephalic aspect. The face is generally flattened and it can be asymmetric. There are front bossas and the occipital is flattened. Most of times there are big fontanels, closing with lateness. The jaws are frequently hipoplasic and a prominent jaw resulting in a moderate prognatism. There is ocular hypertelorism and the orbits are sidelong shallow and oblique, with antimongoloid inclination and protusion of the ocular globes. Frequently there is strabismus. The nose is short and enlarged. The nasal bridge is flattened. The pavilion headphones are generally moved away of the head and they seem big. The palate is arched, narrow and it can be cleft. The maxillary arcade can present a configuration in V with superimposed teeth and protuberant gums. The dental eruption is late. It presents bony and/or cutaneous syndactilism, going from the partial coalition of the fingers, it is generally observed total coalition of the second, third and fourth fingers. Sometimes the last phalanges of the thumbs come increased in volume and with valga position . The fingers can be short. Cutaneous syndactilism of all the ankles, with or without bony syndactilism. The extremity of the great ankle is sometimes thick and malformed. Moderated or serious acne in the teenagers, including in the forearms.

Gonçalves and Silva[5] discussed how a clinic-genetical procedure may help the clinicians. This syndrome demands precocious treatment, even though precocious neurosurgical treatment does not pre-

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vent the mental deficiency. There can be mental deficiency, but cases of normal intelligence were also registered; the incidence of the mental deficiency is unknown.

The objectives of the present stydy are: to accomplish phonological intellectual and neuropsychological evaluations in two children with Apert's syndrome; to compare the discoveries of the phonoaudiological, intellectual and neuropsychological evaluations among the two children with Apert's syndrome.

METHOD

Subjects - Two female children took part in this study, the first child (Case 1) is 8 years and 5 months of age, she attended the second year of first grade in 1998, her economic social level is low; the second child (Case 2) is 12 years and 9 months, she attended APAE, her economic social level is low.

Procedures - The following instruments of evaluation were used: - neuropsychological evaluation –Luria questionary –Nebraska[6]; - the Wechsler Intelligence Scale for Children – WISC[7]; - phonological child evaluation[8]; - awareness phonological test (APT)[9]; - brain scintillography - SPECT.

RESULTS

The first case obtained the following results in the WISC[7]: she presented intellectual revenue within the standart average in relation to the children of the same age group, as it shows the following chart (Table 1).

It was obtained the following results in the second case: the WISC[7] presented the intellectual output below the average in relation to the children of the same group as in chart II (Table 2).

The WISC data in both cases were significant showing that just Case 1 demonstrated higher re-

Table 1. Evaluation of the Case 1 – WISC.

Sub test	Age in the testing	Score	Age difference
Information	7a. 6m.	8	11 m.
Understanding	7a. 10m.	8	7 m.
Arithmetc	7a. 6 m.	5	11 m.
Likeness	9a. 10m.	8	1a. 5m.
Numbers	7a. 2m.	7	1a.2m.
To complete illustrations	8a. 2m.	8	3 m.
Arrangement of illustrations	8a. 2m.	20	3 m.
Cubes	10a. 2m.	18	1a. 8m.
Setting objects	8a. 10m.	17	5 m
Code	7a. 2m.	18	1a.2m.

Table 2. Evaluation of the Case 2 – WISC.

Sub test	Age in the testing	Score	Age difference
Information	8a. 2m.	9	4a.7m.
Understanding	8a.6m.	9	4a.3m.
Arithmetc	6a.6m.	4	6a.3m
Likeness	8a.10m.	7	3a. 11m.
Numbers	6a.2m.	6	6a. 7m.
To complete illustrations	8a.2m.	8	4a. 7m.
Arrangement of illustrations	7a.2m.	12	5a. 7m.
Cubes	8a.6m.	9	4a. 3m.
Setting objects	8a.10m.	17	3a.11m.
Code	8a.10m.	29	3a. 11m.

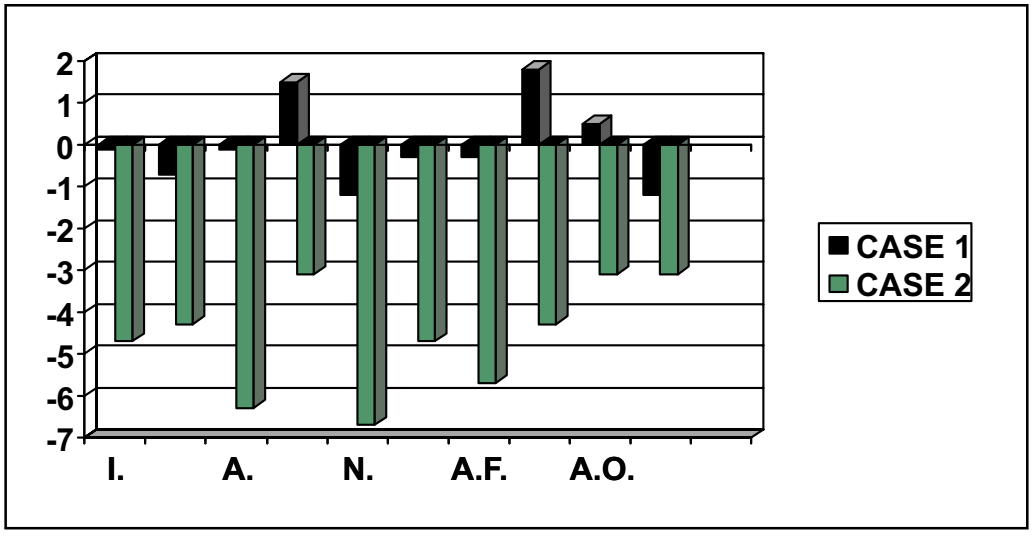

Fig 1. Difference between the achieved age and the cronological age in both cases.

sults in the age group in the tests of likeness, cubes and setting objects, which measure respectively associative thought, abstraction, as it represents Fig 1.

In the neuropsychological evaluation, Case 1 obtained better results in tactile ability, rhythm, speech, expressive language, mathematical reasoning and memory. And in Case 2, it was obtained better results in moving and right ability, writing and reading, as it represents Figure 2.

Concerning the phonological evaluation, the child of the Case 1 presented substitution of deaf phonemes for sound amethodically such as / k / - /g /; / *ʃ* / - /v /, besides distortion in the phonemes /s / and / *s* /. The child of Case 2 presented distortion in the phonemes /s / and / *s* /, systematic omission of the phoneme /l/ in syllabic clamping and asystematic omission of this phoneme in consonantal group, substitution asystematic of /l / for /r / in consonantal group and systematic substitution of /n / for / l /, and language earliness in the phoneme /t /. Although in both cases the children presented lexicon suited to the age.

In concern to the results of the Awareness Phonological Testing (APT)[9], the child Case 1 obtained score of 22 points and the child Case 2, 26 points. In both cases, the children presented difficulties in the execution of the subtests which involved the phonemic and sylabic abilities, indicating therefore those phonological difficulties (Fig 3).

In SPECT the results were as shown in Figures 4 and 5.

DISCUSSION

Gonçalves and Silva[5] described normal neurological evaluation in the clinical suggestive case reported. In previous studies, Patton et al.[10], Léfèvre et al.[11], it was observed among 70 patients, with no sex prevalence, being 50% of men and 50% of women, respectively. After these studies it can be concluded that most of the patients with Apert's syndrome is mental faulty in different levels. The most common causes observed to determine the mental deficiency are: perinatal factors, hydrocephalus, malformation

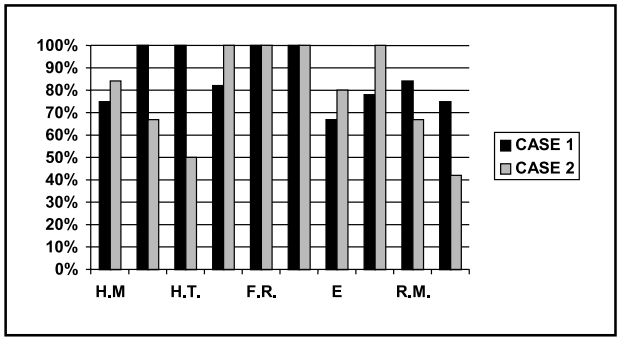

Fig 2. Comparision of the neuropsychological evaluation in both studied cases.

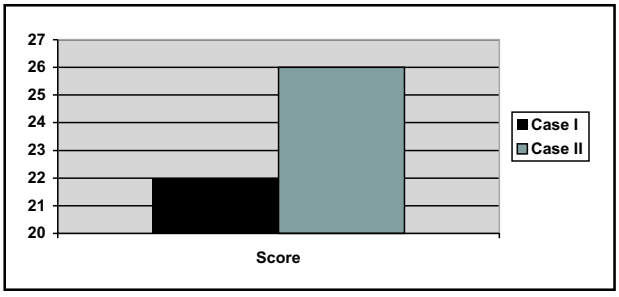

Fig 3. Representation of the scores of the awareness phonological testing (APT).

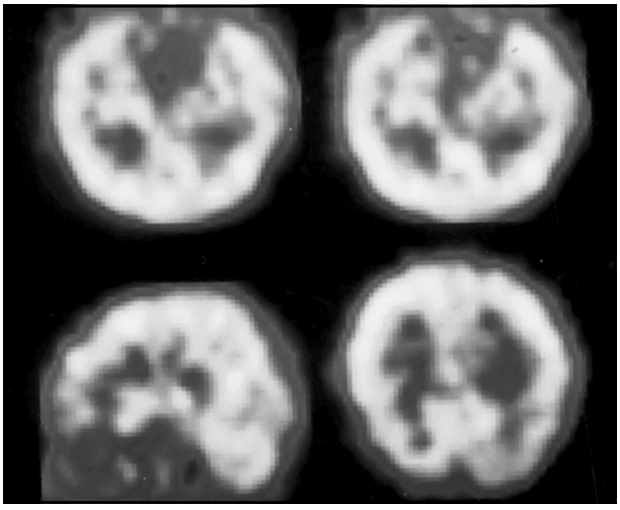

Fig 4. SPECT of Case 1. Superior half of the temporal axis: perfusion of the temporals. Sagittal inferior axis to the left: homogeneous and normal perfusion of the brain cortex and other structures. Inferior transversal axis to the left: homogeneous and normal perfusion of the brain cortex.

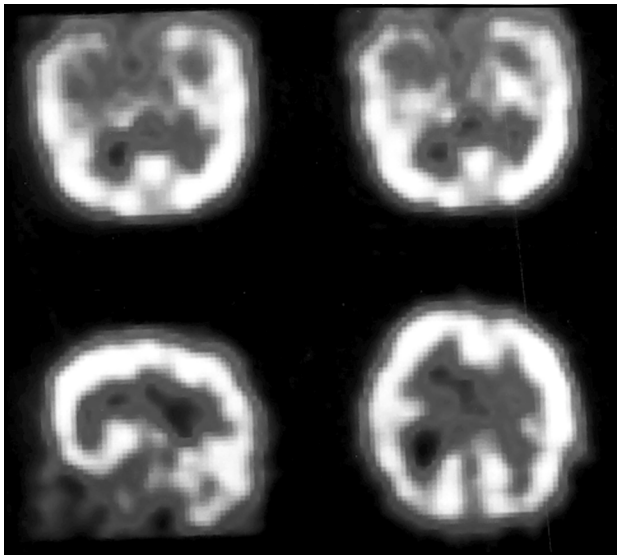

Fig 5. SPECT of Case 2. In the superior half two rebuilt cuts in the temporal axis. Hypoperfusion in the temporal lobe shows an area of moderate hypoperfusion in the medial area of the right temporal lobe. The image of the sagittal inferior axis to the left shows discrete hipoperfusion of the brain cortex and the image in the right inferior transversal axis shows morphological alteration of the brain cortex, the right hemisphere is bigger than the left one and more indirect of ventricular expansion.

of the central nervous system, and the increase of the intrabrain pressure.

The data were correlated to exams of neuroimage (SPECT). According to the accomplished evaluations, it was observed that the results of the second evaluation case confirms the characteristic of mental deficiency presented by the Apert's syndrome shown in the literature, the patient revealed difficulty in the tests : rhythm, tactile ability, memory, mathematical reasoning, arithmetic, numbers and arrangement of illustrations compatible with the hipoperfusion in the right temporal lobe shown by SPECT. However, the first case differs from studies by presenting normal intellectual revenue, without other deficits and presenting a normal SPECT, being an atypical case to the situation, proving that inside the same symptomatology there can be significant differences which justify the neuropsychological diagnosis and the phonological difficulties.

In concern to the phonological evaluation, in both cases it was possible to verify the presence of phonological deviation and difficulty in the manipulation of the sounds of the speech. On the other hand in the second case it was verified that the phonological deviation is compatible with the hypoperfusion in the temporal area shown in the SPECT; in the first case, the SPECT is normal and so there is no link between the findings of the neuroimage and the phonological findings in this case. But it is necessary to perform the neuroimage to stablish the presence or not of the hypoperfusion in the temporal area, mainly because the phonological deviation can be present in both cases : with the presence of the hypoperfusion of this brain area (Case 2) and in cases with no evidence of the same however with the evidence of lateness language development (Case 1).

In concern to the language, few are the studies which were found in the literature. However we can mention the study accomplished by Misquiatti[12], which evaluated 8 individuals' language with the Apert's syndrome, 4 with Crouzon's syndrome and 3 with Pfeiffer's syndrome. As procedures it was accomplished genetic evaluations, psychological evaluation, audiological evaluation and phonoaudiological evaluation. The findings revealed that 9 individuals presented conductive hearing loss with degrees varying from light to moderate and only 2 with normal hearing; just 4 individuals with light mental deficiency; most of the individuals with current syndrome of mutation in the gene FGFR2 presented complaints with relation to the language and hearing, and most were related to the oral delivery; the language alterations related to the emission and oral reception, perceptual processes and basic functions were presented in the great majority of individuals, in different commitment the delay in the acquisition of the language frequently happened in the appraised individuals. The author concludes from this study the

need of specific phonoaudiological studies both in area of the language as of the audiology, in order to determine phonoaudiological manifestations in these syndromes, besides the need to direct these individuals precociously to rehabilitation of the disturbances of communication.

In short, this study reveals the necessity of application of neuropsychological, cognitive and phonological evaluation and correlation of the results with complementary testings because significant differences can be present in the Apert's syndrome.

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