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CLINICAL-GENETIC ASPECTS OF CONGENITAL CLEFT LIP AND PALATE AMONG CHILDREN OF YAKUTIA

ABSTRACT

The comparative analysis of prevalence and structure of congenital cleft lip and palate (CCLP) among children living in Yakutia for the last decade has been carried out. At the same time the assessment of the direct and indirect interrelation has been carried out according to medical-genetic consultation of patients and their parents. Studying of congenital anomalies frequency of maxillofacial area at children characterizes an unfortunate trend of their increase during observation.

The obtained data demonstrate that unilateral clefts of the upper lip and palate and also the isolated clefts of the hard and soft palates prevail in their structure. The uranostaphyloschisis was revealed more often in CCLP structure, further there were unilateral through cleft lip and palate, bilateral through clefts, hidden cleft palate, isolated cleft lip and alveolar process where children with syndromes of hemifacial microsomia, Pierre Robin and Gorlin-Goltz are the hardest in holding complex treatment-and-prophylactic actions and medical-social rehabilitation. Some examined children had severe forms of congenital malformation, due to syndromes of hemifacial microsomia and Pierre-Robin's syndrom. Meanwhile some children were revealed to have the inherited forms of malformation of maxillofacial area and also Van der Woude syndrome.

The analysis of pathological sign or syndrome among direct and indirect relatives characterizes the existence of some features where the main congenital malformation of maxillofacial area is traced from indirect relatives, and the direct genetic interrelation from mother or father. Our analysis has revealed the existence of features by gender. So, palate clefts were often found at girls in comparison with boys. At the same time boys prevailed on the contrary in indexes of unilateral clefts lip and palate. The dominance of left-side localizations of congenital clefts of the upper lip and palate was revealed by localization among these pathologies.

The analysis of the obtained data characterizes that the first stage of perfecting of complex medical-social rehabilitation is the creation of the uniform regional database of children with congenital clefts of the upper lip and palate.

Keywords: congenital clefts of the upper lip and palate, relevance, heredity, Pierre-Robin's syndromes, Van der Woude syndrome, hemifacial microsomia.

INTRODUCTION

The congenital cleft of the upper lip and palate is a heavy malformation of maxillofacial area, shown gross anatomic and functional disorders [3, 6]. It should be noted that congenital malformation of the face is not only medical, but also social problem; therefore a complex treatment approach allows us to achieve the good remote results [1, 2]. Congenital lip and palate clefts have multifactorial origin with negative impact of teratogens, heredity, etc. [7]. Despite constant perfecting of surgical and orthodontic methods of treatment of children with congenital clefts of the upper lip and palate, complex rehabilitation of such patients continues to remain one of the most difficult tasks for a maxillofacial surgeon, a dentist, a pediatrician, a logopedist and a psychologist [4, 5].

It is known that perfecting of health care system is based on knowledge of clinical-epidemiological and etiological features of case incidence. In this regard the researches directed to studying of these problems are relevant. We don't have similar researches in the conditions of Yakutia for the last period.

Materials and research methods

The retrospective and prospective analysis of registration cards and case histories has been carried out. Collecting of clinical material and medical-genetic analysis has been carried out on the basis of children's maxillofacial surgery at otorhinolaryngological department and medical-genetic department at Republic hospital № 1 – National center of medicine». Totally 281 children aged from 3 months up to 14 years and teenagers up to 18 years were examined during 2000-2016 years. There were 136 boys and 145 girls. All children were operated concerning congenital clefts of the upper lip and palate. They took a course of medical-social rehabilitation. The clinical-genealogic research of the families having the child with congenital pathology of maxillofacial area was done for the establishment of family relations, tracing the pathological symptom or syndrome among close and distant, direct and indirect relatives. At the same time we made a family tree and carried out the genealogical analysis. A consulting person and a proband (consulting – the person coming for doctor's consultation; a proband – the patient himself). Collecting information included the common questions: surname, name, proband middle name, date of birth, nationality of a proband, his mother and father, birthplace of a proband and parents, existence of kinship marriage in a family tree.

Statistical processing of clinical material was carried out with the use of standard methods of variation statistics.

Results and discussion

The carried-out dynamic analysis of birth rate of children with CCLP during observation characterizes the existence of some features in indexes. So, since 2000 to 2016 in the region there were born 281 children with congenital clefts lip and/or palate. There were 16.53±0.30 cases on average in a year. In 2014 there were maximum quantity (31 cases) of the birth of children with CCLP (CCLP frequency was 1:548 [1.82±0.02 – on 1000 newborns]), and the minimum index (3 cases) was revealed in 2001 (CCLP frequency was 1:4420 [0.22±0.05 – on 1000 newborns]). At the same time a dynamic increase in the line of a trend during observation was defined that characterized a negative tendency of frequency increase of congenital malformation.

It should be noted that CCLP structure has revealed clefts of hard and soft palates (37.82±0.66%), further there were unilateral through clefts lip and palate (30.33±0.74%). Meanwhile, the data of bilateral through clefts and also hidden clefts of the palate, the isolated clefts lip and alveolar process made 8.23±0.98, 7.49±0.99 and 7.52±0.99% respectively. At the same time, minimum indicators of frequency were the isolated clefts of the upper lip (4.13±1.01%), transversal clefts lip – 0.37±1.07%. As a rule, medical-social rehabilitation children with syndromes of hemifacial microsomia, Pierre Robin and Gorlin-Goltz are the hardest in holding complex treatment-and-prophylactic

actions. The carried-out assessment and analysis revealed these syndromes at 4.11±1.03% of children.

It is necessary to emphasize that the carried-out analysis revealed the existence of features by gender. So, girls have palate clefts more often (64.35±0.71%) whereas boys have 35.65±1.26%. At the same time boys on the contrary prevailed in indexes of unilateral clefts lip and palate (55.56±1.01%). The dominance of left-side clefts localizations was revealed by localization among these pathologies where the indicator was 61.72±0.87%.

At the result of analysis we revealed 31 children with the tainted heredity. 9 children were with a through cleft of the upper lip and palate: 4 - with left-side and 5 – right-side cleft. The quantity of cases with a palate cleft was 13 children, 4 of them with the latent form of a palate cleft. The medical-genetic analysis revealed a presence of bilateral clefts of the upper lip and palate at 5 children, including one child with Van der Woude syndrome. 4 examined children were revealed the hereditary tainted of the isolated clefts lip and/or alveolar process.

The analysis of tracing of pathological symptom or a syndrome among direct and indirect relatives characterizes the existence of some features. So, 35.48±2.81% have the direct genetic interrelation from mother or the father, and 64.52±1.55% - congenital malformation of maxillofacial area from indirect relatives.

Conclusion

The received results have demonstrated a negative tendency of frequency occurrence increase of congenital clefts lip and/or palate among children of the region for 2000-2016. At the same time some of the examined patients showed the genetic interrelation of congenital pathologies. Such situation needs the development and deployment of the complex regional program directed to the perfecting of treatment-and-prophylactic help and medical-social rehabilitation of children with congenital malformation of maxillofacial area with the creation of the unified electronic database.

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