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DENTAL ISSUES IN RETT SYNDROME*

PROBLEMY STOMATOLOGICZNE W ZESPOLE RETTA

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Abstract

The advancements in science and technology allowed saving the lives of children, who had no chance of survival before. Hence the problem of so called rare diseases, usually genetically determined. It is a new challenge for both the physicians and the health services. These children require a coordinated multi specialist oriented health care, which includes also dentists. This situation is reflected by the case of an 18 years old girl with Rett Syndrome, described by us. In this patient despite numerous visits to various dental practices, no decision of a radical surgical extraction of the tooth has been conducted. In our Department the extraction of teeth 22, 16 and 14 has been performed, as a part of 1 day surgery procedures, thus eliminating the dental infections and pain.

Conclusion: *Elaboration and introduction into praxis principles of dental care in children and young adults with rare diseases are needed.*

Key words: Rett syndrome, dental focal infection, treatment

Streszczenie

Postęp w nauce i technice umożliwił ratowanie życia dzieciom, które dawniej nie miały szans przeżycia. Stąd pojawił się między innymi problem tzw. chorób rzadkich przeważnie uwarunkowanych genetycznie. Stanowi on nowe wyzwanie zarówno dla lekarzy, jak i dla organizatorów służby zdrowia. Dzieci te wymagają bowiem skoordynowanej wielospecjalistycznej opieki medycznej, w tym również stomatologicznej. Sytuację tę odzwierciedla opisany przez nas przypadek 18-letniej pacjentki z zespołem Retta, uwarunkowanym genetycznie. U pacjentki, mimo licznych wizyt w gabinetach stomatologicznych, nie podjęto decyzji o radykalnym postepowaniu dotyczącym chirurgicznego usunięcia zęba. W naszym Zakładzie w ramach procedur chirurgii jednego dnia wykonano ekstrakcję zęba 22, 16 oraz 14 eliminując zakażenia pochodzenia zębowego oraz ból.

Wniosek: *Konieczne jest więc opracowanie i wdrożenie do praktyki obowiązujących zasad opieki stomatologicznej u dzieci i młodzieży z tzw chorobami rzadkimi.*

Słowa kluczowe: Zespół Retta, ogniska zapalne zębopochodne, leczenie

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The Rett Syndrome (RTT) was described for the first time in 1966 by the Austrian physician Andreas Rett. It is a progressive neurodevelopmental, and skeletal disorder that manifests itself with profound impairment of learning abilities, disturbances in communicative abilities, stereotypic movements, wringing, seizures, breathing anomalies, dystonia, growth retardation, scoliosis, kyphosis, trophic changes in distal parts of the extremities, autistic features, nocturnal laughing or screaming attacks, also teeth grinding [1]. In 80% of classic Rett cases, mutation in the gene *MECP2* are identified, and in 8 % deletions of a part or even whole *MECP2* gene [2]. It is a genetic disorder that manifests mainly in girls with an incidence of 1 in 10 000-15 000

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[3, 4]. RTT is caused by a X-linked dominant mutation that is lethal in most cases in males. Approximately 99% cases are de novo mutations in *MECP2* gene [5].

The aim of the study was to show the dental aspect of the Rett Syndrome with the specific considerations for difficulties in treatment of handicapped patient with so called rare diseases.

CASE REPORT

An 18 years old girl reported with her mother to our Department, due to an inflammatory infiltration in the area of tooth 22. The patient was disabled (fig. 1). As the interview of the mother showed, first symptoms of the disorder occurred between the 6th and 10th month of age. They consisted of general psychomotor retardation; the child did neither crawl nor walk. It just shifted on the buttocks. Between the 1st and 4th year of age, the mother observed loss of the previously acquired abilities, such as purposeful use of the hands, hand apraxia, loss of language and disability of walking, frequent crying at night, breathing disorders, seizures and drooling. Such symptoms urged the mother to visit a pediatrician. The EEG showed epileptic seizures. From the provided medical documentation, it emerged that in 1997 the Rett Syndrome was diagnosed in the girl with refractory epilepsy and partial secondary generalized seizures at the age of 4.

Family history towards the Rett Syndrome was negative.

Patient's mother reported also an issue with surgical treatment of the tooth 22 in her daughter. The pain of the maxillary second incisor began 2 weeks before their arrival to our Department. Despite numerous visits to various dental practices, no decision of a radical surgical extraction of the tooth has been conducted. Only the pharmacological treatment (antibiotics) has been ordered. There was a big problem for the mother, also the helplessness caused by the lack of help from the dentists.

Extra oral examination showed hand wringing and washing movements (fig. 2), sudden lurching of the head towards the shoulder (fig. 3). Intra oral examination allowed to establish: teeth grinding – bruxism, difficulties in chewing and swallowing, neglect of oral cavity hygiene, oral inflammation with hypertrophic gingiva (fig. 4), also many cavities in teeth: 16, 14, 22, 23 (fig. 5). Tooth 22 presented severe pain to vertical and horizontal percussion, and palpation caused pain of maxillary alveolar bone with visible sinus tract.

Child's mother was presented with the diagnosis and at the same time a surgical treatment plan in procedures of 1 day surgery, which consisted of extraction of 3 teeth with pulp gangrene, in which one with active inflammation. After obtaining a written consent, the appointment has been set in an urgent mode.

In general anesthesia teeth 16, 14 and 22 has been extracted. The intra-alveolar curettage performed and the wounds surgically closed. The intra and postoperative course was uneventful.

After the surgery, the patient was transferred to recovery room, where a physician monitored her condition. Eventual breathing problems, balance disturbances, diplopia, pain, dizziness, nausea and vomiting were of special consideration. Double vision was tested in a sitting position with the patient tracking the examiner's finger, which

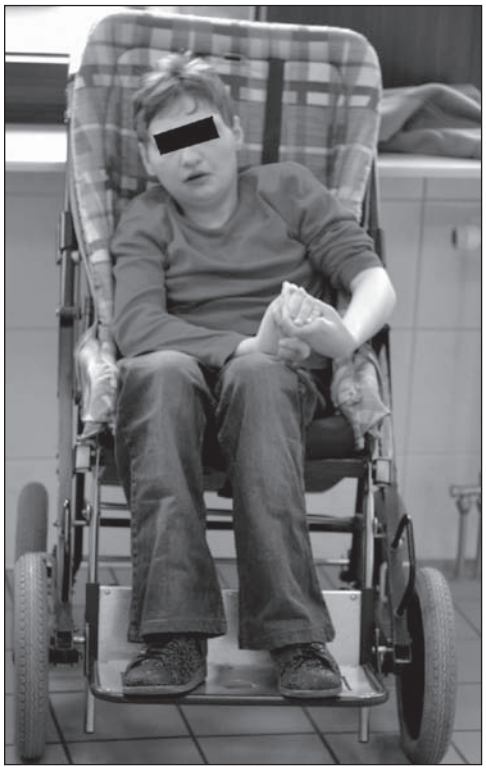

Fig. 1. Disabled patient in a wheellchair.

Ryc. 1. Pacjentka poruszała się na wózku inwalidzkim.

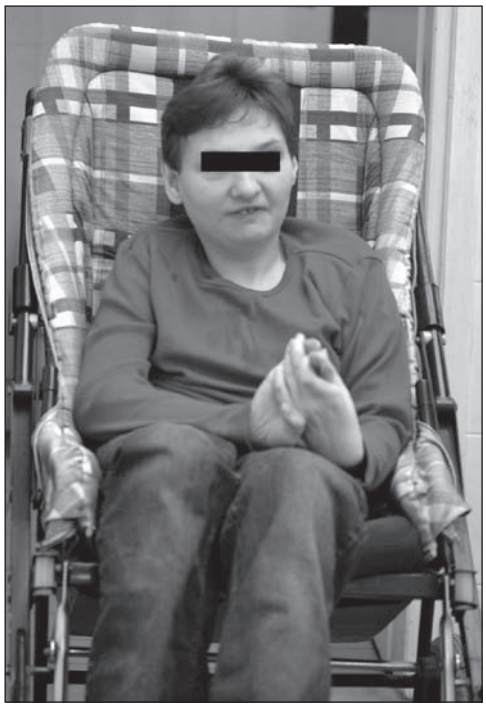

Fig. 2. Rubbing the hands together.

Ryc. 2. Zacieranie dłoni.

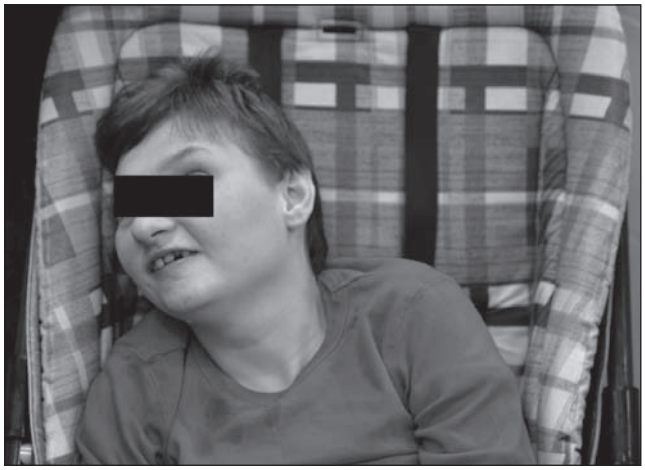

Fig. 3. Sudden head turns and leaning it on the shoulder.

Ryc. 3. Raptowne zwroty głowy do boku i pochylanie jej na ramię.

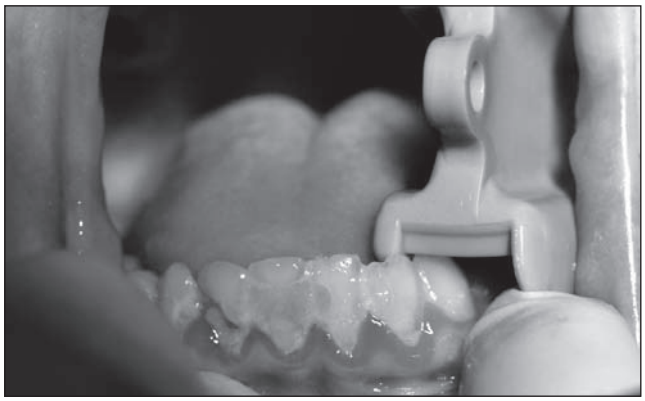

Fig. 4. Poor oral hygiene with overall inflammation and gum hyperplasia.

Ryc. 4. Zła higiena jamy ustnej ze stanem zapalnym i przerostem dziąseł.

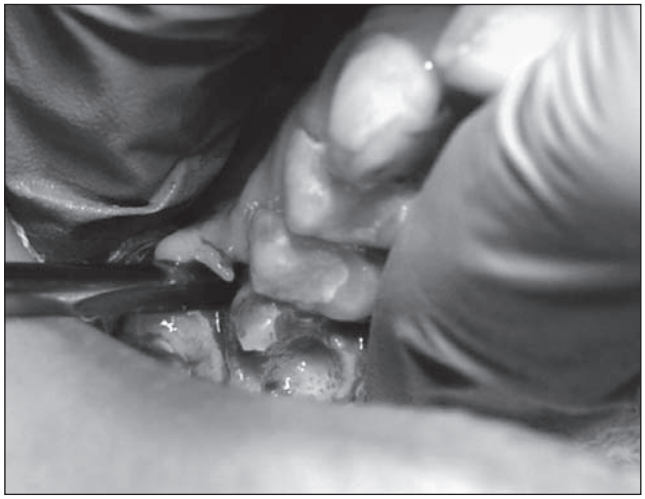

Fig. 5. Many cavities in teeth 16 and 14.

Ryc. 5. Liczne ubytki próchnicowe w zębach 16, 14.

is moved to extreme gaze to the right, left, upward and downward, diagonally to either side, and finally inward toward the patient's nose. In case of seeing two fingers, double vision is diagnosed. After achieving a state of balance, the decision has been made to discharge the patient.

Postoperative examination in 1st 24 hours after the surgery showed no edema of the operated area. Wound rinsing has been performed and further control visits scheduled. In the 7th day after the surgery the sutures were removed. The wound healed correctly.

DISCUSSION

We present a case of a girl with the deletion of *MECP2* gene.

Rett Syndrome is a progressive genetic neurodevelopmental disorder with mental retardation in females. It consists of 4 stages [6]. The beginning of the RTT is usually indistinguishable and lasts from the 6 months to 1,5 year of age. It is characterized by deceleration of head growth, muscle hypotonia, and indifference to surrounding environment and play. Second stage involves losing the acquired communicative and social abilities, and also motor coordination. With simultaneous intensification of stereotypic movements and indifference towards other people, loss of purposeful use of the hands and instead developing uncontrolled wringing and mouthing. This stage lasts from 1 to 4 years [7]. Those latter symptoms were observed in our patient. Third stage, called the apparent stagnation, is observed between the 2 and 10 years of age. Motor deterioration and abnormal hand moves are even more visible. Followed by improvement in child's behavior, manifesting by reduction of crying, calming down and reduced autistic features. Unfortunately, increased muscle tension and scoliosis is observed. The fourth stage is characterized by further loss of motor coordination with muscle atrophy, scoliosis and aggravation of the ability to move [4].

Chahrour and assoc. [8] are of the opinion, that the loss of verbal communication causes the screaming fits, irritation, anger and self-abusing behavior, which in turn result from the fact of feeling helpless due to poor social communication and stress coping. According to Nomura [9], the existing hypersensivity to sound, lack of eye to eye contact and reduced facial expressions resemble autistic features. Weese-Mayer and assoc. [10] underline the loss of motor coordination, development of ataxia and loss of ability to walk, which is the case described by us. The above-cited authors also report disturbances in autonomic nervous system, such as hyperventilation during wakefulness, breath-holding, apnea with the Valsalva effect, air swallowing and forced expulsion of air and saliva. Jian and assoc. [11] reveal, that the significant symptom is the occurrence of seizures, from easily controlled to tonic-clonic seizures. The mother of treated patient reported those symptoms. She also mentioned that the increase of the symptoms and their incidence tend to decrease lately, which should be explained by reaching puberty or effective anti-epileptic drug.

Matijevic and assoc. [12] note loss of weight or obesity and hypotrophic, cold blue feet. Osteopenia is a cause of frequent bone fractures and scoliosis. Authors underline also major cardiac problems, such as tachycardia or sinus bradycardia, and with time, further loss of motor coordination, scoliosis and dystonia. Whereas Roze and assoc. [13] observe that, as the patients get older (6th and 7th decade of life) they often develop Parkinsonian features. It needs to be said, that the type of disturbances and their intensity are individual, which influences the diversity of clinical picture and creates major diagnostic difficulties.

It also deserves to be mentioned that there is an ongoing problem with the patients with various genetic disorders, which, basing on our experience, was signaled by us in many articles [14, 15]. Patients report to the dentists and do not obtain help, and in many cases, there are acute inflammations present, which cannot be postponed because of the risk of complications. Patients are always sent from dentist to another, which is confirmed by our case of a patient with Rett Syndrome. It needs do be underlined, that there are no existing procedures in such cases. That is why we can hardly blame the patients only for the condition of their oral hygiene. The dentists seeing such a problem of performing an oral surgery should refer such patients to the medical center, where adequate medical and laboratory facilities and skilled personnel are at hand. The Number 6 Central Clinical Hospital of Medical University of Lodz, where the patients underwent sanitation of oral cavity, meets those requirements. Nevertheless, the primary goal is not to allow the poor oral hygiene, which means above all caries prophylaxis.

Taking everything into consideration, we recommend that the disabled and patients with so-called rare diseases should be included into the dispensary groups.

CONCLUSION

Current situation of disabled children and those with so-called rare diseases needs a holistic approach of specialists in many fields, including the dentists. There is a need for establishing diagnostic and treatment procedures from the earliest stage of life. Also, including in the group of professionals treating patients with rare diseases, dentists is mandatory.

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