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CLEFT LIP: CAUSES, SYMPTOMS, DIAGNOSIS, AND TREATMENT METHODS

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Abstract: In this article information is presented on congenital clefts of the upper lip in children.

Definition

Cleft lip, or cheiloschisis, is a congenital malformation of the craniofacial region characterized by the failure of the upper lip to fuse properly. Approximately 0.04% of children are born with this defect, with boys being affected more frequently.

Causes of Cleft Lip

The formation of a cleft lip is primarily linked to genetic abnormalities, specifically mutations in the **TBX22** gene. Factors that may trigger such mutations include:

- Use of antibacterial medications during pregnancy;
- Severe stress and emotional disturbances;
- Pregnancy toxicosis;
- Exposure to infections or radiation;
- Use of drugs;
- Smoking and excessive alcohol consumption.

These factors are particularly dangerous during the first two months of pregnancy. Additionally, statistical data indicate that the risk of having a child with a cleft lip increases in women over 40 years of age.

Cleft lip can also be inherited, so parents with this condition are advised to undergo **medical-genetic counseling** before planning a pregnancy.

Classification of the Condition

The cleft is usually located on the upper lip, either to the left or right of the midline. Rare cases involve defects on both sides or affect the lower lip.

Two forms of cleft lip are distinguished:

- **Complete**, when a deep fissure extends from the lip to the nose on one or both sides. This occurs due to the failure of the nasal process to fuse with the right or left maxillary process.
- **Incomplete (partial)**, when there is a partial cleft appearing as an indentation, usually on one side.

Extent and Severity of the Defect

The extent and depth of a cleft lip can vary. In mild cases, the cleft is limited to soft tissues. In severe cases, the non-fusion affects the bones of the palate and upper jaw, indicating the simultaneous development of a cleft lip and cleft palate. The condition is often accompanied by nasal deformities and clefts of the soft and/or hard palate.

Symptoms of Cleft Lip

Cheiloschisis is not only a cosmetic defect but also a cause of significant respiratory and speech disorders. Its main manifestations include:

- Impairments in sucking, swallowing, and chewing;
- Malocclusion and dental abnormalities;
- Speech disorders;
- Other craniofacial developmental anomalies.

Sucking and swallowing difficulties are most pronounced in deep, through-and-through defects that create a direct connection between the oral and nasal cavities. The lack of airtight separation between these cavities prevents the oral pressure necessary for the sucking reflex. If the defect involves the muscles of the soft palate, swallowing is also impaired. In such cases, newborns with a cleft lip are fed via a feeding tube.

In unilateral, shallow clefts of the upper lip, sucking and swallowing reflexes are generally preserved.

Chewing problems occur in two main situations: absence of timely cleft lip repair and improperly developed bite and dental deformities. Weakness of the muscles and ligaments of the pharynx and palate, seen in bilateral clefts of the upper lip with cleft palate, also contributes to difficulties in chewing.

Dental abnormalities occur due to the splitting of the upper jaw, leading to abnormal tooth growth: missing teeth, misaligned angles of eruption, or double teeth. Patients with a cleft lip are more prone to dental caries and orthodontic problems.

Speech disorders often manifest as **rhinolalia** (nasal speech), where speech acquires a pronounced nasal quality and sounds become unclear.

Cleft lip can also be associated with developmental anomalies of the nose and face, as well as congenital malformations of internal organs.

Cleft lip may occur as part of **Patau syndrome**, a condition characterized by the presence of an extra 13th chromosome. This syndrome involves multiple internal organ malformations, such as defects of the atrial septum and blood vessels. Children with Patau syndrome often present with multiple external anomalies, including narrow palpebral fissures, malformed ears, cleft lip, and cleft palate.

Diagnosis of Cleft Lip

A cleft lip can be suspected in the fetus during **ultrasound examination at 14–16 weeks** of pregnancy. In newborns, the defect is immediately visible.

Which Specialists to Consult

Cleft lip is treated by **craniofacial (maxillofacial) surgeons**. In some cases, consultation with a **speech therapist** may also be necessary.

Treatment of Cleft Lip

Thanks to advances in craniofacial surgery, cleft lip can now be successfully treated. Severe cosmetic defects that interfere with chewing and speech are, after surgery, usually reduced to a barely noticeable scar above the upper lip.

Cheiloplasty (Surgical Repair of Cleft Lip)

Cheiloplasty is usually performed in children between **three and six months of age**, provided there are no contraindications. In severe forms of the genetic defect, surgery may be performed in the **first days of life** if the infant is gaining weight steadily and has no cardiovascular, gastrointestinal, nervous, or endocrine disorders, and no anemia. However, due to the small anatomical size of a newborn's lips and the immaturity of some physiological functions, there is a high risk of significant blood loss. Therefore, if there are no urgent indications, surgeons typically wait until the child is **three to six months old**.

Surgical correction of the cleft restores the **anatomical structure and integrity of the lips**, corrects defects of the nose and palate, and addresses other facial abnormalities.

Cheiloplasty also provides the foundation for the **proper development of the craniofacial system** in the future. It is important that the correction of a cleft lip be completed by **three years of age**, when speech development is actively progressing.

Thanks to advances in modern medicine, **90% of children born with cleft lip and cleft palate** can lead normal lives without being self-conscious about their appearance. However, in **70% of cases**, additional surgery may be required later in childhood to correct residual deformities of the nose and lips.

Complications

Due to impaired sucking, infants with a cleft lip may not feed normally. Sometimes, swallowing and chewing disorders also occur, leading to **delayed growth and development**.

Children with cleft lip often develop **ENT problems and bronchitis**.

Abnormal oral and pharyngeal anatomy can impair the growth of facial bones, resulting in **micrognathia** (underdevelopment of the upper jaw) and **macrognathia** (overdevelopment of the lower jaw). The midface may appear sunken, and the eyes may appear prominent.

Prevention of Cleft Lip

To minimize the risk of having a child with a cleft lip and palate, prospective parents should:

- Avoid harmful habits (smoking, alcohol, drugs);
- Screen for **TORCH infections** (toxoplasmosis, rubella, cytomegalovirus, herpes);
- Consult a **geneticist** if a family member has this condition;
- Avoid unnecessary use of medications during pregnancy.

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