SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR SYNDROME – A CASE REPORT AND LITERATURE REVIEW

Zespół pojedynczego siekacza w szczęce – opis przypadku i przegląd piśmiennictwa

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Abstract

Introduction: Solitary median maxillary central incisor syndrome is rare, with only single reports in the literature. This congenital defect affects both primary and permanent teeth. It is rarely an isolated anomaly, but usually a set of general symptoms. **Objectives:** The aim of the study was to collect reports on single incisor syndrome and analyze them in the context of our own case. **Materials and methods:** We assessed available literature reports on solitary median maxillary central incisor syndrome, including case reports. We also presented our own case based on systemic, dental, and radiological examination. **Results:** The described case was a single maxillary incisor syndrome confirmed by dental and radiological examinations. The patient presented with oral symptoms typical of the disorder, and a systemic interview confirmed extraoral symptoms. **Conclusions:** Solitary median maxillary central incisor syndrome is a rare midline defect that is usually not an isolated genetic disorder. The multitude of symptoms and disorders throughout the body requires an interdisciplinary and individualized approach to the patient.

Streszczenie

Wstęp: Zespół pojedynczego siekacza szczęki występuje rzadko. Dostępne są nieliczne doniesienia na jego temat w literaturze. Jest to wada wrodzona, która dotyczy uzębienia mlecznego i stałego. Rzadko występuje w sposób izolowany, zwykle jest to zespół objawów ogólnych. **Cel:** Celem pracy było zebranie doniesień na temat zespołu pojedynczego siekacza i prześledzenie tych doniesień w kontekście przypadku z doświadczenia własnego. **Materiał i metody:** Istniejące w literaturze doniesienia na temat zespołu pojedynczego siekacza szczęki wraz z przedstawionymi w literaturze opisami przepadków. Obserwacja pacjenta (przypadek własny) na podstawie badania ogólnoustrojowego, stomatologicznego oraz radiologicznego. **Wyniki:** Opisywany przypadek to potwierdzony badaniami stomatologicznymi oraz radiologicznymi zespół pojedynczego siekacza szczęki. Występują w nim charakterystyczne dla zburzenia objawy w obrębie jamy ustnej, a przeprowadzony jednocześnie wywiad ogólnoustrojowy potwierdza objawy występujące poza jamą ustną. **Wnioski:** Zespół pojedynczego siekacza szczęki to rzadko występujące zaburzenie w linii pośrodkowej ciała, które zwykle nie jest izolowanym zaburzeniem genetycznym. Mnogość objawów i nieprawidłowości w obrębie całego organizmu wymusza interdyscyplinarne i indywidualne podejście do pacjenta.

Keywords: solitary median maxillary central incisor; genetic disorders; dental anomalies

Słowa kluczowe: zespół pojedynczego siekacza szczęki; zaburzenia genetyczne; anomalie zębowe

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Introduction

Solitary median maxillary central incisor syndrome (SMMCIS) is a disorder affecting the structures in the midline of the body, mainly the head. It is a rare congenital defect characterized by the presence of a single maxillary central incisor. Both primary and permanent dentition may be affected. The anomaly is associated with abnormal development of the oral vestibule and face. This rare disorder may pose a diagnostic and therapeutic challenge and requires an interdisciplinary approach [1].

Epidemiology

SMMCIS is relatively rarely discussed in medical literature due to its rarity. However, isolated reports may be found [2].

The syndrome is most often diagnosed in early childhood and may affect both primary and permanent dentition. Its incidence is estimated at 1 in 50,000–100,000 births [3]. Data shows that the incidence of SMMCIS varies among different ethnic groups [3]. The epidemiology of the disorder is based mainly on individual reports and case descriptions available in the medical literature, therefore it is difficult to precisely determine the incidence of this defect and its population or ethnic distribution [3].

Etiology

SMMCIS is a rare anomaly caused by various etiological factors, including genetic, environmental, or other developmental factors. It is a set of multiple congenital defects mainly affecting the midline parts of the body. They are caused by an unknown factor [4].

SMMCIS is generally considered a genetic defect that may arise from gene mutations. Several potentially involved mechanisms have been described in the literature. The most important are:

- Missense mutations in the *SHH* gene at locus 7q36;
- Mutations in Homeobox (HOX) genes. These genes are responsible for regulating cell growth and differentiation, including teeth, during embryonic development;
- Mutations in genes that control midline facial movements. Disturbances in these genes during embryonic development may lead to defects in the structure of the frontal part of the face;
- Genetic factors related to maxillary incisors. Functional abnormalities of these genes can cause SMMCIS. A solitary incisor may be an indicator of disturbed tooth morphogenesis;
- Gene expression during the embryonic period, as confirmed by relevant research. Abnormal gene expression at a specific time and place during embryonic development may contribute to facial skeleton defects, including SMMCIS [4, 5].

Abnormal migration of mesoderm in the frontonasal process and its fusion in the midline between 35 and 38 days of fetal life are considered to be the most probable cause of the defect [6, 7].

SMMCIS may be inherited in an autosomal dominant manner. There are associations between this disorder and other syndromes and genetic defects. Authors of case reports postulate a link between SMMCIS and Pallister-Hall syndrome caused by a mutation in the *GLI3* gene. Spontaneous mutations may occur in some cases, causing SMMCIS independently of family history of defects [5].

Clinical symptoms

The presence of a single maxillary incisor in the midline is the main symptom of SMMCIS. Instead of two central incisors, patients have one tooth in the central location. The tooth has a symmetrical crown [8].

In addition to the primary and most obvious symptom, SMMCIS patients may present with many other craniofacial abnormalities. Facial dysmorphia may vary from patient to patient. The most common dysmorphic features include:

- Changes in the structure of the nose, e.g. its altered shape, a wide bridge, a rounded tip or other deviations from the correct structure, nasal malformations in the form of choanal atresia, pyriform aperture stenosis (in about 90% of patients).
- Changes in the structure of the lips, usually defects of the upper lip: altered shape, atypical vermilion.
- Asymmetries or changes in facial structure: altered shape of the cheekbones, changes in the structure of the chin in the form of micrognathia (mandibular hypoplasia).

In addition to abnormal central maxillary incisors, other dental anomalies may also occur, such as abnormal position and/or number of other teeth and hypodontia [9, 10]. Defects in the number and position of teeth give rise to occlusal problems in these patients.

In addition to craniofacial anomalies, disorders of various systems may develop, with the heart, urinary system, and nervous system most often involved [11]. SMMCIS malformations also generate problems with the auditory system, speech development, and chewing functions [12].

SMMCIS-associated middle ear defects in the form of malformations of the auditory bones or organs may impair sound conduction and affect hearing. In some patients, underdevelopment of auditory structures may occur, leading to hearing deficits, especially in childhood.

Furthermore, patients with SMMCIS present with phonation disorders. These result from abnormal craniofacial structure, including nasal hypoplasia, as well as abnormal chin and airway structure. The affected patients may also suffer from a cleft palate, which additionally compromises speech development. All these disorders may generate problems with the articulation of sounds.

SMMCIS seems to have a major impact on chewing function. Here, the potential structural effects of SMMCIS on the mandibular articular-ligamentous apparatus and the general structure of the oral cavity play an important role. Developmental defects of the oral cavity, such as dental anomalies, hypodontia, malocclusion, and palatal hypoplasia, will undoubtedly affect chewing function. All these abnormalities affect the way food is chewed. Patients have particular difficulty chewing hard foods or foods that require long chewing. This in turn generates abnormal loading of the dental system and promotes the development of malocclusions [12].

Other developmental disorders that may be associated with SMMCIS include congenital heart defects (25% of cases), including tetralogy of Fallot (15%), scoliosis (14%), esophageal atresia (10%), hypoplasia of the clavicles, anosmia, renal agenesis, hypothyroidism and chronic anterior pituitary insufficiency, brain malformations, mild to

severe mental retardation (50%). About 50% of children with SMMCI have short stature [13].

Treatment

Treatment of patients with SMMCIS requires an interdisciplinary approach. The team should consist of a dentist, orthodontist, speech therapist and otolaryngologist.

The affected patients often require orthodontic teeth alignment and bite correction. Cooperation between speech therapist, orthodontist, and otolaryngologist is necessary in the treatment of speech, breathing, and chewing disorders. Surgical intervention may be needed in some cases. Surgery is indicated in the case of structural defects of the facial skeleton [14].

Case report

A 10-year-old boy was admitted to a dental clinic for diagnostic purposes and a possible referral for treatment at a specialist clinic. His psychosomatic development was normal. He had no history of chronic conditions or regular pharmacotherapy. He developed allergies to grasses the summer, as reported by the mother (no tests were run to confirm this). According to the mother, the boy did not sustain any oral injuries in his early childhood, or the mother did not recall any. The child expressed a desire for treatment for aesthetic reasons and social problems at school.

Materials and methods

The case analysis was based on the data collected during the patient's visit to the dental clinic. Dental and general medical history, radiological documentation, as well as extraoral and intraoral radiographs were used for case description.

Results

The patient presented with the typical feature of SMMCIS in the form of a single permanent maxillary central incisor (Fig. 1). The tooth had an symmetrical crown and lacked differentiation of the incisal angles. Both angles resembled the distal angle of a normal incisor (Fig. 2 and Fig. 3).

Characteristic changes in oral soft tissues included the absence of the superior labial frenulum and the incisive papilla, whereas changes in maxillary structure included V-shaped palate. The patient also had a prominent ridge in the midline of the hard palate and width growth deficiency. External anomalies included indistinct philtrum and an arched upper lip.

Discussion

SMMCIS is a rare abnormality that usually does not occur as an isolated genetic defect [15–19]. Although some authors classify this syndrome as a separate disease entity [20, 21], SMMCIS is more often considered to accompany other developmental abnormalities, e.g. holoprosencephaly [17–22].

According to Hall, unknown etiological factors affecting the embryo between the 35 and 38 weeks gestation are

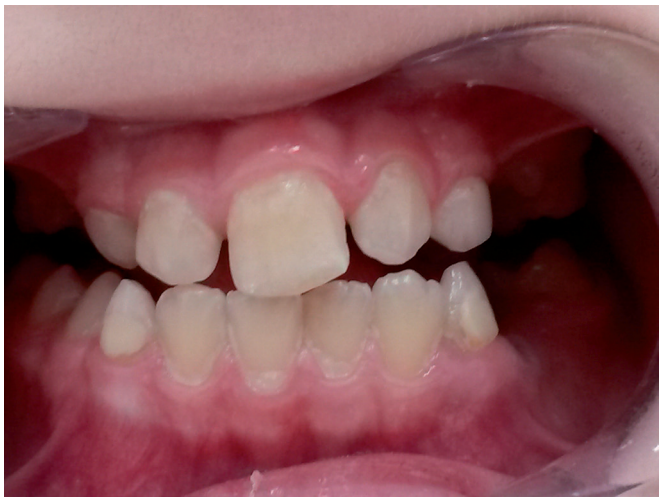

Figure 1. Extraoral X-ray showing a single maxillary incisor

Figure 2. Model of the patient's jaw showing a single maxillary central incisor

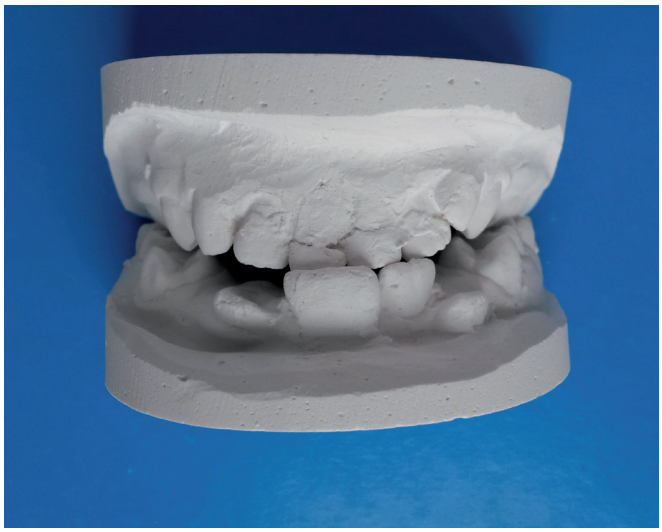

Figure 3. Comparison of the patient's maxillary and mandibular models. The panoramic X-ray shows a single central incisor located in the midline. Apart from this defect, the buds of all permanent teeth are present, including the maxillary and mandibular wisdom teeth (Fig. 4)

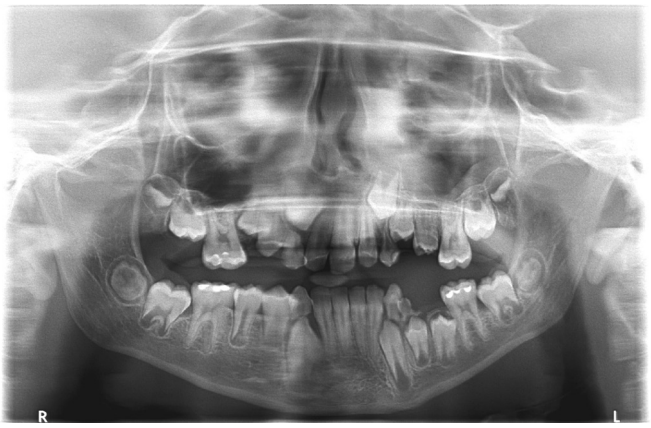

Figure 4. Patient's panoramic radiograph

the cause of SMMCIS [2]. They inhibit or slow down the transverse growth of the maxilla and the midline structures, which in turn leads to fusion of the left and right dental lamina [20]. These disorders result in the formation of a symmetrical single upper incisor, as well as impaired formation of the palatine suture and the superior labial frenulum [20].

Similarly to reports by other authors, our patient presented with clinical features indicating SMMCIS.

A reduced number of teeth is one of the basic symptoms accompanying a solitary maxillary central incisor. Hypodontia is more likely to involve permanent (1–10%) than primary teeth (0.4–0.9%) [14–16]. In the discussed case, apart from the anomaly in the form of a single central incisor, no missing teeth were observed, what is more, the buds of all molars were present. However, the shape of the upper incisor and its position in the midline of the body indicated SMMCIS.

Other symptoms typical of SMMCIS and mentioned by other authors were observed in the discussed patient: absence of the superior labial frenulum and flattened, arched upper lip without Cupid's bow [15, 20, 22]. The patient also had a short, narrow V-shaped maxilla and a prominent palatine ridge on the highly arched palate. According to Kjaer et al., these features constitute the diagnostic criterion for holoprosencephaly. This is the most severe manifestation of SMMCIS, which involves structural abnormalities of the central nervous system (CNS), the base of the skull, and the face [8].

Due to the patient's narrow jaw and mixed dentition, treatment with removable appliances could be considered, as recommended by Barcelos et al. Its aim was to improve the inclination of the central incisor [22]. According to the treatment protocol proposed by Hall, intervention is not recommended at the stage of mixed dentition, but is started after primary tooth shedding is completed. According to this protocol, maxillary expansion, shifting of central incisor and recovery of space for prosthetic reconstruction of the second central incisor are implemented [2]. Machado et al. recommend rapid maxillary expansion (RPE) in the first stage of treatment and fixed appliance in the next stage [17].

As pointed out by some authors, maxillary expansion may not proceed as planned. This is due to the lack of anterior palatal suture. Maxillary osteotomy is recommended before starting orthodontic treatment to successfully expand the maxilla [16].

Conclusions

Reports on SMMCIS are scarce due to its rarity and the limited number of cases described in the literature.

Dysmorphic features in SMMCIS may differ significantly among patients, therefore they should be diagnosed individually and carefully. Additional investigations (specialist consultations, imaging) are often needed. Only thorough diagnosis allows for identification of patient's symptoms and therapeutic needs in detail. Patients with SMMCIS should be regularly monitored by an interdisciplinary team consisting of dentists, orthodontists, speech therapists, otolaryngologists and internal medicine specialists.

Due to potential ear defects, SMMCIS patients need to be put under otolaryngological monitoring for hearing deficits. Such monitoring should be started already during the developmental period.

Regular monitoring of speech development is recommended in children with SMMCIS. Early detection of problems in this area allows for the implementation of timely and individualized speech therapy. Such intervention improves the patient's everyday functioning, and allows for proper development and learning in a peer environment.

If chewing disorders are diagnosed, appropriate therapeutic intervention should be implemented by a speech therapist or orthodontist. Early intervention in a child with SMMCIS can significantly improve their chewing and swallowing functions.

Patients with SMMCIS require interdisciplinary care. Collaboration between orthodontist, dentist, maxillofacial surgeon, pediatrician and speech therapist supports the proper physical, intellectual and social development of children.

Parental and patient education is an important aspect of treating a child with SMMCIS. The awareness of the need for regular monitoring and treatment of developmental defects and its benefits for the child's future health and functioning is crucial.

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