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Surgical extraction of a supernumerary tooth in a patient with Angelman syndrome - case report

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ABSTRACT:

The article discusses the problem of dental-occlusal disorders in patients with rare Angelman syndrome, based on the case report of an 8-year-old patient.

KEY WORDS: Angelman syndrome; mesiodens; extraction

INTRODUCTION:

Angelman syndrome (AS) is a rare genetic disorder that occurs with prevalence estimates of 1: 15,000 - 1: 20,000 [1,2]. The most common cause are molecular defects of chromosome 15 [2].

In the clinical picture of Angelman syndrome there occurs a significant impairment of intellectual development with speech acquisition disorders and disorders in the locomotor system, such as gait ataxia, balance disorders and tremor limbs. Characteristic symptoms include facial dysmorphia with hypertelorism, a large mouth gap, narrow upper lip, "gothic" palate, open and/or cross bite, as well as significant prognathism. The following accompanying symptoms are also observed in Angelman syndrome: abnormal, pathognomonic EEG, frequent epileptic seizures, sleep and behavioral disorders [3-6,9]. On top of the previously mentioned craniofacial disorders, the dental and radiological examination of all patients reveals: diastemata, gaps, abnormal frenulum attachments, teeth eruption disorders, supernumerary teeth and teeth with abnormal anatomy and histology [7-9]. With age, an increase in dental problems occurs, resulting from inadequate hygiene, lack of cooperation on the part of the patient, absence of dental care or accessibility to treatment under general anesthesia [8].

The article is a case report of an 8-year-old patient with Angelman syndrome who underwent surgical extraction of mesiodens in the area of teeth 11 and 21 under general anesthesia as part of one-day surgery. Supernumerary teeth are a feature occurring in over 80% of patients with AS [7].

THE CASE REPORT:

The 8-year-old patient with Angelman syndrome reported to the Department of Dental Surgery of the Medical University in Łódź for surgical consultation and possible extraction of a supernumerary tooth located in the area of teeth 11 and 21. Due to co-existing epilepsy, the patient was taking sodium valproate.

Physical examination revealed characteristics of the Angelman syndrome: delayed psychomotoric development, gait ataxia, imbalance and excessive excitability. Phenotypically, the patient showed hypopigmentation of hair, iris and skin compared to both parents, which is

specific for Angelman syndrome. Due to scoliosis and muscular hypotonia, the patient wore an orthopedic corset.

Extraoral examination revealed symptoms characteristic for the syndrome: facial dysmorphism with hypertelorism, large mouth-gap and a narrow upper lip. The intraoral examination revealed a partially erupted mesiodens, causing a pseudodiastema, an abnormal attachment of the upper lip frenulum, multiple gaps, macroglossia, teeth eruption disorders, a high-arched palate and an open bite in the anterior segment.

Due to difficult contact, lack of cooperation and the concomitant epilepsy, the patient was qualified for surgical extraction under short-term intravenous anesthesia in one-day surgery. At the Department of Dental Surgery of the Medical University of Łódź, such procedures are performed under general anesthesia. The Department is one of the few centers in Poland where surgeries are reimbursed by the NHS for patients with a disability certificate. In addition to this economic advantage, such one-day surgery allows for discharging the patient on the same day, thus reducing stress and facilitating recovery in case of patients with rare diseases or minors [8].

Before the surgery, the patient was instructed to do blood tests: APTT, complete morphology and white blood cell image. Due to the lack of cooperation on the part of the patient, it was impossible to take an OPG before the surgery. The parents were presented with the surgical treatment plan, which they accepted, and they were also given pre- and post-operative recommendations, including the mandatory follow-up visit on the day after the surgery.

During surgery under general anesthesia the supernumeral tooth was removed from the area of teeth 11 and 21. The surgical procedure and positioning of the patient during the operation was adjusted to her general condition. The extraction wound was surgically treated. After the surgery and recovery from anesthesia, the patient left the Dental Surgery Department in good general condition.

During the follow-up visit on the following day, a slight edema, normal wound healing and good general condition of the patient were observed. The convalescence took place at home under parental care, in an environment familiar to the patient. The one-day surgery allowed the minor to avoid stressful long-term hospitalization away from her family . Due to extensive malocclusion the patient remains under further care of the Department of Dental

Surgery of the Medical University in Łódź. Unfortunately, as a result of the lack of cooperation, the orthodontic treatment attempt has proved ineffective.

DISCUSSION:

Angelman's neuro-behavioral syndrome was first described in 1965 by pediatrician Harry Angelman [2,6]. In 70 to 80% of cases, the main cause is the de novo deletion in the 15q11-q13 region of the long arm of maternal chromosome 15 [5,6,10].

There are also 3 other mechanisms responsible for the occurrence of Angelman syndrome, i.e.:

1) intragenic mutations in the UBE3A maternal gene (5- 20%)

2) a defect in gene imprinting on chromosome 15q11-13q (3-5%)

3) uniparental disomy of paternal origin in the paternal chromosome 15q11- q13 (3-5%) [1,5,6].

The disease occurs with prevalence estimate ranging from 1:15,000 - 20,000 births. This number may be inaccurate due to the lack of diagnosis in the first 3 years of a child's life [1,2]. In the "happy puppet" syndrome, mutation occurs in the same chromosomal region as in Prader-Willi syndrome. The distinguishing feature for both anomalies is the place of deletion. In Prader-Willi syndrome, the deletion occurs in the 15q11-q13 region of the paternal chromosome, while in Angelman syndrome it occurs in the same area of the maternal chromosome [1,5,11]. The deletion in the 15q11-q13 region leads to the development of the clinical picture of Angelman syndrome with considerable intellectual disability, severe speech disorders and characteristic physical and behavioral features [6].

Craniofacial dysmorphism in Angelman syndrome is manifested by: microcephaly, narrow-skeletal skull, prominent cheeks, deep-set wide-apart eyes, a large mouth-gap with narrow upper lip, progeny and macrogeny. Kollemann's face harmony proportions are also impaired. The jaw section is usually elongated [8,9]. The specific morphological features can be diagnosed only at 3 years of age [2]. Intraoral symptoms include: numerous gaps and diastemas, abnormal upper lip frenulum attachments, teeth with abnormal anatomy and defects in hard tissue, supernumerary teeth, eruption disorders, malocclusion in the form of open and cross bites [7-9,13]. The most common supernumerary teeth are mesiodentes which

often disturb the correct eruption of upper incisors, thus aggravating the malocclusion [7]. Patients exhibit a high percentage of caries and its consequences [8]. The literature provides ample information on abnormalities in the structure of hard tooth tissue (enamel hypoplasia and hypomineralization) and in the anatomy, in the form of single-root single-canal first molars [8].

Due to the impaired suction function caused by a hypertrophied tongue, excessive salivation and abnormal development of the mandible the patients suffer also from gastro-intestinal reflux disease [1,13]. Holding a hypotonic tongue between the front teeth is a common parafunction [8]. Xerostomy, gingival hyperplasia, excessive salivation, glossitis, taste disturbances, or erythema multiforme may occur as side effects of anti-epileptic drugs administered to the patients. According to Gonzales-Serrano, swallowing disorders become an additional cause of excessive salivation [9].

In adolescence, the patients develop a tendency for obesity, developing into monstrous obesity in adulthood. This trait is more frequent in women [1,11]. The development of primary and secondary sexual characteristics is normal [1].

Another characteristic of Angelman syndrome is behavior manifested by: hyperactivity, disorders of sleep, concentration and facial expression, as well as laughter attacks inadequate to situation. The attacks of laughter are accompanied by uncoordinated movements of the limbs [12,13]. Sleep disturbances may occur in the form of insomnia, decreased sleep demand and total disruption of the circadian rhythm [13]. Due to the occurrence of the above behavioral disorders, people affected by AS are referred to in the literature as: "Happy puppets" or "Puppet children" [2,13,14]. The patients with Angelman syndrome have severe speech disorders while most patients can pronounce only 2-3 words. According to Larson's study, only 13% pronounce 5 words [6,17].

Communication problems cause strong attacks of aggression and an increase in frustration in people with AS, which exacerbate the concomitant mental illnesses [1]. Communication with the environment is largely limited to gestures, showing objects or paintings, or using special electrical devices [17]. Patients exhibit severe mental retardation with an IQ oscillating between 20-40 and suffer from autism [3,6,15]. 80-90% of patients are diagnosed with epilepsy between 1 and 3 years of age with frequent and prolonged seizures [3,6,13]. There is an excessive fascination with water [3,13,16].

Another symptom characteristic of this syndrome is a delay in motor development. It manifests itself in gait ataxia, balance disorders and tremor limbs of varying severity [16,17]. The characteristic way of moving is most often caused by muscular hypotension, scoliosis and flat foot [17,18]. Children start walking only between the 3rd. and 4th. year of life [11]. According to Larson's research, 68% of people with AS can walk alone [17]. In some patients with Angelman syndrome hypopigmentation of the skin, hair and iris is observed, compared to family members [2,19].

The symptoms increase with age [1]. Diagnosis can be made on the basis of the set of symptoms, but, above all, through genetic and molecular tests [18]. Unfortunately, the molecular basis has not been fully explained yet[1,5,6]. The features vary from patient to patient, and some subjects exhibit the correct phenotype and head size. The EEG is helpful in making the diagnosis [1]. According to Sachdev, in 80% of cases genetic and molecular tests can confirm the diagnosis of AS, the remaining 20% of patients are diagnosed basing on clinical features [18].

We differentiate Angleman's syndrome from Rett, Prader-Willi's and Bekwith-Wiedermann's syndrome, as well as Christanson's syndrome and X-thalassemia conjugated to the X-chormosome [1,5,18]. An early diagnosis allows for prompt implementation of treatment of coexisting epilepsy and for genetic counseling. Currently, thanks to proper choice of treatment patients survive to 60-70 years of age. This average life expectancy is only slightly shortened due to the predisposition to injury [11].

The treatment of patients with AS is multidisciplinary, requires involvement of doctors of many specializations and is focused on the treatment of concomitant epilepsy and autism as well as improving the quality of life of the patient and his family [6,18]. The sleep and circadian disturbances are treated pharmacologically and through behavioral therapies. The speech problems are solved by means of alternative forms of communication such as sound devices, pictures and gestures [6]. The early rehabilitation of scoliosis, flatfoot and joint and muscle contractures helps prevent postural defects [18].

Improving the quality of life comprises, among others, dental treatment, including dental surgery and orthodontics. The treatment of many types of malocclusions and dental abnormalities improves the functioning of the entire stomatognathic system, allows for proper food intake and has a positive effect on speech development [18]. Due to the difficult contact

and lack of cooperation with the patient, surgical treatment should be performed under general anesthesia as part of one-day surgery [8].

SUMMARY:

The treatment of patients with Angelman syndrome requires cooperation of medical specialists in various fields, including dentists and dental surgeons. The patients, due to malocclusion and oral hygiene problems, require intensive dental care. The therapy can be difficult and often ineffective. An early diagnosis allows for quick treatment and improvement of the quality of life. Due to the lack of cooperation with patients with AS, it is important to perform dental surgery procedures under general anesthesia, preferably as part of one-day surgery, which reduces stress, shortens the recovery period and allows the patient to avoid another hospitalization. In many countries treatment under anesthesia is a procedure of choice for patients with rare diseases.

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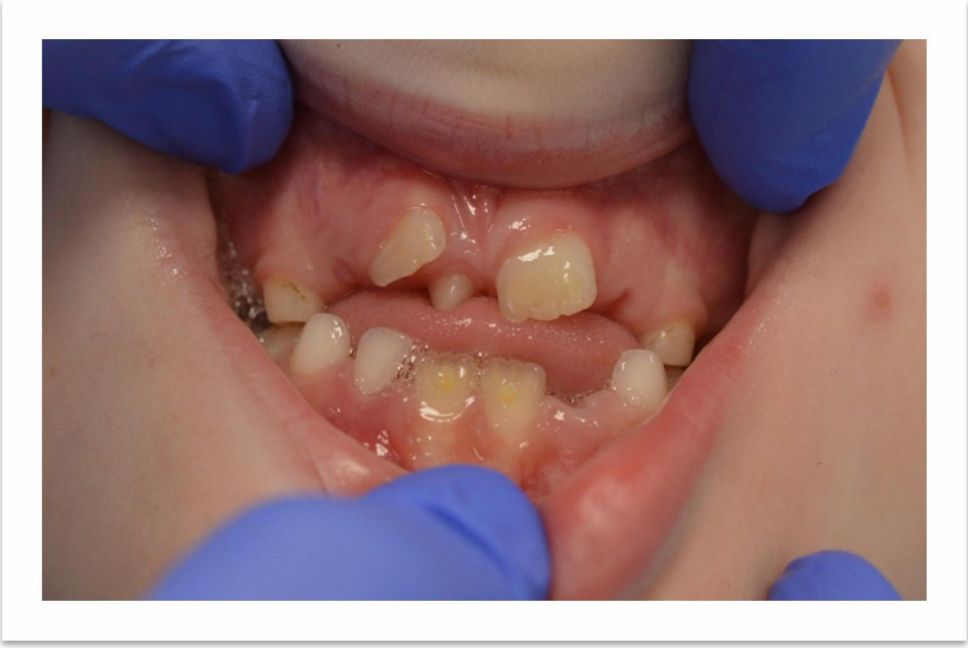

Fig 1. Intraoral examination of the patient

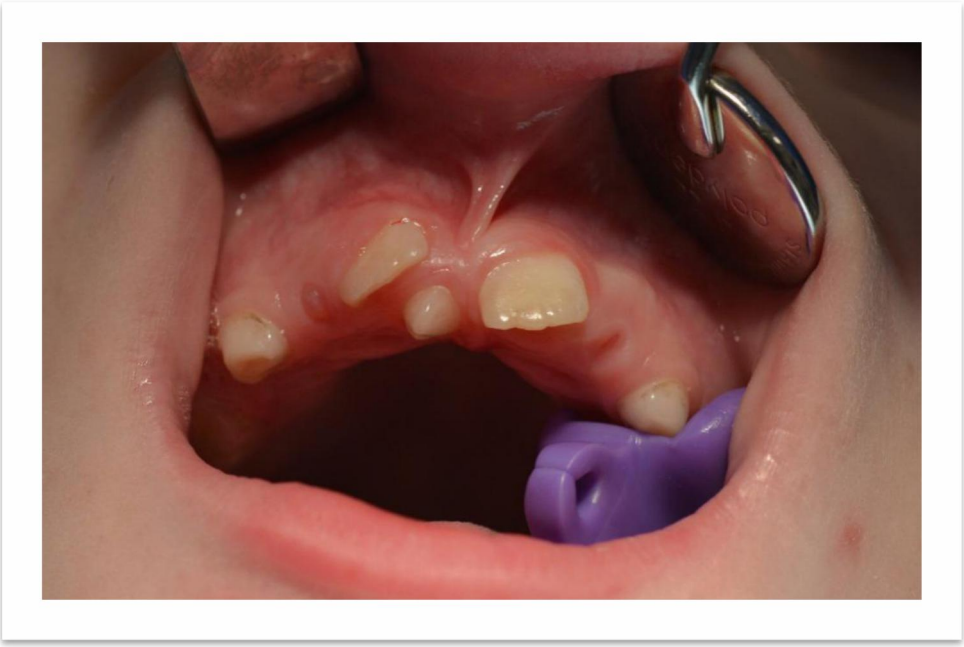

Fig 2. The visible mediodens and other dental anomalies.

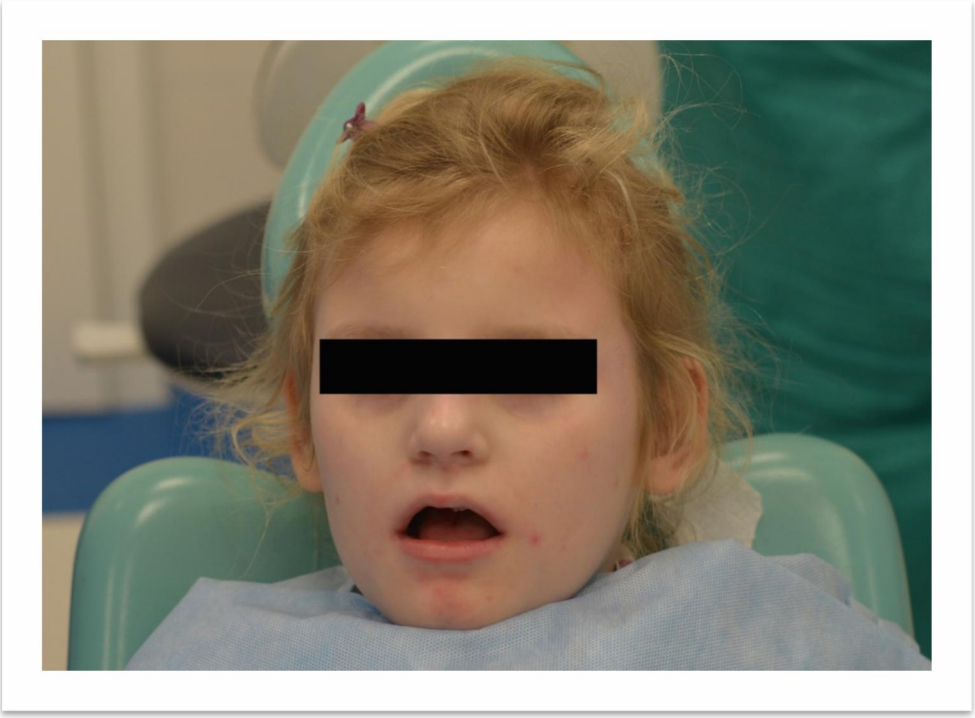

Fig 3. Extraoral examination

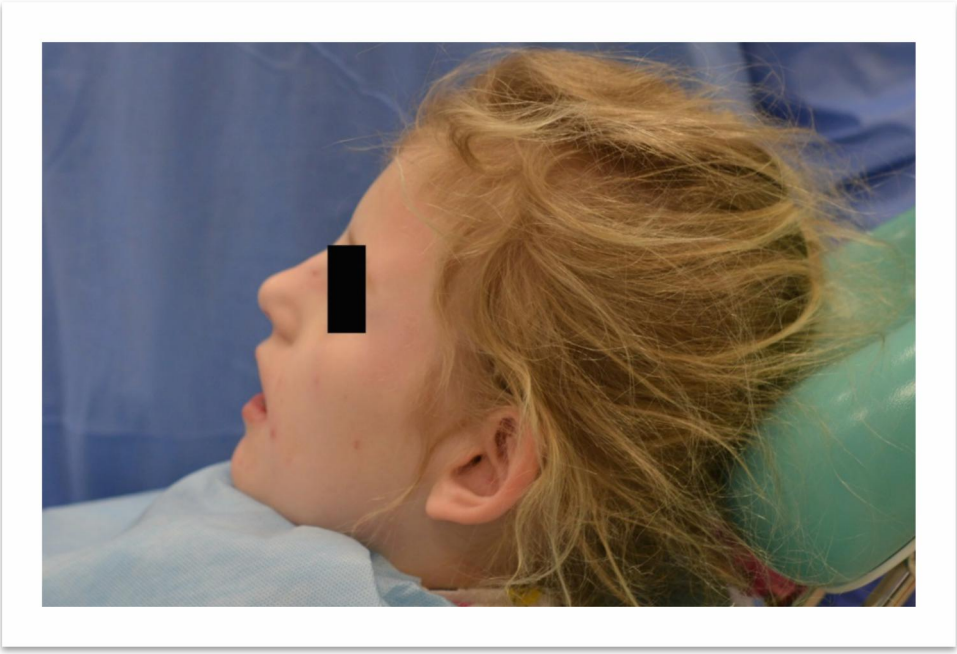

Fig 4. The patient's left profile

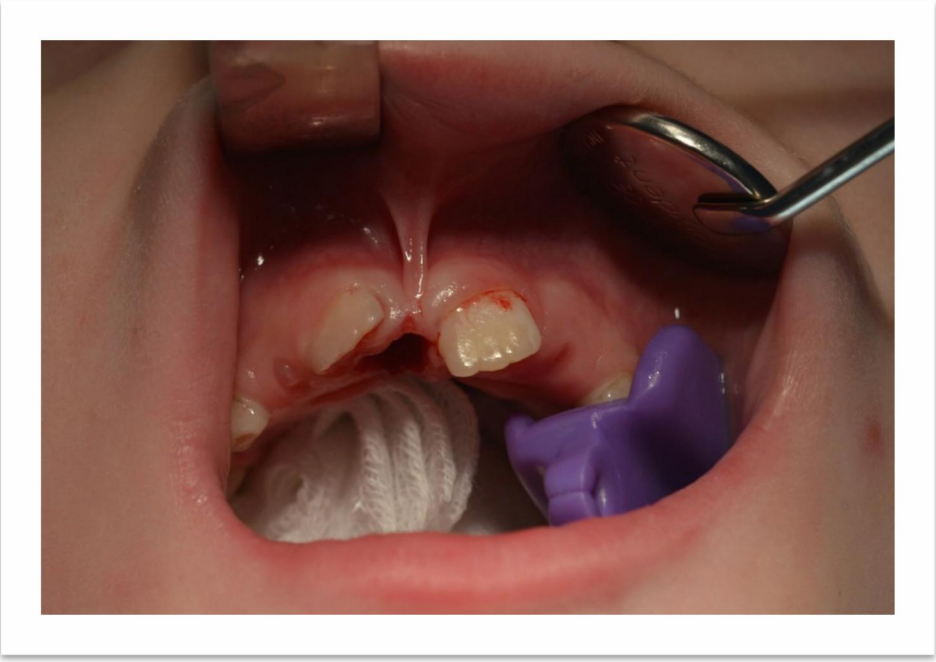

Fig 5. Condition immediately after removal of the supernumerary tooth before suturing

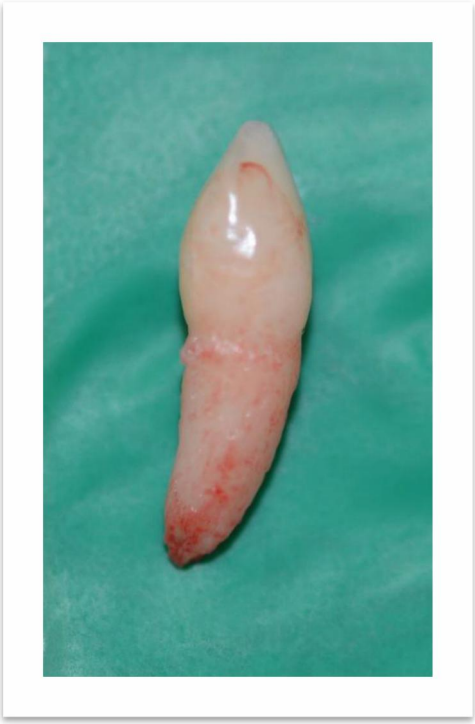

Fig 6. The extracted supernumerary tooth