ANALYSIS OF THE INCIDENCE OF CONGENITAL MALFORMATIONS OF THE MAXILLOFACIAL REGION IN THE NIZHNY NOVGOROD REGION OF RUSSIA

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Abstract. A major problem of medicine is the prevention, treatment and rehabilitation of patients with congenital malformations of the maxillofacial region. This is due to their frequency, the severity of anatomical and functional disorders, as well as the complexity of social adaptation of patients. Cleft lip and palate are highly common malformations with a significant variation of risk factors. The updating of epidemiological data in certain regions of Russia makes it possible to identify the dominant causal mechanisms of the occurrence of defects, to determine the necessary directions for improving preventive and diagnostic measures, treatment and tactics of dispensary supervision of patients. The analysis of the incidence of congenital malformations of the maxillofacial region in the city of Nizhny Novgorod and the Nizhny Novgorod region of Russia over the past 19 years has been carried out. The risk factors for the development of anomalies, the structure of morbidity, the type and nature of the therapeutic measures carried out were determined.

Keywords: cleft palate, cleft lip, congenital malformations, maxillofacial anomalies, orofacial disorders, craniofacial defects, maxillofacial malformations, epidemiology, interdisciplinary approach.

List of Abbreviations

CM – congenital malformations
LS-ChP – left-sided cheiloplasty
LS-RChP – left-sided rhino-cheilo-plasty
LS-RChPP – left-sided rhino-cheilo-palate-plasty
MSS – musculoskeletal system
NNR – Nizhny Novgorod region
RS-ChP – right-sided cheiloplasty
RS-RChP – right-sided rhino-cheilo-plasty
RS-RChPP – right-sided rhino-cheilo-palate-plasty
WHO – World Health Organization

Introduction

The prevention, treatment and rehabilitation of patients with congenital malformations is an actual problem of medicine and maxillofacial surgery (Howe *et al.*, 2020; Hamzan & Sulaiman, 2020). This is related to their frequency, severity of anatomical and functional disorders, and the difficulty of social adaptation of patients (Corsello & Giuffrè, 2012; Taib *et al.*, 2015; Worley *et al.*, 2018; Ferrari-Piloni *et al.*, 2021).

Congenital deformities of the craniofacial region are ranked second among all human malformations, of which congenital clefts of the lip and/or palate account for 88% (Demikova *et al.*, 2015; Worley *et al.*, 2018).

The frequency of human CM cases is an important characteristic of a population's health condition (Fuangtharnthip *et al.*, 2021; Zhu *et al.*, 2021). According to WHO statistics, the birth rate of children with cleft lip and palate averages 1:750 newborns (in Russia this figure varies from 1:1000 to 1:600 in different regions), which is 20-30% of all human malformations and 86% of malformations of the maxillofacial region (Kasimovskaya & Shatova, 2020; Solodukhina & Shlyaptsev, 2021).

In recent decades, there has been no tendency towards a decrease in morbidity. This situation is largely attributed to the effect of toxic substances on the body as a consequence of the intensive development of industry, in particular chemical industry (Kiryushin *et al.*, 2023). The influence of teratogenic factors (physical, chemical, including pharmaceuticals) and biological agents (viruses) results in embryonic de-

velopmental disorders involving morphological anomalies and malformations, which are caused not only by external disharmony, but also by severe functional disorders (Azimov *et al.*, 2015). In addition, the period of embryonic development from the third to the eighth week is the most critical due to the fact that in this period embryonic cells are highly differentiated and susceptible to many teratogenic environmental factors (Kiryushin *et al.*, 2023).

The severity of anatomical and functional maxillofacial disorders explains the complexity and duration of treatment, requiring a comprehensive interdisciplinary approach to patient rehabilitation, including the engagement of various specialists (pediatrician, orthodontist, cleft surgeons, otolaryngologist, orthopedic dentist, speech therapist, psychologist, osteopath). Persisting anatomical deformities cause dysfunction of the directly affected and organs connected organs. The risk of developing middle ear disease, velopharyngeal dysfunction and malocclusion is often increased.

In this regard, the use of epidemiologic methods becomes extremely relevant not only for decision-making in clinical and preventive medicine, but also for the formation of a specific strategy aimed at improving the treatment and diagnostic process of congenital maxillofacial anomalies in the region.

Objective: to study the epidemiologic features and structure of the incidence of congenital maxillofacial malformations in Nizhny Novgorod and Nizhny Novgorod region of Russia from January 2005 to December 2023.

Materials and Methods

The medical records for 19 years: 2005-2023 (form 043-1/u – 251 medical records) of patients with malformations of the maxillofacial region – cleft lip and palate, admitted for orthodontic treatment were analyzed on the basis of Regional Children's Clinical Hospital of Nizhny Novgorod (Nizhny Novgorod, Russia). Clinical examination of 251 patients was conducted, who were divided into two groups: those under dispensary supervision (n = 135) and those under treatment (n = 116). The nosological form of the disease, localization of pathology (right- and left-sided, uni- and bilateral cleft lip and palate), the type of surgical intervention performed, and the patient's sex have been analyzed. Parents who gave birth to children with congenital pathology of the maxillofacial region were surveyed using a specially composed questionnaire consisting of 24 questions concerning the features of the parents' health status, living conditions, mother's lifestyle before and during pregnancy, and the features of the course of pregnancy. Anamnestic data were analyzed, including the factors identified by the parents of the patients as the presumed cause of pathology development. During the study, two groups were formed: the first included children under dispensary supervision, and the second group consisted of children under treatment. The study was conducted in accordance with the Helsinki Declaration (adopted in June 1964, revised in October 2013) and approved by the Local Ethics Committee. Voluntary informed consent was obtained from each patient for medical examinations, interventions and participation in the study. The information obtained in the course of the conducted research was systematized and compiled into a common database. After that, it was processed using Microsoft Office ® 365 software packages (Microsoft Corporation, Seattle, USA), Microsoft Excel and the Wolfram Mathematica (version 11.3.0.0) statistical package. For the qualitative variables the absolute frequency of congenital anomalies of the maxillofacial region and the frequency of manifestation as a percentage were calculated. The statistical significance of the difference between qualitative features in the samples was assessed using the Chi-square test.

Results

According to the data of medical records analysis, the number of patients for 19 years amounted to 251 people (Table 1). The morbidity rate has been defined as stable over 19 years in the territory of Nizhny Novgorod and the region of city. It was noted that the number of boys with congenital maxillofacial malformations prevails over the number of girls in the ratio of 1.5:1.

Table 1

Distribution of patients with congenital malformations of the maxillofacial region by sex and nosological forms

№	CM of the maxillofacial region (nosology)	Boys, case/%	Girls, case/%
1	Unilateral complete cleft of the upper lip and palate with cartilaginous nasal deformity	75/36%	34/16%
2	Bilateral complete cleft of the upper lip and palate with cartilaginous nasal deformity	22/11%	10/5%
3	Cleft palate	15/7%	28/14%
4	Unilateral complete cleft of the upper lip with cartilaginous nasal deformity	10/5%	5/2%
5	Bilateral complete cleft of the upper lip with cartilaginous nasal deformity	2/1%	–
6	Cleft lip	2/1%	5/2%

The null hypothesis (H_0) was formulated as «the probability of the manifestation of congenital anomalies of the child does not depend on the sex of the child». At the same time, the Chi-square test (calculated) was 20.410, with Chi-square test (critical, for the number of degrees of freedom 5 and the significance level 0.01) – 15.086. Thus, H_0 was rejected. Therefore, it can be concluded that the probability of the manifestation of congenital anomalies of the child depends on the sex of the child. This provision dictates the need to study the hereditary factor using special laboratory genetic methods in large samples, over a longer period of time and in various regions not only of the one country, but also of the world.

The number of children with CM of the maxillofacial region under treatment in Nizhny Novgorod region is 1.9 times higher than in Nizhny Novgorod city (Nizhny Novgorod city – 39 people, NNR – 77 people).

The number of girls under dispensary supervision in NNR is 1.7 times higher than in Nizhny Novgorod city (42 and 25 cases, respectively). The number of boys with congenital maxillofacial malformations in NNR is 1.5 times less than in Nizhny Novgorod city (27 and 42 cases, respectively).

It was noted that the maximum incidence of morbidity among boys with congenital maxillofacial malformation was in 2015, among girls – 2015-2016. During the observation period (19 years), the number of referrals to Regional Children's Clinical Hospital of Nizhny Novgorod increased from 0.8% to 3.8% of cases.

The study assessed the distribution of children with congenital maxillofacial malformations under treatment and dispensary supervision at the Regional Children's Clinical Hospital of Nizhny Novgorod according to the type and localization of pathology (right-sided, left-sided).

It was found that a large proportion of children on dispensary supervision is represented by cases of rhino-cheilo-palate-plasty performed to treat congenital left-sided complete cleft lip and palate with cartilaginous nasal deformity (Fig. 1). In contrast, the majority of children under treatment have congenital right-sided complete cleft lip and palate with cartilaginous nasal deformity (Fig. 2, Fig. 3).

According to the results of a questionnaire survey of mothers/parents who gave birth to children with congenital maxillofacial malformation, it was noted that during pregnancy, mothers mostly experienced toxemia in the first trimester, as well as a risk of pregnancy termination. In 100% of cases, the questionnaires indicated that the children were born at term and the social status was favorable. All interviewed parents were in agreement with doctors' recommendations on child treatment.

The age of the parents at the time of birth of children with CM of the maxillofacial region ranged from 25 years to 40 or more, with an average maternal age of 27.8 ± 2.2 years.

In the family by order of birth, the newborn with CM of the maxillofacial region was the first in 80% of cases. The second and third child was born in the families in 13.3% and 6.7% of cases, respectively.

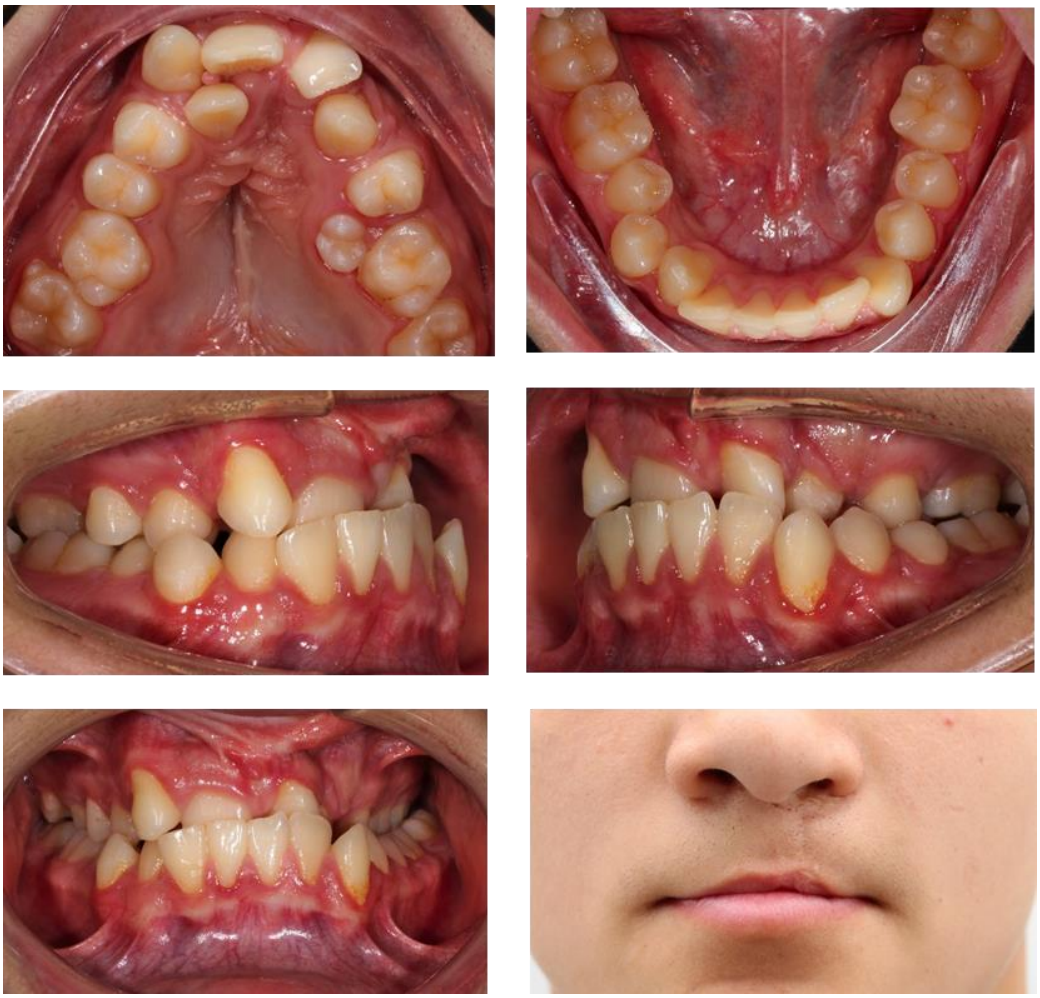

Fig. 1. Clinical cases of rhino-cheilo-palate-plasty performed to treat congenital left-sided complete cleft lip and palate with cartilaginous nasal deformity

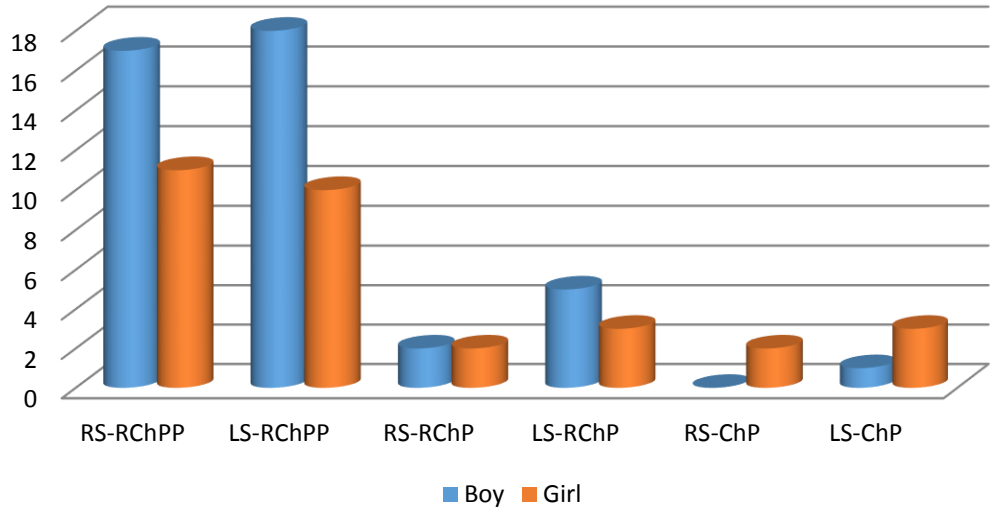

Fig. 2. Distribution of children with congenital maxillofacial malformation under dispensary supervision at the Regional Children's Clinical Hospital of Nizhny Novgorod by type and localization of pathology

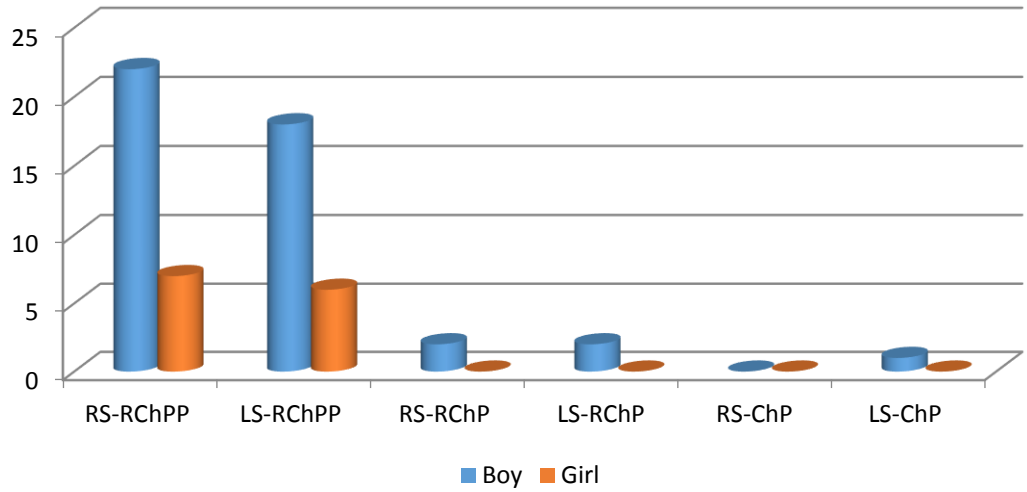

Fig. 3. Distribution of children with congenital maxillofacial malformations under treatment at the Regional Children's Clinical Hospital of Nizhny Novgorod by type and localization of pathology

It was also noted that during pregnancy expectant mothers were healthy in 73.3% of cases. However, in 26.7% of cases, expectant mothers had chronic diseases (gastritis, cholecystitis, duodenitis, pyelonephritis).

On the basis of the questionnaire the risk factors influencing the formation of congenital pathology of the maxillofacial region were assessed.

Among the presumed factors that provoked the development of pathology, parents mentioned: infectious diseases of the mother in early stages of pregnancy – 21.5%, environmental factor – 14.8%, threat of pregnancy termination – 13.3%, harmful chemical agent – 13.1%, chronic diseases of mother (gastritis, cholecystitis, duodenitis, pyelonephritis) – 12.4%, combined impact – 9.1%, congenital anomalies in family history, hereditary factor – 9.1%, occupational hazards – 6.7%, It is worth noting that in most cases the combined effect of these endogenous and exogenous risk factors was revealed.

Discussion

According to WHO data, the incidence of malformations of the maxillofacial region averages 3-6%; very severe combinations of developmental anomalies, resulting in disability or death of infants, are common. The etiology of CM is different, the main factors are environmental, social, hereditary, etc. (Korolenkova *et al.*, 2018). Data from local and foreign scientists clearly indicate the relationship between atmospheric air pollution and the formation of CM (Kiryushin *et al.*, 2023). Ubiquitously deteriorating environmental situation leads to chronization of the process and increased disability among the child population. And, unfortunately, there is an increasing tendency in the number of children born with cleft lip and palate (Preidl *et al.*, 2019; Salari, *et al.*, 2021).

Congenital clefts of the upper lip and palate occupy the 4th-7th place among all congenital anomalies in terms of the combination and complexity of the anatomical and functional disorders (Cobourne, 2004). The severity of facial malformation is determined not only by external disfigurement, pronounced functional disorders, conflict tension and negative psychological background in the family, but also by the fact that the deformity causes somatic disorders leading to growth retardation and underdevelopment of the child's body as a whole (Marginean *et al.*, 2018 Martín-del-campo *et al.*, 2019). Persisting anatomical disorders cause dysfunction of the organs directly affected and connected with them. The harmony of development of several areas is disturbed, and combined secondary deformities occur

(François-Fiquet *et al.*, 2014; Ozdemir & Esenlik, 2018; Ushnitsky *et al.*, 2019; Verma *et al.*, 2021).

The features of children's neuropsychic state differ sharply from the norm. Unfavorable conditions of upbringing and traumatization of children's psyche in early school age play an essential role in the emergence of psychogenic disorders (Worley *et al.*, 2018).

By affecting speech, hearing, appearance and psychology, the abnormalities lead to long-term adverse health and social adaptation of patients. Typically, children with these disorders need multidisciplinary medical care from birth to the end of rehabilitation. Surgeon, pediatrician, orthopedist, otolaryngologist, psychoneurologist are involved in identifying somatic abnormalities and associated malformations, carrying out the necessary correction of abnormalities in the preoperative period.

Using modern methods of diagnosis and treatment, complex rehabilitation of patients should include timely predictable results, treatment at different stages of rehabilitation of patients with this pathology, in order to eliminate the anatomical defect, restore facial contours, form intelligible speech and adaptation of patients in society (Demikova *et al.*, 2015).

The choice of the method of restoration of maxillofacial defects depends on the type and state of cleft, as well as on the equipment of the medical institution and the capabilities of the multidisciplinary medical team. Despite the fact that access to medical care has increased in recent years, especially in developing countries, the quality of care still varies considerably (Ignatieva, 2013).

The algorithm of rehabilitation measures largely depends on the severity of the malformation. It determines the sequence of diagnostic and therapeutic interventions, including rhino-cheilo-palate-plasty depending on the defect according to the indications, the choice of removable orthodontic construction as a preparation for treatment with bracket systems, and the scope of reconstructive intervention on the alveolar process with the subsequent choice of orthopedic treatment at the final stage of rehabilitation. The psychological readiness of the parents for each stage of treatment is essential for the beginning of each stage of treatment. It has been observed that early orthodontic correction is more effective.

The problem of diagnosis and treatment of this group of patients requires the development and implementation of a protocol for their complex rehabilitation depending on age and the degree of severity of morphologic signs of maxillofacial anomaly during the period of correction of developmental abnormalities.

Conclusion

For a long time, congenital malformations occupy one of the first places among the most urgent medical and social problems, which is caused by the high incidence of this pathology among newborns, the lack of a downward tendency in morbidity and the difficulty of eliminating their consequences.

Thanks to modern medical achievements, significant progress has been made in the treatment and rehabilitation of patients with congenital clefts, the aim of which is to eliminate the anatomical defect, create harmonious facial contours, intelligible speech and social adaptation. Modern hardware methods of orthodontic treatment in combination with soft tissue plasty at the early stages of treatment are designed to form favorable conditions for the subsequent restoration of bone structures in the anatomical areas of defects of the alveolar process on the upper jaw with subsequent orthopedic rehabilitation to restore the dentition.

The study of epidemiological features, incidence and structure of morbidity in each specific region should help to optimize diagnostics, treatment and rehabilitation measures, and to form a vector of prevention of congenital maxillofacial malformation. The presented data indicate the need for further prospective cohort studies aimed at specifying the degree of exposure of etiological factors, creating new preventive programs, improving diagnostics, treatment, and dispensary supervision that meet the current trends in medical development and contribute to improving the quality of life of patients.

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