

## Case Reports

# THE APPLICATION OF STIMULATOR IN THE TREATMENT OF CLEFT LIP AND PALATE IN GOLDENHAR SYNDROME, TRISOMY 13 AND LOBAR HOLOPROSENCEPHALY WITH A MEDIAN CLEFT LIP

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**Abstract.** *The clinical picture of a newborn with a syndromic cleft lip and palate is severe. The orthodontic and surgical treatment of the cleft is complex and long-term. It is further complicated by various birth defects which can be life-threatening for a newborn or can make the therapy itself more difficult. The induction of a newborn into total anesthesia with a view to performing the surgery of a cleft is often made difficult or time-limited. This paper presents pre-surgical orthodontic therapy in newborns with three severe types of cleft, UCLP, BCLP and premaxillary agenesis with median cleft lip which occurred within three rare syndromes - Goldenhar syndrome, lobar holoprosencephaly with a median cleft lip and trisomy 13 (47XX+13). Pre-surgical orthodontic therapy was conducted by means of RBJ stimulators without extra oral fixation, whose construction was conditioned by the type of cleft. With active treatment of RBJ stimulators, the cleft area in all three types of cleft was significantly reduced, as well as the protrusion of the premaxilla in BCLP. By directing the growth of cleft segments of newborn's upper jaw, the most approximate shape to a healthy newborn's jaw shape is achieved. All three types of described stimulators used in the therapy of syndromic cleft lip and palate enabled primarily the feeding of newborns, and thus their survival. With their orthopedic treatment they created optimal conditions for successful performing of surgical care of syndromic cleft lip and palate.*

**Key words:** *Prenatal development, craniofacial growth, infant orthopedics, feeding.*

## Introduction

The early orthodontic therapy by means of stimulators in different types of cleft lip and palate is a useful therapeutic procedure in the earliest period of life. Its two advantages are enabling the function of feeding in newborns and pre-surgical therapy. There are numerous assertions which confirm the efficiency of its application in nonsyndromic cleft lip and palate [1–4].

Clefts which occur within various types of syndromes further enlarge the severity and seriousness of the clinical picture of these syndromes. Over 400 syndromes within which clefts occur were described in the literature [5]. Thirty percent of all orofacial clefts fall into the category of syndromic clefts. They were described for the first time as early as 1940s when anatomic classification of clefts was attempted and it was noticed that in many individuals affected by clefts there were adjoined defects which, to a great extent, affect the survival of newborns or affect the therapy. Cleft lip and palate most commonly occur within following syndromes: Van der Woude syndrome (CLP and CP), Microdeletio n22q11.2 syndrome (CP), Stickler syndrome (CP), Treacher Collins syndrome (CP), Opiz syndrome

(CLP), Kabuki syndrome, Smith Lemli Opiz syndrome (CP), etc.

Many of these are quite rare, having been reported in a handful of families or individuals.

The surgery of cleft lip and palate in newborns is made difficult due to broad communication between nasal and oral cavity, the lack of available tissue for closing communication, protruded premaxilla, in BCLP, the lack of bony foundation in median cleft lip, dislocation of cleft segments, etc. Pre-surgical orthodontic therapy has a goal of directing a cleft segment of the upper jaw in the direction of closing the cleft and forming one regular shape of the upper jaw from the cleft segments.

The treatment of syndromic cleft lip and palate is further complicated by numerous birth defects; heart, nephrological, metabolic, etc. The induction of a newborn into total anesthesia for the surgical therapy of cleft lip and palate in severe cases is sometimes unmanageable or time-limited. For these reasons, a proper pre-surgical orthopedic preparation of newborns is of essential importance. It not only facilitates performing of surgery and achieving the best possible results, but it also reduces the time duration of anesthesia in newborns.

The paper describes the application of RBJ stimulators in three different entities: Goldenhar syndrome, lobar holoprosencephaly with a median cleft lip and trisomy 13 (47XX+13).

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## Patients and Methods

### Patients

Goldenhar syndrome/oculo-auriculo-vertebral spectrum, hemifacial microsomia, facio-auriculo-vertebral syndrome, and first and second branchial arch syndrome represent similar errors in morphogenesis with gradation of severity [6]. The syndrome is characterized by varying degrees of prevalently unilateral underdevelopment of the craniofacial structures (orbit, ear, and mandible) in association with vertebral, cardiac, renal, and central nervous system defects (Fig. 1). The frequency of occurrence is estimated to be 1 per 3000 to 1 per 7000 with a slight male predominance [6]. The syndrome is a sporadic condition. Cleft lip and palate has been reported in 16% of affected individuals, and 36% have mental retardation.

Trisomy 13 or syndroma Patau (47 XX +13) (Fig. 2) is the third most common autosomal trisomy occurring with an incidence of 1:5000 live births. The most characteristic malformations include varying degrees of holoprosencephaly, microphthalmia, scalp defects (Fig. 2D), cardiac defects and polydactyly (Fig. 2C). CL/P occurs in 60-80% of cases and is midline when associated with HPC. Newborns most commonly live up to 7 days. Growth deficiency and severe cognitive impairment are typical. Most cases of trisomy 13 are a result of nondisjunction often associated with advanced maternal age. Families are typically counseled that there is less than 1% recurrence risk. Types of clefts which are described within this syndrome are: midline cleft lip and palate, bilateral cleft lip and palate and unilateral cleft lip and palate [7].

Lobar holoprosencephaly with a median cleft lip is a complex malformation of the brain associated with the median facial defects (Fig. 3). Variability of the clinical picture is the characteristic of this anomaly. In most cases, the degree of severity of the facial anomaly correlates with degree of damage to the brain [8] Facial

anomalies are divided into five groups: cyclopia, ethmocephaly, cebocephaly, premaxillary agenesis with median cleft lip, and other less severe manifestations [9]. Premaxillary agenesis with median cleft lip is a complete median lip cleft, and when associated with hypotelorism, it is the fourth type of facial anomaly within HPC. A face with this anomaly is characterized by the absence of crista galli, the nasal bone, the complete premaxilla, and nasal septum. Its occurrence is 1:16 000 of live-born infants [9].

These three anomalies are followed by different types of clefts, which, according to severity belong to severe forms of clefts. A newborn with Goldenhar syndrome, according to the severity of the clinical picture has a milder form of this syndrome, but unilateral complete cleft of primary and secondary palatum influences that it obtains the shape of a middle-degree anomaly (Fig. 1C). There is a great dislocation of a bigger segment of palatal extension along with a nose septum to one side due to the existence of complete cleft of primary and secondary cleft. A nose wing and unformed nostril are also broadly long-drawn on the side of cleft. Lateralization, which is a characteristic of this syndrome, is reflected in this type of cleft.

The severe form of complete bilateral cleft of primary and secondary palate even further emphasizes the severity of the clinical picture of a newborn with trisomy 13. The premaxilla carried by the nose septum is clearly distinguished from the rest of the face. There is a big cleft at the level of palatal extensions which achieves complete communication between oral and nasal cavity with the protruded premaxilla (Fig. 2B).

The most severe type of cleft, which is at the same time very rare, is median cleft lip (which occurs together with congenital, complete lack of the premaxilla) (Fig. 3B). Due to the non-existence of frontonasal process which participates in the forming of the middle part of the face, a newborn does not have nasal bones, premaxilla, so that there is almost no boundary between the two cavities.



**Fig. 1** The face appearance of a newborn with Goldenhar syndrome. Note auricular tag, epibulbar dermoid non-typical UCLP.



**Fig. 2** The face appearance of a newborn with trisomy 13. In addition to BCLP, features that are apparent include the bulbous nose, up-sweep to the scalp hair pattern and postaxial polydactyly.



**Fig. 3** The face appearance of a newborn with Lobar holoprosencephaly with a median cleft lip. Note the lack of crista galli, the nasal bone, the complete premaxilla, and nasal septum [8].

## Methods

All three types of clefts are very difficult for surgical care. In unilateral cleft, the most difficult thing is to achieve the symmetry due to lateralization of structures that are involved in the formation of a nose and lips. It is difficult to

surgically care for bilateral cleft due to the protruded maxilla, since it often comes to dehiscence due to tissue tension, but in comparison with surgical repair of the palate, it is by far easier than the median cleft with premaxillary agenesis in which it is almost impossible to repair the palate due to the lack of bony tissue.

The role of RBJ stimulators has a great significance in all three types of clefts within pre-surgical therapy. The active effect of stimulators is mostly expressed by activating an orthodontic screw which is implemented in the construction of stimulators in all three types of clefts, as well as the specific cutting of an acrylic plate. Some of the basic principles which are common regardless of the type of cleft are: the screw is always placed at the right angle in relation to the line that joins two sides of acrylic plate, the big segment serves as the

strongpoint for the movement of a smaller segment, the acrylic plate covers palatal segments to the boundary between mobile and immobile mucous membranes, as well as the fact that the basic condition for efficient and the only action of stimulators is a perfectly taken impression between the upper jaw of a newborn. The difference between stimulators used in orthopedic and surgical therapy of these three clefts is in the line of plate cutting. In unilateral cleft of primary and secondary palate the line of acrylic plate cutting is in the shape



**Fig. 4** Taking impressions, impression, cast model and stimulator in Goldenhar syndrome (A, D, G, J), trisomy 13 (B, E, H, K) and lobar holoprosencephaly with a median cleft Lip (C, F, I, L).

of Z. In such a way, a smaller palatal segment moves in the medio-mesial direction after activating the screw, thus reducing the cleft area between palatal segments as well as the cleft at the level of alveolar edges and at the same time it achieves the closest possible regular shape of the upper jaw (Fig. 4J). Since the problem with BCLP lies in the protruded premaxilla, the cap with the premaxilla is most often pulled backwards between cleft palatal segments by means of the open screw (Fig. 4K). After that, different types of stimulators are possible. Median cleft lip with premaxillary agenesis only has transversally separated palatal segments, so that by means of activating the open screw in a straight line, we direct segments towards each other (Fig. 4L).

The basic requirement for efficient action of stimulators is a perfectly taken impression of a morphologically altered upper jaw in all three syndromes. On a proper impression, after the pouring of a cast model, a complete mucosa of a hard palate and alveolar edges should be outlined. Each crease, plica, lateral sulcus on a hard palate, frenulum on the alveolar edge can act as an excellent retainer, but also quite the opposite, as destabilizers of a stimulator (Fig. 4G–I). This is why it is necessary to release these anatomic details from the acrylic plate of stimulators, to bypass every plica and frenulum.

Taking impressions has its specificities Fig. 4A–C. The impression is always taken when a newborn is awake, it is even desirable that the infant is crying. This is important, on one hand because we can then see the face color of a newborn, and on the other hand, in such a way, anatomic details are better marked. It is especially important that it then comes to the lowering of the soft palate, by which the boundary between hard and soft palate is clearly outlined, and at the same time it represents the distal boundary of a future stimulator. Even a slightly elongated distal stimulator can cause an urge to vomit, which is completely undesirable. The impression consists of two phases. The first anatomic impression is always taken with alginates, and the other corrective impression with addition silicones. The anatomic impression gives only basic outlines. The addition impression (corrective) taken with addition silicones represents a key step towards the application of stimulators (Fig. 4D–F). It shows the most minute details, since it is taken with the help of hydrophilic and hydrophobic material that is in a liquid state. Taking impressions is based on doctor's expertise and good knowledge of the characteristics of impression masses.

The role of stimulators is twofold. Besides its role in pre-surgical therapy in all three syndromes, its invaluable importance is to enable the function of feeding immediately after the birth. Namely, due to the existence of broad communication between a nasal and oral cavity, there is no possibility of creating negative pressure in the mouths of newborns which is necessary for sucking milk. At the attempt of feeding it comes to choking, coughing, aspiration of milk, often cyanosis or developing pneumonia. In such situation parents succumb to panic and they are in constant fear of their newborn's survival. After the

application of a stimulator that closes communication between an oral and nasal cavity, it serves as an artificial palate. In such a way, conditions are made for a tongue to resist from the hard side of the acrylic plate and to perform its function that it has during breastfeeding. Apart from that, in this way, the tongue is disabled to stay inserted high towards a nasal cavity into the empty space between cleft palatal segments ever broadening it. The tongue position becomes physiological and has the power to participate in the regular development of occlusion and other orofacial functions. Orthopedic pre-surgical therapy by means of stimulators is based on biological foundations and the aspect of individuality. Numerous studies have shown that the growth of bony segments of the upper jaw affected by cleft is not disturbed [10–13]. Only the directions of the growth of bony segments are disturbed. The role of stimulators is in directing growth. The growth is led in the direction of closing the cleft area and achieving the shape of the upper jaw as regular as possible. In this way, an excellent pre-surgical preparation is performed that creates conditions not only for facilitated performing of surgery, but making the aesthetic and functional results of surgery as good as possible.

The aspect of individuality in pre-surgical orthodontic therapy by means of stimulators is also very important. Our observations are in compliance with many authors who confirmed that regardless of the classification of clefts, there are great individual differences within the same group. They relate not only to the differences in the morphology of the cleft-affected upper jaw, but to the type of growth as well. This is why this should be taken into account when constructing stimulators. A good orthodontist must recognize the type of growth at the very beginning of newborn's life, to 'listen to' it later and to design stimulators in accordance with that, changing them every month until the first surgical intervention.

Stimulators presented in this paper have one more characteristic that distinguishes them in relation to other appliances described in orthodontic therapy of newborns with clefts, and that is the lack of extra oral fixation which helps in the retention of appliances. Different means of retention are described. From caps on the heads of newborns, various elastic bands or even pins directly placed in palatal segments which prevented appliances from falling out [14]. The retention of stimulators is achieved with construction that was made after a perfectly taken impression of a cleft upper jaw. In such a way, with a strong strongpoint on the segments of the upper jaw, a stimulator gives excellent orthodontic results. The effects of its action were confirmed with a 3D computer analysis [4].

## Conclusion

The role of RBJ stimulators in pre-surgical orthodontic therapy is multiple. By enabling the function of feeding at the very beginning of life, it solves the essential problem of newborn's survival. With its active effect it

directs the growth of cleft segments, reduces the size of cleft and creates the conditions for successful achievement of the best possible aesthetic and functional results of surgical interventions to come. With proper pre-surgical therapy, they reduce the duration of surgical therapy, and thus the duration of total anesthesia which is often impossible in syndromes due to the existence of multiple birth defects. The therapy is comfortable since it does not damage the sensitive mucosa of newborns. The lack of extra oral fixation positively affects parents'

psychic state and their environment. Positive effects of therapy with stimulators are confirmed with a 3D computer analysis, and their use is therefore recommended in the therapy of syndromic clefts.

**Research ethics and patient consent.** This research involving human subjects is conducted according to the World Medical Association Declaration of Helsinki and with approval of the institution.

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