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## Clinical factors in prosthodontic treatment of children with genetic defects

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**Abstract****Background**

Prosthodontic treatment of children with genetic disorders is an area that is rarely examined in the current specialist literature. Few prosthodontists will undertake treatment of such patients, who will more often be referred to an orthodontic specialist. After examining the four cases of children with genetic disorders described in this paper, it can be concluded that when a prosthodontist includes a few additional procedures in the treatment process, he or she can successfully help such patients.

**Objectives**

The aim of this paper is to indicate the clinical difficulties faced by prosthodontists who undertake prosthodontic rehabilitation of children with genetic disorders.

**Material and methods**

The paper is based on data collected during the prosthodontic treatment of four children, aged 5-12 with genetic defects, and analysis of the body of work concerning these defects and their treatment.

**Results**

Presentation of guidelines for the prosthodontic treatment process and creation of dentures for treated children based on extended procedures.

**Conclusions**

A prosthodontist is a crucial person in a team of specialists treating disorders within the face among children with a genetic predisposition. A basic knowledge of orthodontics and psychology facilitates the treatment. Prosthetic restoration in the treatment group does not always require complicated operations. Individualization of the tools for downloading orthodontic impressions, designing denture elements and an increased number of checkups are the additional procedures. For the clinician, the emotional aspect of the treatment is the main impediment. Maintaining a good relationship with a patient and his or her caregivers requires interpersonal skills.

**Key words:** cardio-facio-cutaneous syndrome (CFCs), ectodermal dysplasia (ED), cherubism, prosthodontic treatment of children

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**Short title**

Prosthetic treatment of children

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**Introduction**

If not met with prompt and effective treatment, the acquired or congenital absence of teeth, originating in the developmental period, can have serious consequences in the form of dental defects, gnathic defects, and even disruption of the proportions of the whole face. Prosthetic treatment in the early developmental stages is not only meant to rebuild the lost issue, but also to stimulate toothless sections to develop properly, improve speech, aesthetics, and the chewing function, to prevent parafunctional reflexes and occlusive disorders, as well as to eliminate the trauma resulting from the loss of teeth [1].

Anodontia (total lack of teeth) or hypodontia (reduced number of teeth) often occur as one of the symptoms of the genetic diseases mentioned in this article. ED (ectodermal dysplasia) is a developmental deficit disorder of the tissues derived from ectoderm. Hair growth and sweating disorders, toothing abnormalities and nail dysplasia comprise the four clinical symptoms of this disease. The major toothing reduction and teething disorders require earlier prosthetic rehabilitation. Cherubism is a disorder that causes prominence in a lower portion in the face and progressive fibrous bone degeneration, which is the main cause of tooth formation abnormalities and the incorrect positioning of the teeth in the arch. CFCs (cardio-facio-cutaneous syndrome) is a genetic disorder that effects many parts of the body, particularly the heart, facial features, skin and hair. People with this condition have delayed development and intellectual disability. The most common conditions in the mouth region are high arched palate and maxilla size disorder (micrognathia).

Prosthetic treatment of patients with congenital dental defects begins in preschool and continues to maturity and therefore coincides with the period of intensive growth of the patient's body. When undertaking the treatment of a young patient with a genetic defect, one must take many factors into

account, such as the patient's age, symptoms associated with the genetic disease and the type of dental and gnathic disorders, as well as the relationship with the patient and his caregivers. This paper aims to provide a broad and comprehensive description of the various problems faced by a prosthodontist working to promote better functioning of the stomatognathic system.

### **Aim**

The aim of this publication is to take notice of the need for the prosthetic treatment of children who, by genetic disease, have a reduced number of teeth or have no teeth at all, namely children diagnosed with ED, CFCs and cherubism. The work depicts the specificity of the treatment of pre-school and early-childhood patients with emphasis on the emotional aspects of the checkups. Furthermore, one should also mention, both all the difficulties in the treatment process of children with developmental defects, and an attempt to show how to overcome them.

### **Material and Methods**

This paper is based on data collected during the prosthodontic treatment of four children with genetic defects and an analysis of the body of work concerning these defects and their treatment.

When discussing the prosthodontic treatment of children, specific aspects of the clinical situation need to be taken into account. When the patient is a young child suffering from a genetic disease, who comes to the clinic with his caregivers, this is considerably different than the usual scenario of a prosthodontic patient's visit. The prosthodontist needs to adapt to this situation and analyze a variety of clinical factors, such as the symptoms of the genetic disease (the degree of disorders related to chewing apparatus, disorders of muscular tension, disorders of the masticatory function), the patient's age (his level of intellectual and emotional development) and possible mental retardation which can be associated with the disease. Only a comprehensive approach to the patient can contribute to a proper execution of the prosthodontic restoration process and successful rehabilitation.

A group of four children was observed, which was divided into 2 age groups (preschool age and young school age). This division was based on the classification used in developmental psychology.

“When researching the psychological development of a child, the following development periods can be distinguished: infancy, postinfancy, preschool age, junior school age and adolescence” [2]. According to Joanna de Flassilier-Popławska, “knowledge of subsequent stages, especially developmental changes in cognitive and emotional spheres, allows to better understand a child patient’s situation and take it into account in the course of treatment” [2].

Impulsive action and emotional instability are characteristic for preschool age (3-7 years). Children at this age require help from adults, especially emotional support in new and unfamiliar situations. They like to expand their knowledge about the world around them by asking frequent questions. The prosthodontist should be aware of that and therefore should allow the child’s caregivers to accompany the child during visits to the dentist’s office, fully explain all the procedures that will be used in the course of treatment, answer questions about the tools and instruments used and attempt to minimize the child’s fear by speaking in a calm and friendly voice as well as acting in a firm and confident manner. To gain the trust and respect of the child patient, the clinician should communicate both with the child and his caregivers. Subsequent visits should be scheduled during morning hours when the patient is well rested. Particular procedures that are done during the visit preferably shouldn’t last longer than 20 minutes.

It is recommended that the prosthodontist presents the outline of the treatment plan to caregivers as well as clarify all their doubts during the first visit. This is important as caregivers’ attitude, positive or negative, will affect the child’s attitude towards the treatment process. “The behavior of people accompanying the child plays an important role in modeling his behavior in the dentist’s office. For example, fear, anxiety or a negative attitude to the doctor shown by caregivers is later imitated by the child” [2].

However, it has been observed that there is a different kind of behavior among school children (7-12 years old), who quit relying solely on their parents' opinion. At this stage, the peer group’s opinion becomes important to the children. Any deviations are not tolerated, and the importance of physical appearance grows. Patients in this age group cooperate rather well during the dental treatment, as their

motivation is higher. They are also able to understand the purpose of the treatment and have better control over their emotions. “The way they function in both cognitive and emotional spheres enables good cooperation with the dentist” [2].

## **Results**

### **Subject 1**

Julia, a 5-year-old girl diagnosed with cardio-facio-cutaneous syndrome, was referred to the Department of Prosthodontics, Wroclaw Medical University, in January 2014. “As with other dysmorphic syndromes the diagnosis of CFC syndrome depends upon a characteristic picture rather than any specific diagnostic test. Demonstration of the underlying genetic abnormality requires sophisticated methodology beyond the scope of a standard clinical or genetic laboratory” [3]. “Therefore clinically, CFC syndrome is a RASopathy characterized by craniofacial, dermatologic, gastrointestinal, ocular, cardiac and neurologic anomalies” [4]. The disease is a result of de novo mutations, i.e. a new dominant gene being added to the gene pool. Research has identified shared characteristics of craniofacial deformities that included macrocephaly, bitemporal narrowing, convex facial profile and hypoplastic supraorbital ridges [4]. Dental phenotype characteristics for CFC include malocclusion with open bite, posterior crossbite and high-arched palate [4].

It was necessary to provide Julia with a removable dental prosthesis, since she had prematurely lost deciduous teeth due to their disturbed structure. Intraoral examination revealed the presence of four teeth in the maxilla deciduous canines and secondary molars (Figure 1). Prosthodontic restoration was meant to improve the ability to chew food as well as the aesthetics of the face, which was also significant because Julia attended preschool and had been in contact with other children.

Initial difficulties were encountered while attempting to take a dental impression during the first visit, as contact with Julia was limited due to the medium degree of mental retardation. Despite being 5 years old, the patient behaved as a 1.5 years old child. Under these circumstances, the doctor had limited possibilities to explain the course of treatment to the patient, therefore he needed to gain her trust through acting in a calm and confident manner. During each visit, the patient sat on her mother’s lap, which

made her feel safe and secure. Cooperation with her parent was an important factor making the treatment process easier and more efficient. Julia's mother was calm, strong-minded and motivated to help her child. She was not afraid to ask questions related to the stages of the treatment process and she acted as a good intermediary between her child and the doctor, making the communication process more efficient. She was a model example of a parent's behavior in the prosthodontist's office. The success of the treatment process was largely dependent on gaining the mother's favor.

Determining the central occlusion and taking all the measurements required for the denture was a big challenge, as the patient manifested excessive nervousness. The doctor had to be patient and composed, but at the same time carry out all the necessary procedures quickly and efficiently. After making a partial denture, the last step which proved to be difficult was checking if it fit correctly as well as convincing Julia that the denture should stay in her mouth and be used as if it were her own teeth. In most cases it is difficult to convince an adult that the denture adaptation period is cumbersome and painful, but only temporary. To convince a child, it's recommended to present treatment as fun and attractive event. Julia's denture required Adams' braces for additional support, because initially the patient tried to remove the denture from her mouth, as she didn't understand the purpose of the treatment (Figure 2). Additionally, muscular hypotension could have an adverse effect on the way the denture was fixed in the mouth.

Unfortunately, lack of cooperation from the patient's side made it impossible to make a panoramic X-ray, which would make it possible to plan further treatment. Julia's denture should be checked every 2 months and if the need arises, should be adjusted with a Fischer screw to fit it to bone development dynamics or alternatively should be replaced. During follow up visits, the doctor should always check the oral hygiene and provide the family with detailed instructions concerning hygiene habits, because the patient, suffering from the aforementioned genetic disease, may not be able to take care of that herself [4]. Furthermore, occlusion development should also be monitored and, if necessary, the patient should be referred to an orthodontist for further treatment.

## Subject 2

Young patients that visit the Prosthodontics Clinic most frequently are children diagnosed with ectodermal dysplasia. Cwajda describes this disease as “syndromes of disorders caused by anomalies in ectodermal structures”. She goes on to say that “154 syndromes related to ectodermal tissues were identified, 120 of which result in symptoms in the mouth area, mainly hypodontia” and that “[s]ymptoms characteristic of ectodermal dysplasia include: frontal bossing, collapsed middle third of the face, saddle nose, thick and everted lips, low-set ears called satyr ears, fair eyes, wide set orbits” [5]. Labda et al. Have also shown that small children with ED often have problems with speech and eating due to missing teeth or anodontia [6]. They also feel that they are different from their peers [6]. Self-awareness connected with physical appearance can start to form when a child is 4-5 years old, therefore it is important to start aesthetic as well as prosthodontic procedures around that age [7].

Dawid (aged 5), came to the clinic for a new denture for the maxillae and mandible, because he had lost the previous one. After a meticulous oral examination and careful analysis of anatomic factors, a decision was made to create removable partial acrylic dentures for the maxillae and mandible. “The use of partial and complete acrylic prostheses is an interesting and practical alternative that provides a relatively quick, easy, acceptable and economical solution to the functional aesthetic oral rehabilitation and psychological benefit in young patients with pronounced edentulism” [8]. This should only be treated as a temporary solution however, and parents should be informed about the other treatment possibilities given by implant prosthodontics, which can be applied in the future, after the child’s developmental stage is completed [7]. Dawid was cooperating well considering his age, however he was also having mood changes.

Taking his dental impression during the first visit was difficult as it was not possible to fit standard children’s trays to his anatomy. The small size of the maxillae and mandible caused by missing teeth and underdevelopment of the alveolar process is one of the disease’s symptoms. In order to achieve an accurate impression of the prosthetic field as well as prosthetic abutment (2 primary maxillary second molars and 1 primary mandibular second molar), the decision was made to create custom trays (Figure

3) after taking anatomical impressions using standard children trays. The teeth remaining in the mouth were relatively small sized. Additionally, due to prior lack of full adaption to the denture, Adams' braces were used as supplementary supporting elements.

Patients with diagnosed ectodermal dysplasia can have denture adaptation problems related to the difficulty in achieving retention and stabilization, as this disease is often accompanied by dry mucosa and underdevelopment of maxillary tuberosity and the alveolar process [9]. It's recommended to widen the base of the denture for such patients in order to make the denture fit the mouth better [9]. Dawid's treatment required performing additional procedures, scheduling visit hours optimal for the child's good mood in order to improve cooperation, explaining the course of treatment and applying additional denture elements - Adams' braces and Fischer's screws (Figures 4 and 5). Thanks to adjusting the treatment to suit the specific needs of the child, prosthetic rehabilitation was successful at this development stage. "Long term success, however, depends on the patient's regular follow-up visits and maintaining meticulous oral hygiene" [9]. Follow-up visits were schedule for Dawid every two months.

### **Subject 3**

Bartek came to the Prosthodontics Clinic at 12 years old, at rather late age. He had been diagnosed with ectodermal dysplasia a year earlier. The patient came to the Genetics Clinic as his parents were worried about his hypodontia and nail dysplasia (Figure 7). During the interview, it was established that the deciduous teeth had erupted correctly, just when the patient was 11 years old and after he had lost his lower incisors, there were no permanent teeth. Panoramic X-ray showed that the patient lacked most of the tooth buds for permanent teeth (Figure 6). The remaining deciduous teeth were not developing at the same rate as the rest of the body, therefore Bartek required overdenture to replace the missing teeth and restore the proper height of the occlusion (Figure 8). Using Fisher's screw in the denture made a longer use of the restoration possible, as it could be increased along with the facial skeleton. The course of the treatment was good, the patient was motivated and cooperated well. Bartek's treatment was aimed to restore the balance in the stomatognathic system as well as improve the smile's aesthetics, which is an important factor for a patient at that age (Figure 9). "Well-fitting and functioning prosthesis could be a

great help during their schooling years as it will improve appearance and thus boost their self-confidence” [6].

#### **Subject 4**

9 year old Dawid, who had been diagnosed with cherubism, came to the Prosthodontics Clinic in 2014. Yilmaz et al. report that “Cherubism is a rare inherited developmental abnormality that causes bilateral enlargement of the maxilla and/or mandible”. Moreover, “[i]t is caused by the autosomal dominant gene located on chromosome 4p16.3 and typically affects men. It is generally accepted that it is a benign disease of bone beginning at the age of 2-3, that progresses in childhood with a peak at the age of 5, and shows spontaneous regression at the end of adolescence” [10]. Published sources report that, depending on the severity of the bone changes, the effects the disease has on the mouth can include dislocation of teeth, dental crowding, root resorption, premature loss of deciduous teeth, tooth agenesis and tooth bud involution [11][12]. Dental abnormalities and phonation disorders as well as chewing and swallowing disorders can affect patients with the aggressive form of cherubism [11][12].

A report from Dawid’s craniofacial CT from 2010 indicated the extensive destruction of both maxillae and mandible, which were transformed into a sponge-like structure and expanded. These changes did not affect either mandibular condyles nor zygomatic bones. They had the biggest impact on the body of the mandible, which displayed significant bilateral enlargement. Conducting a thorough interview revealed no family history of the disease in the patient’s family. His mother said that the first alarming signals appeared when Dawid was 3.5 years old, when he started to lose deciduous teeth prematurely. A panoramic radiograph of Dawid’s mouth confirmed a lack of tooth buds for some of the permanent teeth and also revealed dislocation of the remaining teeth, which was caused by the deformed bone structure of the maxilla and mandible (Figure 10). It also showed teeth located in radiolucent areas, which is a characteristic image for this anomaly called “floating teeth” [11][12]. The disease has not affected the boy's mental development.

During subsequent visits to the clinic, a functionally unstable denture with Fischer’s screw and other orthodontic elements helping proper tooth eruption was created (Figure 12). It was challenging to obtain

stabilization of the lower denture due to the sponge-like alveolar process of the mandible with a flat bottom of the vestibule and sublingual space (Figure 11). Low, dislocated abutment teeth, caused by the disease, proved to be an additional difficulty. The course of Dawid's prosthodontic treatment will last several years. Currently, it is planned to achieve contact of the teeth and restoring the aesthetics of the smile (Figure 13). Further steps include removal of the deformed bone tissue along with the retained teeth, transplantation of the removed bone and probably creating an overdenture type prosthesis.

This patient's treatment is a complicated and multi-stage process, but thanks to the proper motivation, determination and willingness to cooperate from him, satisfactory effects of the treatment can be expected. In the case of patients with such diseases, it is important that the dental prosthetist knows both the clinical and radiological symptoms of the disease, because he may be the first person to diagnose the patient when the patient visits the clinic wanting to improve the aesthetics of the teeth. The doctor should recommend further diagnostics in a genetics clinic after the interview with the patient and intraoral examination and X-ray interpretation.

## **Discussions**

The prosthodontic treatment of children is not a well-developed field. Often doctors who are specialized in prosthodontics tend to leave the treatment of such patients to orthodontists. But children suffering from genetic disorders often require medical assistance from a prosthodontist, who can additionally include orthodontic knowledge in his actions. A prosthetist ought to belong to the group of specialists undertaking the rehabilitation of the masticatory system of such patients. After analyzing the treatment of the above-mentioned children, it is worth noting that doctors undertaking the treatment of a young patient should have the necessary qualities. He or she should have proper interpersonal skills, be well prepared and have the right attitude. Interpersonal skills are important in order to establish good contact with the child and his caregiver. Good preparation includes having a basic knowledge regarding orthodontics, pediatric dentistry and psychology. When it comes to the right attitude, we must highlight a comprehensive look at the case and an open mind to new challenges. In the present examples, dentures were made for children taking part in clinical monitoring. This leads to the conclusion that it is not a

difficult treatment, but it requires some additional actions and is more time-consuming. Another conclusion that's worth noting is the importance of cooperation between the doctor and patient's guardians. Successful treatment of the patients described in this paper was often possible thanks to a good relationship between the prosthodontist and the child's parents, who presented a positive attitude towards the process. In 1983, Murray stated that "cooperation between the doctor and guardians is a fundamental condition for dental treatment. After all, it's parents that shape hygienic and dietary habits of the child, as well as take care of prevention and bring child to subsequent visits" [2]. **[Murray, 1983 is not included in the list of References, and it looks as if the quote belongs to Murray, but then a citation [2] for de Flassilier-Poplawska is given. Which is it?]**

### **Clinical Implications**

All the children described in this paper accepted their dentures and are using them, despite considerable difficulties with making the restoration and a long adaptation period. They also remain under systematic prosthetic and orthodontic care and report regularly to the designated check-ups.

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Figure 1. Patient without artificial replacement

Figure 2. Patient with partial denture

Figure 3. Functional impressions of maxilla and mandible on individual trays

Figure 4. Acrylic denture for mandible with Fisher's screw and Adam's braces, lingual view

Figure 5. Denture for maxillae with Fisher's screw and Adam's braces, lingual view

Figure 6. Panoramic X-ray showing missing permanent tooth buds

Figure 7. Distorted structure of the nails - one of the ectodermal dysplasia symptoms

Figure 8. Patient without denture, lowered occlusions visible

Figure 9. Patient with overdenture for maxilla and mandible

Figure 10. Panoramic X-ray showing distorted structure of mandible bones and dislocation of teeth

Figure 11. Mouth without denture, wide and irregular alveolar process of mandible and elevation of the bone in the sublingual space as well as decreased depth of mouth vestibule visible

Figure 12. Denture with orthodontic elements

Figure 13. Patient wearing maxillary and mandibular dentures