CLINICAL CASE

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BEATA BIELICKA, AGNIESZKA NĘCKA, MARTA ANDRYCH

Interdisciplinary Treatment of Patients with Goldenhar Syndrome – Clinical Reports

Interdyscyplinarne leczenie pacjentów z zespołem Goldenhara – opis przypadków

Department of Facial Orthopedics and Orthodontics Silesian Piasts University of Medicine in Wrocław

Abstract

Goldenhar Syndrome known as dysostosis otoauricularis or hemifacial microsomia is a rare congenital disease. The clinical picture of this condition varies from a little asymmetry in the face to severe under-development of one half of the face with orbital deformation and microtia or sometimes total absence of the ear. Moreover, the chin and the facial midline are off-centered. Although hemifacial microsomia refers to one half of the face, the condition is bilateral in 31% of the cases, with one side being more affected then the other. Intra-orally patients with Goldenhar Syndrome reveal: malocclusions, tooth discrepancies, agenesis of third molars and second premolars on the affected side, supernumerary teeth, enamel and dentin malformations, delay in tooth development. The treatment of patients with hemifacial microsomia is very complicated and long-lasting, therefore it requires a cooperation of many specialists from medical and stomatological fields. Orthodontic therapy begins with removable orthodontic appliance, and when secondary dentition appears it is possible to continue the treatment using fixed orthodontic appliance to create a proper occlusal plane and to correct malocclusions and tooth discrepancies. Unfortunately, not only orthodontic treatment but surgical treatment is necessary to elongate ramus and corpus of lower jaw. There are two methods of surgical treatment: early operations (when the growth of patient is not yet ended) and late operations (when the growth is ended). In this paper there are presented the divergent opinions about selecting the operation method and moreover, 3 case reports of patients with Goldenhar syndrome are described (**Dent. Med. Probl. 2006, 43, 3, 458–462**).

Key words: Goldenhar syndrome, distraction osteogenesis, costo-chondral rib, orthodontic treatment.

Streszczenie

Zespół Goldenhara, zwany inaczej dysplazją oczno-uszną lub połowiczym niedorozwojem twarzy jest rzadką chorobą rozwojową. Obraz kliniczny tego zespołu jest zróżnicowany: od niewielkiej asymetrii rysów twarzy do znacznego niedorozwoju połowy twarzy z deformacją gałki ocznej i małżowiny usznej lub całkowitym brakiem ucha. U pacjentów tych stwierdza się ponadto przemieszczenie bródki i zaburzenie linii pośrodkowej. Mimo że zespół Goldenhara odnosi się do połowy twarzy, to w 31% przypadków choroba ta występuje obustronnie z przewagą jednej strony. Z innych objawów klinicznych w obrębie układu stomatognatycznego stwierdza się wewnątrzustnie: wady zgryzu, wady zębowe, brak zawiązków zębów trzonowych i przedtrzonowych po stronie zaburzenia, jak również obecność zębów nadliczbowych, hipoplazję szkliwa i opóźnione wyrzynanie zębów. Leczenie pacjentów z tym zespołem jest długotrwałe i wymaga współdziałania specjalistów z różnych dziedzin medycyny i stomatologii. Terapię ortodontyczną rozpoczyna się stosując aparaty ruchome, natomiast po wyrznięciu się stałych zębów można przejść do leczenia aparatami stałymi, dzięki czemu jest możliwe uzyskanie prawidłowej płaszczyzny zwarcia oraz korekty wad zgryzu i wad zębowych. W celu wydłużenia gałęzi i trzonu żuchwy jest jednak niezbędna interwencja chirurgiczna. Istnieją dwie metody chirurgicznej korekty wady: wczesne zabiegi (przy niezakończonym wzroście pacjenta) i późne zabiegi (po zakończonym wzroście). W pracy przedstawiono istniejące w piśmiennictwie rozbieżności poglądów co do wyboru metod operacyjnych stosowanych w leczeniu pacjentów z połowiczym niedorozwojem twarzy oraz przedstawiono 3 własne przypadki pacjentów z zespołem Goldenhara (**Dent. Med. Probl. 2006, 43, 3, 458–462**).

Słowa kluczowe: zespół Goldenhara, dystrakcja kostna, przeszczep kostno-chrzęstny, leczenie ortodontyczne.

Goldenhar syndrome known as otoauricular dysostosis or hemifacial microsomia (HM) is a rare condition, which is a part of first and second branchial arch syndrome [1]. This disease was first described in 1952 by Maurice Goldenhar, who found under-development of one half of the face, disturbances of the auricle and dermoid cyst of eyeball in one of the monozygotic twin [2, 3]. Etiology of this condition is not yet fully established, although it is known that it has genetic origin [4]. The latest genetic research on HM indicates 13 chromosomal aberrations and also 22 chromosomal pair trisomy [2]. The occurrence of Goldenhar syndrome ranged from 1 in 3000 to 1 in 5600 births. Males appear to be more frequently affected than females, and also the right side is affected more often than the left side (the proportion is 3 to 2) [5].

The clinical picture of HM varies from a little asymmetry in the face to a severe under-development of one half of the face with orbital deformations and microtia or even with total absence of auricle. The chin as well as facial midline are off-centered and usually deviate to the affected side. Moreover, patients with Goldenhar syndrome suffer very often for the auditory problems, which are induced by malformations in the middle, external and internal ear. 30 to 50% of this patients reveal conduction deafness, which is caused by dysfunction of facial nerve, in particular the temporal and zygomatic ramus of facial nerve. Intra-oral structures are also affected in this condition. The following symptoms can be observed: malocclusions, tooth discrepancies, macrostomy caused by underdeveloped lower jaw, agenesis of third molars and second premolars on the affected side, supernumerary teeth, enamel and dentin malformations and delay in tooth development. Moreover, patients with HM often reveal asymmetric development of masticatory system muscles as well as agenesis of salivary glands, and, rarely, palate clefts. The treatment of patients with hemifacial microsomia is very complicated and long-lasting, therefore it requires co-operation between many specialists from medical and stomatological fields, such as: laryngologist, ophthalmologist, speech therapist, mandibulo-facial surgeon and plastic surgeon. Orthodontic therapy begins with removable orthodontic appliance (functional appliance), and when secondary dentition appears it is possible to continue the treatment using fixed orthodontic appliance to restore a proper occlusal plane and to correct malocclusions and tooth discrepancies. Unfortunately, it is usually impossible to elongate alveolar bone using only orthodontic treatment, therefore the surgical intervention is needed in order to lengthen the mandibular ramus and mandibular corpus, which will reduce facial asymmetry. There are essentially two approaches: either an early operation (during growth), or a late treatment (after the active growth period). In the early approach, two methods are possible: the conventional surgical procedure or the distraction osteogenesis [6]. During the conventional operation, the deficient ramus or/and corpus of the mandible is partially replaced by an autologous osseo-chondral rib, which is usually taken from area of iliac crest or bone rib. The second possibility, i.e. distraction technique is a method of forming bone through corticotomy (separating the bone segments) and sequential stretching of the healing callus. This process is accomplished with the aid of a distraction device, which is secured with screws placed directly into the bone segments for a predetermined length of time.

The late surgical procedures consists of either a classical osteotomy (bimax surgery with canting the maxilla in combination with advancement of the mandible and lengthening of the ramus) or a bimaxillary distraction osteogenesis (Ortiz-Monasterio method) [7]. So, it is possible to use distraction technique either in young age of patient, when the growth is not yet ended or in later time, after the active growth period. Other surgical interventions important from esthetic point of view, such as correction of the auricle and soft tissue augmentation, are performed by plastic surgeons.

In this paper there are presented 3 cases of patients with Goldenhar syndrome treated in Department of Facial Orthopedics and Orthodontics, University of Medicine in Wrocław.

Case Reports

Patient 1

Patient K.N., now 13 years old, was referred by a paediatritian with a diagnosis of Goldenhar syndrome in 1992. From may 1992 she was also under periodical care in Plastic Surgery Hospital in Polanica Zdrój. On the basis of medical history of the patient, extra-oral and intra-oral clinical examination, analysis of diagnostic models and analysis of lateral skull roentgenogram, the congenital condition – hemifacial microsomia was confirmed. Clinical investigation revealed asymmetric lower half of the face, remarkable facial hypoplasia on the right side with the chin deviated to the affected side and concave facial profile as well as macrostomia (Fig. 1). Intra-oral examination showed severe asymmetric occlusion, II class occlusion, deep bite, tooth discrepancies, crowding and staphyloschisis – bifid tongue. Analysis of panoramic radiogram, one made at the age of 10 years and next at the age of 12 years, confirmed the loss

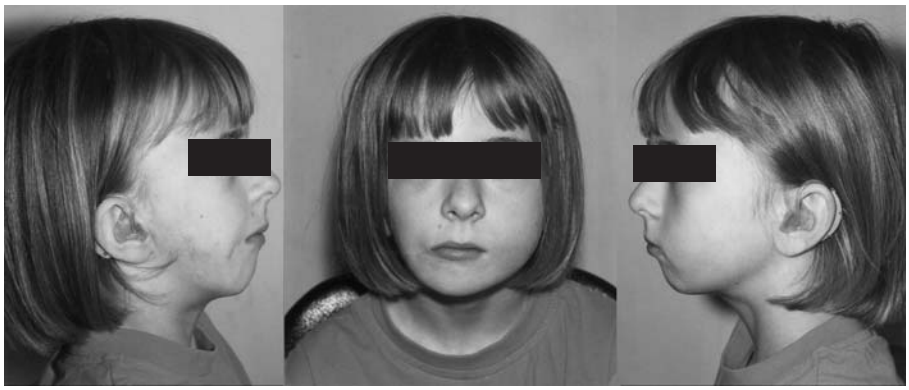

Fig. 1. Extra-oral view of patient K. N. – profile and en face

Ryc. 1. Pacjentka K. N. Zdjęcia zewnątrzustne z profilu i en face

of second lower molar tooth germ on the right side and moreover, showed severe degree of mandibular asymmetry (Fig. 2). Cephalometric analysis according to Hasund and Segneer' method revealed: retrognathic face, angle SNA = 66°, SNB = 61.4°, SNpg = 61.4°; increase of following angles ANB, ML-NL and mandibular angle, and also increase of WITS index (WITS = 10.5 mm). During ENT examination the bilateral hearing loss (hypoacusia) was diagnosed.

The patient underwent orthodontic treatment at the age of 2 years. The removable orthodontic appliance (Schwarz plate with a tongue crib) was made to correct tongue thrusting. Also, logopedic treatment was advised and then performed. After 4 years of orthodontic treatment in order to make osteotomy of mandible and later to close staphyloschisis the patient was send to Hospital of Plastic Surgery in Polanica Zdrój. Post-operatively, the Schwarz plate was substituted by bimaxillary functional orthodontic appliance to reach I class occlusion and to create acceptable overbite and overjet. At the age of 12 years, when the patient had secondary dentition, the fixed orthodontic appliance on the upper and lower jaw was made. Then, it was possible to treat crowding and other tooth discrepancies. At present, the patient is provided with a special program of orthodontic care over children with congenital anomalies of facial cranium. The further orthodontic treatment depends on the effect of elongating the mandible bone. The results of up to now treatment performed in the patient we can accept as being satisfying, because the occlusal condition as well as the function of stomatognathic system have been significantly improved. However, in order to improve esthetic appearance of the patient's face, the further plastic operations are needed.

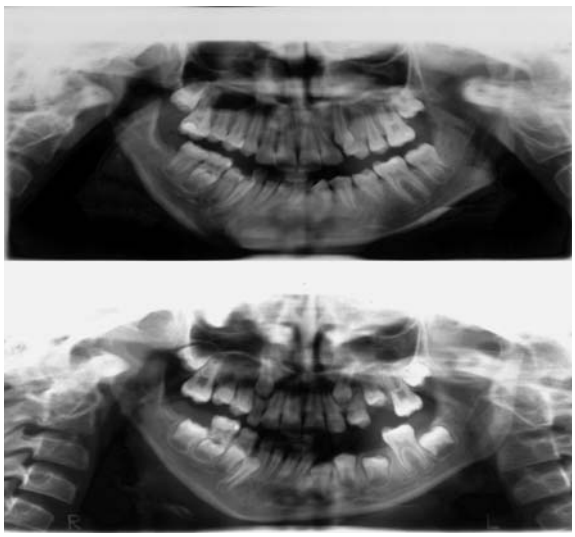

Fig. 2. Panthomografic X-ray of patient K. N made of the age of 10 and 12 years old

Ryc. 2. Pacjentka K. N. Zdjęcia pantomograficzne wykonane w wieku 10 i 12 lat

Patient 2

Patient D.M. now 10 years old was referred by an orthodontist to our department with suspected dysostosis mandibulofacialis. Clinical extra-oral examination revealed severe asymmetry of the lower half of the face, with the chin deviated to the right side, and also concave facial profile, bilateral atrophy of acoustic ducts and macrotia. Moreover, in the past, the patient underwent implantation of hearing aid type BAHA (Fig. 3). The patient revealed hypotonia of orbicularis oris muscle and hypertonia of geniohyoid muscle. Intra-oral examination showed: contraction of upper jaw, midline deviated to the affected side, protrusion of upper incisors and premature loss of primary dentition, especially molar teeth (Fig. 4). Cephalometric analysis according to Hasund and Segneer' method revealed: ortognathic face, angle ANB = 10.7°, SNB = 71.4°, SNpg = 71.2°, moreover increase of following angles ML-NL, Ml-NSL and also severe increase of WITS index (WITS = 7.7 mm).

The orthodontic treatment began from removable orthodontic appliance-Schwarz plate in order to expand maxilla and in this way to treat bilateral cross bite. Moreover, using this appliance, it was possible to eliminate protrusion of upper in-

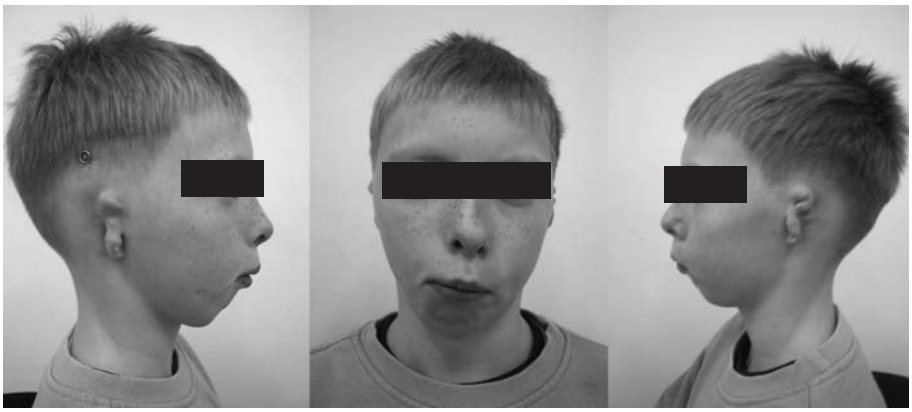

Fig. 3. Extra-oral view of patient D. M. – profile and en face

Ryc. 3. Pacjent D. M. Zdjęcie zewnątrzustne z profilu i en face

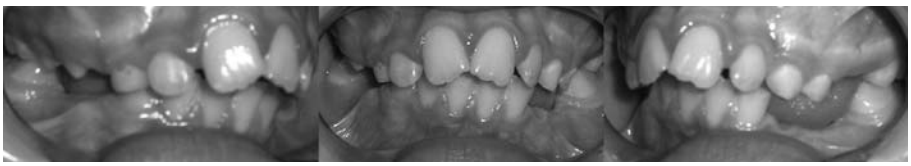

Fig. 4. Intra-oral view of patient D. M.

Ryc. 4. Pacjent D. M. Zdjęcia wewnątrzustne

cisors in significant degree. At the same time, the patient was referred to Hospital of Plastic Surgery in Polanica Zdrój. At present, the patient is qualified to surgical lengthening of mandibular ramus using distraction technique.

Patient 3

Patient M.S. now 7 years old, came with her parents to our department in order to treat malocclusions. Extra-oral examination revealed: asymmetric lower half of the face, severe facial hypolasia of the left side, with the chin deviated to the affected side and concave facial profile. Analysis of the panoramic radiogram showed shortened ramus of mandible on the left side (Fig. 5). Moreover, the patient revealed hypertonia of geniohyoid muscle. Clinical intra-oral intervention revealed: primary dentition with intensify caries, premature loss of upper right deciduous incisor and erupted permanent first molars and incisors (Fig. 6). At first step, the patient underwent assanation of oral cavity**,** and also removable orthodontic appliance was made. After that, the patient was send to Plastic Surgery Hospital in Polanica Zdrój, where she was qualified to surgical operation–distraction osteogenesis. Post-operatively, we are planning to continue orthodontic treatment using functional orthodontic appliance.

Discussion

Treatment of patients with Goldenhar syndrome usually starts in the early age of the patient and it is long-lasting therapy. This therapy is complex and requires cooperation of many specialists from medical and stomatological fields. It is possible to obtain good esthetical and functional effect of this congenital condition by applying interdisciplinary treatment. In the literature we can find different opinions on the proper time to begin the treatment of the syndrome. Some authors have preferred the early treatment intervention. For example, Munro et al. [8] have claimed that the early surgical procedure is the best option for the further proper growth and, moreover, for the psychological reasons. The other authors have preferred the surgical investigation to be delayed until the growth of the patient is ended, because then, the whole deformation is exposed and it is possible to plan the most suitable surgical procedures [5].

The other essential problem is selection of method of the surgical treatment. There are several methods of therapy: from one hand, the conventional surgical procedure such as costo-chondral rib graft or classical osteotomy, and from the other one, the distraction technique. All the presented methods have advantages and disadvantages. One of the contra-indication for using costo-chondral rib graft is the fact, that it has another rhythm of growth than the condyle and moreover, it is independent from the opposite healthy condyle growth [5, 9]. So, it is sometimes seen overgrowth at the grafted side. Moreover, when the costo-chondral rib is growing too much and too fast, this 3-dimentional overgrowth can also result in bulk of tissue that can diminish the range of mandibular movements. The good alternative for the classical intervention is distraction osteogenesis. Using this technique, it is possible to lengthen the jaw and the ramus of the mandible to the desired size. Unfortunately, this method is not free from disadvantages. First, the most important limitation is a situa-

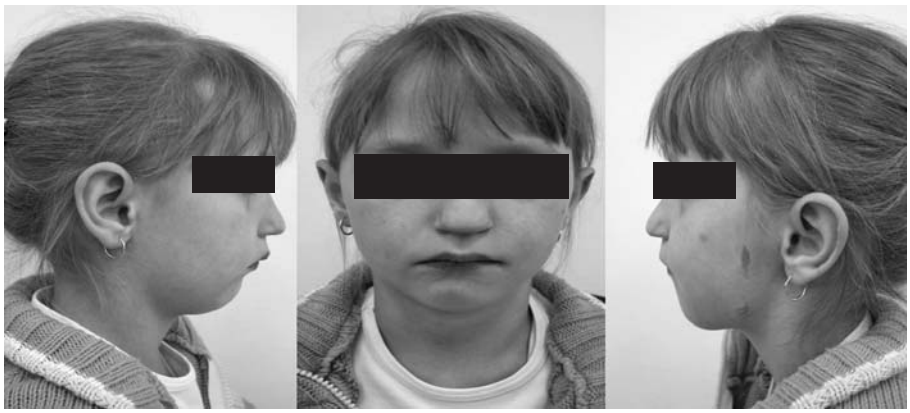

Fig. 5. Extra-oral view of patient M. S. – profile and en face

Ryc. 5. Pacjent M. S. Zdjęcia zewnątrzustne z profilu i en face

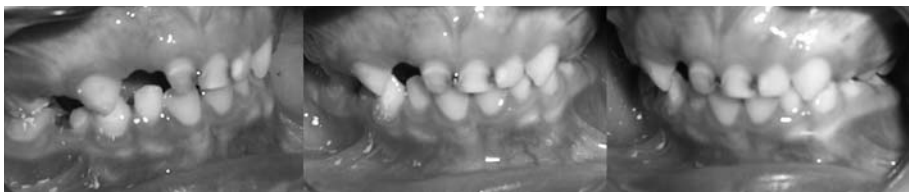

Fig. 6. Intra-oral view of patient M. S.

Ryc. 6. Pacjent M. S. Zdjęcia wewnątrzustne

tion when TMJ-reconstruction is needed. Distraction osteogenesis can lengthen the jaw, but can not create normal growing and functioning of temporo-mandibular joint. The second side-effect is a risk of mild infection during the active and passive period of lengthening.

Taking into account our experience in treating patients with Goldenhar syndrome, we can conclude that good results of treatment of such complicated congenital condition as hemifacial microsomia, are possible only by performing the complex and interdisciplinary medical care. Such therapy requires the cooperation of many specialists, and, what is not less important, the good communication and understanding between this team of specialists and a patient, as well as his or her family.

References

[1] Burdzińska-Gołowin J., Bryniarska-Łasisz D., Ritter A., Maszkiewicz W.: Zespoły zaburzeń rozwojowych łuków i kieszonek skrzelowych w świetle obserwacji kardiologicznych. Ped. Pol. 1981, 56, 1157–1164.
[2] Obrębowski A., Kraśny J., Wojnowski W., Kurywczak-Grykiel K.: Niedosłuch mieszany w zespole Goldenhara-Gorlina. Otolaryngol. Pol. 1999, 53, 639–641.
[3] Lisicka E.: Trzy przypadki zespołu Goldenhara. Klin. Oczna 1984, 86, 273–274.
[4] Chrzanowska K., Fryns J. P., Krajewska-Walasek M., Van Den Berghe H., Wiśniewski L.: Nager type of acrofacial dysostosis: An example of autosomal dominant transmission with variable expression. Genetica Polonica 1991, 32, 69–76.
[5] Moulin-Romsee C., Verdonck A., Schoenaers J., Carels C.: Treatment of hemifacial microsomia in a growing child: the importance of co-operation between the orthodontist and maxillofacial surgeon. J. Orthodont. 2004, 31, 190–200.
[6] Kobus K., Pisulska-Otremba A., Kobus-Zaleśna K.: Kortykotomia i dystrakcja kości twarzowo-czaszkowych. Pol. Przegl. Chir. 2002, 74, 933–940.
[7] Ortiz Monasterio F., Molina F., Andrade L.: Simultaneous mandibular and maxillary distraction in hemifacial microsomia in adults. Avoiding occlusal disasters. Plast. Reconstr. Surg. 1997, 100, 852.
[8] Munro I. R., Philips J. H., Griffin G.: Growth after construction of the temporomandibular joint in children with hemifacial microsomia. Cleft Palate J. 1989, 26, 303–311.
[9] Kahl-Nieke B., Fischbach R.: Effect of early orthopedic intervention on hemifacial microsomia patients: An approach to a cooperative evaluation of treatment results. Am. J. Orthod. Dentofacial Orthop. 1998, 114, 538–550.

Address for correspondence:

Beata Bielicka
Katedra i Zakład Ortopedii Szczękowej i Ortodoncji AM
ul. Krakowska 26
50-425 Wrocław
Poland
tel.: +48 71 784 02 80
e-mail: beti@kn.pl

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