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ATYPICAL RETT SYNDROME FORM – CASE STUDY
NIETYPOWA POSTAĆ ZESPÓŁU RETTA – OPIS PRZYPADKU

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Summary

Rett syndrome is a rare, progressive neurological disorder that affects mostly female patients. It is a syndrome of pervasive developmental disorders of the neural, skeletal and gastrointestinal systems. According to the statistics Rett syndrome occurs in approximately 1 : 10 000 – 15 000 live births. Besides the classical form of the syndrome, there exist an atypical variation and the clinically distinct form with preserved speech. The authors present a case of a patient suffering from atypical Rett syndrome variant, with no MECP2 gene mutation.

Key words: Rett syndrome, MECP2, atypical Rett Syndrome form.

Streszczenie

Zespół Retta jest rzadkim, postępującym schorzeniem neurologicznym występującym głównie u dziewczynek. Jest to zespół całościowych zaburzeń rozwoju współistniejących w obrębie układu nerwowego, szkieletowego oraz żołądkowo-jelitowego. Według danych statystycznych częstość występowania zespołu Retta szacowana jest na 1: 10 000 – 15 000 urodzeń. W zespole Retta można wyróżnić klasyczną postać, nietypową postać oraz postać odmienną klinicznie z zachowaną mową. Autorzy przedstawiają opis przypadku pacjentki z nietypową postacią zespołu Retta, u której nie występuje mutacja genu MECP2.

Słowa kluczowe: zespół Retta, MECP2, nietypowa postać zespołu Retta.

Introduction.

Rett syndrome is described as a syndrome of pervasive developmental disorders of neural, skeletal and gastrointestinal systems. [1,2,3,4]. The syndrome is a rare, progressive neural disorder which affects mainly girl patients [5]. According to the statistics Rett syndrome occurs in approximately 1 : 10 000 – 15 000 live births; the actual number of patients is probably greater though, due to the syndrome not having been diagnosed [6,7].

Rett syndrome occurs in its classical form, atypical form or in the clinically distinct form with preserved speech [8,9]. The classical variant of Rett syndrome has a specific course. Until the 6-18 month of age there are no abnormalities. Psychomotor development and head growth are normal. Developmental regression begins between the 1-4 year of child's age. There occur deceleration in the rate of head growth, developmental regression and loss of purposeful hand use. Repetitive stereotypical arm movements are also present. The next stage oscillates between 2 and 10 years of age. It is the stage of apparent stagnation, in which despite the fact motor disturbances constantly progress, some improvements in behaviour are to be observed. The last stage lasts until the adult age. The characteristic features of this stage include movement regression, atrophy, spasticity and scoliosis [10,11,12,13].

The atypical Rett syndrome form includes abnormal child's development, congenital muscle hypotonia and flexion spasms, and is connected with the more severe course of disease. There also exists the clinically distinct form with preserved speech. Onset occurs late and child does not lose speech [14]. The diagnosis of Rett syndrome is based on diagnostic criteria set by the group of experts. They distinguished main, supportive and exclusion criteria. There also exist diagnostic criteria according to ICD-10 [15,16].

Case study.

A child without positive family history of hereditary disease, born prematurely, weighing 1800g, with 29cm head circumference. Developmental retardation observed from her early infancy. She was the patient of the Clinic in Bydgoszcz, where she was thoroughly examined and rehabilitated. At the age of 3 years the girl was examined for severe psychomotor developmental retardation. She was diagnosed with severe developmental retardation (she does not sit) and microcephaly (43,5 cm).

Biochemical analysis did not affirm any congenial metabolic disease. Neurological examination showed hypotonic form of cerebral palsy. The patient was advised to remain under

neurological and rehabilitation care near the place of residence. The patient has been treated for epilepsy since she was 5 years old. CT showed neuronal migration and cortex differentiation disorders. Generalised seizures, initially tonic-clonic, were well-controlled; at the beginning of 2003 there occurred right-sided generalised nocturnal seizures which were more difficult to control. In 2004, after being admitted to the Provincial Children Hospital in Bydgoszcz, the patient was diagnosed with bilateral pyramidal tract syndrome with left-side dominant, leg joints' flexion contractures, stereotypic hand movements, severe developmental retardation; there was no logical communication with the child. The MRI examination showed hypogenetic corpus callosum, agenetic septum pellucidum and slight cortical atrophy.

In 2005 molecular examination was conducted, as the child's phenotype indicated Rett syndrome. No MECP2 gene mutation was found. In 2006 the patient was once more admitted to the Provincial Children Hospital in Bydgoszcz. Neurological consultation showed pyramidal-extrapyramidal syndrome and well as epilepsy. Psychological consultation affirmed severe retardation (assessment according to the Brunet-Lezine scale). Psychological development level of a 11-month old child. No speech, unintelligible vocalisation. Emotional reactions were simple but qualitatively diverse. The girl was completely non self-reliant.

Th-L spine and hip joints were X-ray examined. Examination showed significant Th-L scoliosis to the left, with vertebral bodies being rotated to the left. Hip joint X-Ray examination showed underdevelopment of right-sided pelvic bones, dysplasia and subluxation of the right hip, right femur shaft rotated to the outside.

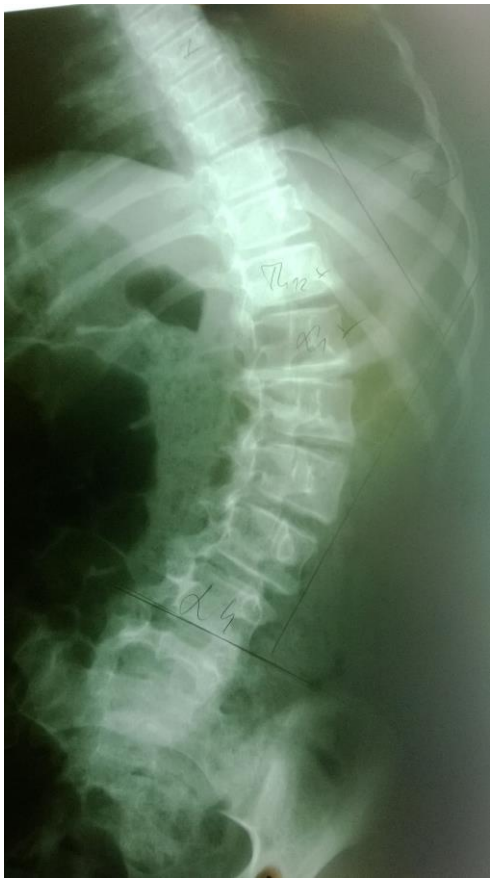

Fig. 1. X-ray of the spine.

Furthermore, stenosis of both hip joint articular spaces in their upper-lateral parts was diagnosed. In addition, the patient suffers from symptoms which are classified as supportive criteria of diagnosing atypical Rett syndrome form, i.e. irregular breathing, abdominal bloating, muscular atrophy, sleep disorders, bruxism, decreased sensitivity to pain stimuli, cold feet.

The patient has been constantly rehabilitated. Her treatment concentrates on some of the symptoms, as it is impossible to modify the course of the disease. The treatment mainly includes kinesiotherapy using the NDT-Bobath method, unloading and passive exercises. The patient is given regular dorsal muscles dry massage and undergoes physiotherapy using sollux - red filter, as well as laser- and magnet therapy. Gel compresses are applied to Achilles tendon, the patient's shank muscles are treated with tonolysis. The patient needs multi-specialty care and remains under strict dietary, cardiological, psychological and pedagogic control.

Discussion.

In the recent years Rett syndrome has been gaining interest and many unusual and atypical case studies have been recorded. Diagnostic procedure in Rett syndrome is a difficult task which requires vast knowledge and experience. Patients suffering from Rett syndrome often show complicated clinical picture, which can become even more complex due to overlapping coexisting diseases. It is interesting to notice that with the patient described in this article there was no MECP2 gene mutation found, although she shows phenotypic Rett syndrome picture. According to the research and observations conducted by Amir R.E, Van den Veyver and Wan M. [17] not finding the MEXP2 gene mutation does not exclude diagnosing the syndrome, and the MECP2 gene mutation is presumably non-specific to Rett syndrome. The presence of MECP2 gene mutation is observed at the part of girl patients, but also probably in other similar neurogenetical disorders. Punctual MECP2 gene mutations are observed at 80% of classical syndrome form cases and at 40% of patients suffering from atypical form [1]. It is worth mentioning that there exists constant need of expanding knowledge of Rett syndrome, as it may result in improving the effectiveness of diagnosing and treating this disease.

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