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OPIS PRZYPADKU
CASE REPORT

Long-term, complex orthodontic treatment of patient with Apert syndrome – from severe malocclusion to functional and aesthetic result

Długoterminowe, wieloetapowe leczenie ortodontyczne pacjentki z zespołem Aperta – od nasilonej wady zgryzu do funkcjonalnego i estetycznego rezultatu

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ABSTRACT

This case report describes the orthodontic treatment of a 2-year-old female with Apert syndrome, initially admitted to the Clinic of Congenital Abnormalities in the University Dental Centre of Medical University of Silesia in Katowice in 2004. Following craniofacial surgery for premature skull fusion, the patient exhibited characteristic Apert syndrome features, including premature fusion of skull bones, midfacial hypoplasia, and syndactyly. Removable appliances were used between 2007 and 2013 to manage dental development and teeth loss, followed by craniofacial osteoplasty in 2013––2014 to correct significant maxillary underdevelopment. Fixed orthodontic treatment was initiated in December 2014, focusing on aligning teeth, correcting malocclusion, expanding the upper arch, and managing crowding. Despite treatment challenges, such as poor oral hygiene the 6-year orthodontic treatment yielded a satisfactory functional and aesthetic outcome. The patient achieved correct overjet and overbite, reduced crowding, and improved jaw relations, though some occlusal problems, including a residual posterior crossbite and minor crowding, persisted. Almost 4-year follow-up demonstrated stable results, although bruxism was developed, requiring a nightly splint. Continued follow-up is essential for managing long-term stability in this complex case.

KEYWORDS

Apert syndrome, orthodontic treatment, acrocephalosyndactyly, craniosynostosis, multidisciplinary treatment

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STRESZCZENIE

Prezentowany opis przypadku dotyczy leczenia ortodontycznego dziewczynki z zespołem Aperta, przyjętej do Poradni Wad Rozwojowych Uniwersyteckiego Centrum Stomatologii Śląskiego Uniwersytetu Medycznego w Katowicach w 2004 r. w wieku dwóch lat. Przed rozpoczęciem leczenia ortodontycznego u pacjentki przeprowadzono operację w celu korekty przedwczesnego zrośnięcia szwów czaszki. U pacjentki obserwowano charakterystyczne cechy zespołu Aperta, w tym przedwczesne zrośnięcie kości czaszki, niedorozwój środkowej części twarzy oraz syndaktylię. W latach 2007–2013 u pacjentki zastosowano aparaty ruchome w celu kontrolowania rozwoju zgryzu oraz uzupełnienia braków zębowych. W latach 2013–2014 wykonano osteoplastykę czaszkowo-twarzową w celu skorygowania znacznego niedorozwoju szczęki. Leczenie ortodontyczne aparatami stałymi rozpoczęto w grudniu 2014 r., koncentrując się na uszeregowaniu zębów, korekcie relacji przednio-tylnej szczęk, poszerzeniu górnego łuku zębowego oraz rozładowaniu stłoczeń. Pomimo trudności, takich jak niedostateczna higiena jamy ustnej, leczenie ortodontyczne przyniosło zadowalające efekty zarówno funkcjonalne, jak i estetyczne. Pacjentka uzyskała prawidłowy nagryz poziomy i pionowy, w znacznej mierze rozładowano stłoczenie zębów oraz poprawiono relacje szczęk. Niemniej jednak pewne problemy zgryzowe, w tym zgryz krzyżowy boczny oraz niewielkie stłoczenia, utrzymywały się po leczeniu. Prawie 4-letnia obserwacja efektów leczenia wykazała stabilne wyniki, choć u pacjentki rozwinął się bruksizm, co wymagało zastosowania nocnej szyny relaksacyjnej. Dalsza kontrola pacjentki jest niezbędna do utrzymania długoterminowej stabilności efektów leczenia.

SŁOWA KLUCZOWE

zespół Aperta, leczenie ortodontyczne, akrocefalosyndaktylia, kraniosynostoza, leczenie interdyscyplinarne

INTRODUCTION

Apert syndrome, also known as acrocephalosyndactyly type I, is a rare genetic disorder, with the incidence estimated to be 1 in 65.000 to 88.000 live births [1]. Apert syndrome is characterized by craniosynostosis (premature fusion of skull bones, which leads to deformation and disfunction of head structures) and syndactyly (fusion of fingers and toes). It was first described by French physician Eugène Apert in 1906. The condition is caused by mutations in the fibroblast growth factor receptor 2 (FGFR2) gene, leading to abnormal development of bones and other tissues during foetal development [2].

The primary clinical features of Apert syndrome include craniofacial abnormalities such as a high forehead, shallow eye sockets, midfacial hypoplasia, and underdeveloped maxilla, often resulting in restriction of airway and multiple dental problems [2]. Additionally, patients usually exhibit complex syndactyly, usually affecting all fingers and toes, which may significantly impair hand function. Neurological issues such as developmental delays and cognitive impairment can also be observed due to the premature fusion of the skull bones affecting brain growth [3].

The management of Apert syndrome is multidisciplinary. Diverse problems often require surgical intervention, e.g. to correct craniosynostosis, syndactyly, and other associated anomalies. Early diagnosis and treatment are essential to improve outcomes and quality of life for patients with Apert syndrome [4]. This case report discusses the clinical presentation and multidisciplinary treatment approach of a patient with Apert syndrome. The case report is emphasizing the challenges and long-term care considering malocclusions, functional and aesthetic outcomes during orthodontic treatment of individual with this rare congenital disorder.

CASE REPORT

A 2-year-old female was admitted to the Clinic of Congenital Abnormalities (Zabrze) in the University Dental Centre of Medical University of Silesia in Katowice in February 2004 to initiate diagnostic and orthodontic treatment due to Apert syndrome. Prior to the visit in Clinic of Congenital Abnormalities, patient had a craniofacial surgery in the Department of Plastic Surgery in Specialized Medical Centre in Polanica Zdrój (24 September 2003), to manage premature fusion of skull bones. Unfortunately, patient was not referred to Clinic of Congenital Abnormalities right after birth, so there was no medical history prior to February 2004, apart from hospital documentation provided by the parents. Initial examination revealed edentulous arches, pseudo-prognathism, concave profile and insufficient maxillary development. Characteristic features of Apert syndrome were observed: high forehead and midfacial hypoplasia as well as syndactyly, affecting both fingers and toes (Figure 1). Patient was under observation for several years, during early childhood and the period of deciduous teeth eruption (from 2004 to 2007). In May 2007 due to poor oral hygiene and numerous caries lesions patient was referred to paediatric dentist for general oral sanitation (Figure 2). After sanitation only canines and second molars were present in patient's mouth and it was decided to engage removable, child's prosthesis in the treatment process, which was replacing missing teeth. Patient was using removable prosthesis from 2007 to 2013. New appliances were created throughout that time in order to accommodate

patient's growth and eruption of permanent teeth. In February 2012, lower Schwarz appliance with Fisher's screw, Adam's clasps and short labial bow was included in treatment process. Patient's guardian was instructed to expand appliance once a week.

In 2013 and 2014 patient received series of two surgeries in Dallas, USA, in order to perform craniofacial osteoplasty. Serious maxillary under-development was corrected, facial features were significantly improved, and patient's profile was no longer concave (Figures 3 and 4).

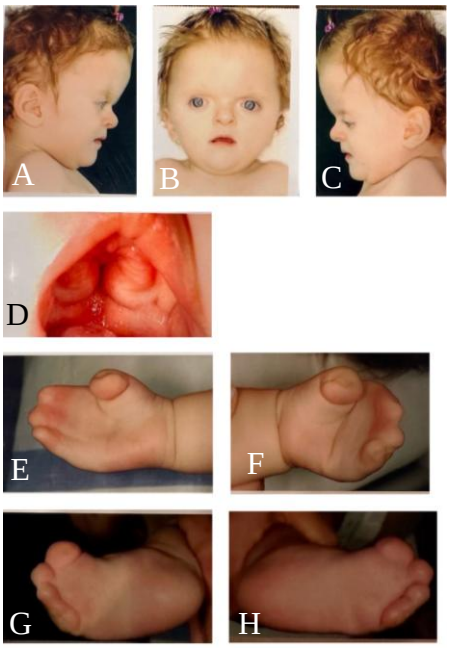

Fig. 1. Patient aged 2 years, during initial visit at Clinic of Congenital Abnormalities. Figures A–C present patient's face and profile before any orthodontic interventions, figure D presents cleft palate, figures E–H show hands and feet with syndactyly before surgical intervention.

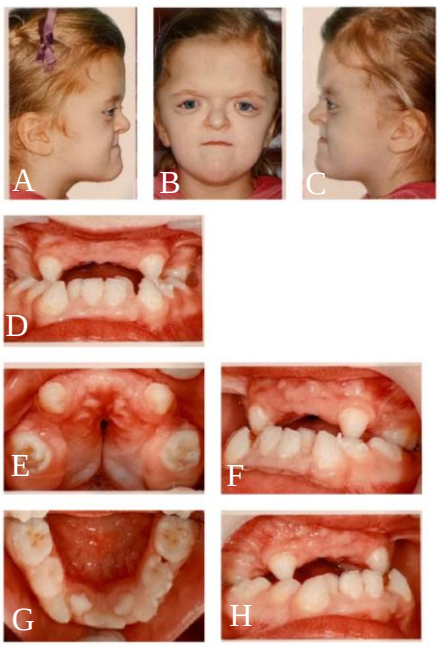

Fig. 2. Patient aged 5 years. Figures A–C present patient's face and profile in early childhood period, figures D–H show teeth and occlusion in deciduous dentition after general dental sanitation.

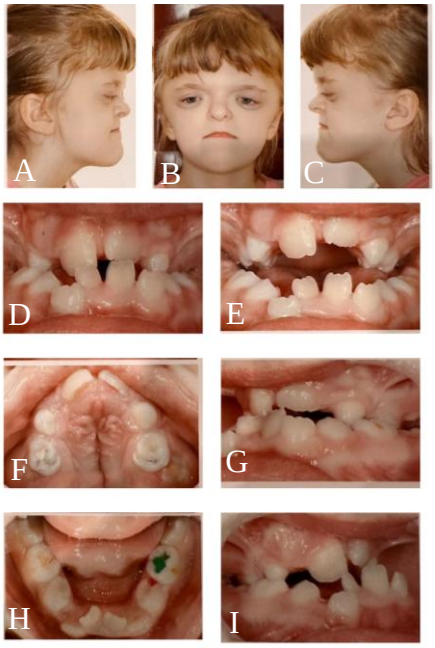

Fig. 3. Patient aged 10 years. Figures A–C present patient's face and concave profile before surgical interventions, figures D–I show teeth and occlusion in mixed dentition period, crossbite, severe crowding and reverse overjet is visible.

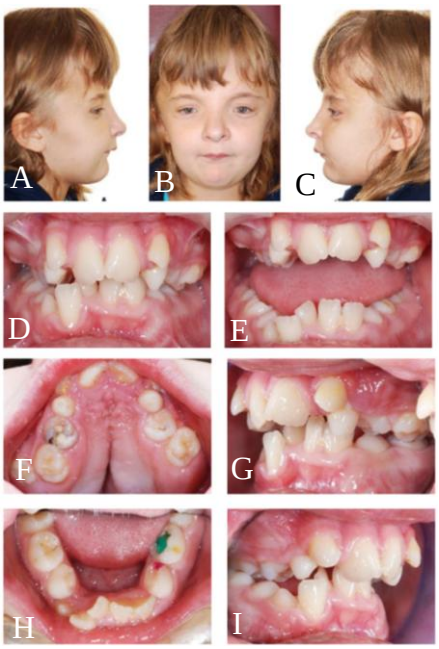

Fig. 4. Patient aged 12 years. Figures A–C present patient's face and convex profile after surgical interventions, figures D–I show teeth and occlusion in mixed dentition period, crossbite and severe crowding persist. However, correct overjet is now visible.

In December 2014 treatment plan was created based on panoramic radiograph, cephalometric radiograph and cephalometric analysis (according to Bjork's method), patient was 11 years 10 months old at the beginning of the treatment with fixed braces. The main treatment objectives were:

- aligning teeth in upper and lower arch
- correction of jaws relation, patient after surgical treatment (craniofacial osteoplasty) presented skeletal II class malocclusion
- space gain and management for severely crowded lower incisors
- expansion of upper arch and management of crowded and palatally erupted permanent teeth in maxilla, first left upper premolar in 180° rotation
- correction of right lateral crossbite
- correction of incisors relation: overjet and overbite
- aesthetical and functional improvement of patient's occlusion.

The treatment with fixed braces (0.022″ MBT (McLaughlin-Bennett-Trevisi) prescription) in the upper arch was initiated on 15 December 2014. The brackets on teeth 14, 12, 11, 21, 22, 24 and bands on 16, 26 were bonded. Fixed braces in lower arch were added on 25 February 2015, initially engaging also deciduous teeth present in patient's mouth, successively replaced with permanent dentition. The main objectives which determined the time of treatment were: complete lack of space and 180° rotation of first left upper premolar and palatally positioned right canine (Figures 5 and 6). Active treatment with fixed braces finished on 4 December 2020 and lasted almost 6 years. At the end of treatment patient aged 17 years 10 months. After removing fixed braces clear, removable retainers were created (Figure 7).

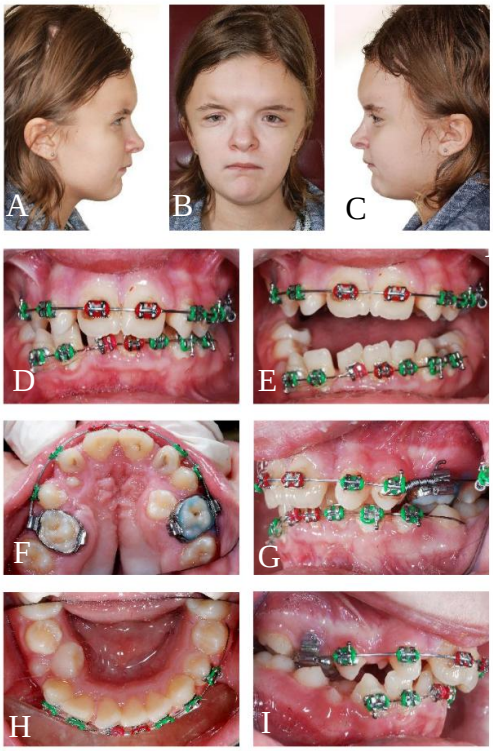

Fig. 5. Patient aged 13 years. Figures A–C present patient's face and profile after first few months of orthodontic treatment, figures D–I show teeth and occlusion after levelling phase.

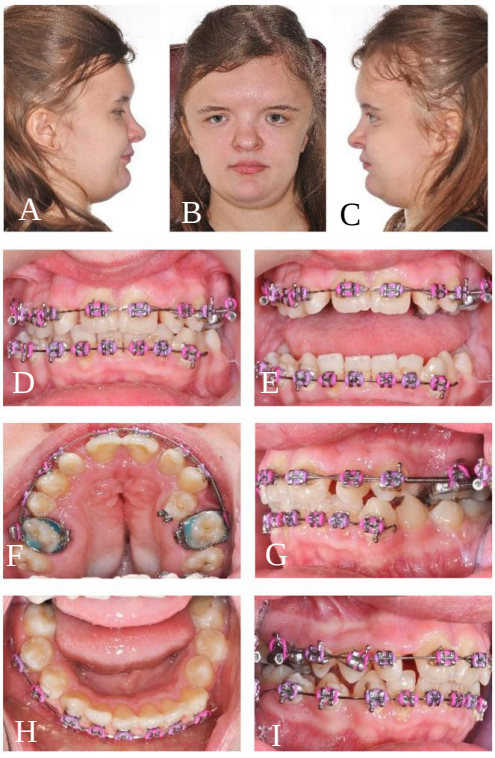

Fig. 6. Patient aged 16 years. Figures A–C present patient's face and profile during active orthodontic treatment with fixed braces, figures D–I show teeth and occlusion during finishing phase of orthodontic treatment.

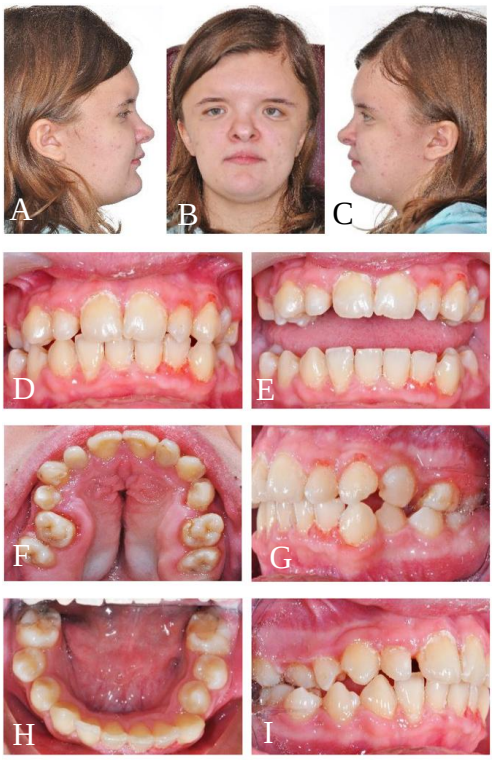

Fig. 7. Patient aged 17 years. Figures A–C present patient's face and profile after orthodontic treatment, figures D–I show teeth and occlusion with braces removed.

The outcome of the treatment is not ideal however, rather satisfying, considering all limitations and complexity of presented case. At the end of the treatment patient had correct overjet and overbite, moderate correction of right lateral crossbite (first right upper molar remained in crossbite), no severe crowding in upper and lower arch (however, some minor crowding still persist in lower arch), right canine aligned within upper arch, first left upper premolar partially derotated and aligned within upper arch, as well as acceptable jaws relation, satisfactory aesthetic and function. Nevertheless, due to the lack of second upper premolars, I Angle's class was impossible to achieve. The main impediment throughout treatment process was poor oral hygiene, despite numerous schoolings and oral hygiene instructions. Furthermore, frequent mechanic failures of bonded elements and wires leaded to elongation of active treatment process. Finally, orthodontic treatment was financed from the Polish National Health Fund (Narodowy Fundusz Zdrowia – NFZ). Patient was included in the governmental programme of orthodontic care for children with congenital defects of the facial skeleton. Even though the programme offered wider range of sponsored treatment methods compared to treatment of children who did not present any defects, some innovative solutions were not sponsored and therefore could not be included during treatment. NFZ financing was a serious limitation which played a key role during creation and realization of treatment plan which based on accessible appliances and protocols.

During almost 4-year follow-up occlusion and treatment results were stable. Patient had few sets of retainers done during retention period. Quick destruction of retainers indicated that bruxism may occur, especially during the night. In 2022 patients was diagnosed with bruxism and she is currently using nightly upper mouth splint. Patient is still visiting Clinic of Congenital Abnormalities every six months for follow-up.

DISCUSSION AND CONCLUSIONS

Apert syndrome is a rare congenital disorder characterized by craniosynostosis, syndactyly, and midface hypoplasia. It presents a significant challenge in orthodontic management. Patients with this condition typically exhibit a complex combination of craniofacial and dental anomalies, including severe class III malocclusion, narrow dental arches, often accompanied by a posterior crossbite, anterior open bite and crowding, necessitating a multidisciplinary approach to treatment [5]. The timing of orthodontic treatment in Apert syndrome is critical and must be well coordinated with planned surgical interventions [6]. Early orthodontic treatment is often delayed due to the need for cranial surgeries aimed at correcting craniosynostosis, which typically take priority during infancy and early childhood [7]. However, removable appliances used during early orthodontic treatment could be beneficial throughout treatment process, by modulating shape of arches and increasing patient's cooperation and systematicity [8]. Orthognathic surgery plays a pivotal role in managing the skeletal discrepancies in Apert syndrome. Maxillary advancement, often through Le Fort III osteotomy or distraction osteogenesis, is frequently required to correct midface retrusion, to advance the maxilla and improve facial aesthetics, airway function, and occlusion [9,10]. Intraoral management of crowding, crossbite and malposition of the teeth requires detailed diagnostics and often multi-annual treatment with fixed braces [11].

Due to the progressive nature of craniofacial growth in Apert syndrome, orthodontic treatment often extends over a long period, sometimes well into adulthood. Long-term retention is essential to prevent relapse. Fixed retainers or removable appliances may be employed depending on the severity of the case and the specific dental movements achieved during treatment. In presented case use of removable retainer might have been the reason why bruxism was developed. There are studies which indicate that disclussion caused by retainer may affect masticatory muscles and their response [12,13]. Nevertheless,

advanced malocclusion, complex treatment procedures and comprehensiveness of treatment often results in recurrence of some initial occlusal problems [14]. In presented case there was a relapse of right posterior crossbite. Moreover, continued follow-up is necessary as the craniofacial skeleton remains dynamic and can continue to change as the patient matures [15]. Beyond the physical challenges, patients with Apert syndrome also face significant psychosocial difficulties related to their appearance and speech. Orthodontic treatment can have a profound impact on a patient's quality of life by improving facial aesthetics and dental function, which in turn can enhance self-esteem and social interactions [16].

Apert syndrome often requires surgical intervention to address cranial, facial, and dental deformities. Early surgical management is crucial to prevent neurological complications, optimize appearance, and enhance function. Cranial vault remodeling is a cornerstone of surgical management, aimed at reducing the risk of increased intracranial pressure and improving neurodevelopmental outcomes. This procedure typically involves a frontal-orbital advancement to expand the skull, thus addressing craniosynostosis and preventing brain compression. Timing of surgery is critical, usually performed in infancy or early childhood, to facilitate optimal brain development. Distraction osteogenesis has also emerged as a valuable technique for facial reconstruction. This method involves the gradual lengthening of bones, particularly the maxilla, to correct midfacial hypoplasia and improve facial aesthetics and airway function. Distraction osteogenesis is often used in combination with other surgical approaches to address skeletal deformities and facilitate gradual, controlled expansion. This technique allows for better facial balance while minimizing the need for extensive bone grafting. Surgical management also involves correction of ocular anomalies and ear deformities. Timing and multidisciplinary coordination are essential for optimal outcomes in these patients [17,18,19,20].

The orthodontic treatment of patients with Apert syndrome is highly complex and requires a multidisciplinary approach that includes orthodontists, maxillofacial surgeons, and other healthcare providers. Timing of treatment, the use of advanced surgical techniques and long-term retention strategies are crucial for achieving successful outcomes. Despite the challenges, significant advancements in both orthodontic and surgical techniques have greatly improved the prognosis for these patients. Early diagnosis, careful planning, and ongoing collaboration between specialists remain key to optimizing both functional and aesthetic results in the orthodontic management of Apert syndrome.

In the presented case successful planning of surgical procedures and well-thought orthodontic treatment provided satisfying result for both the patient and the doctors. Despite numerous publications of successful management of Apert syndrome worldwide it is still challenging to achieve such a good result of treatment working only with the methods approved by governmental programme of orthodontic care for children with congenital defects of the facial skeleton of NFZ.

Authors' contribution

Study design – A. Ledwoń, N. Giża, L. Rodziewicz, U. Rojek
Manuscript preparation – A. Ledwoń, N. Giża, L. Rodziewicz, U. Rojek
Literature research – A. Ledwoń, N. Giża, L. Rodziewicz, U. Rojek
Final approval of the version to be published – A. Ledwoń, N. Giża, L. Rodziewicz, U. Rojek

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