

A Review on Craniofacial Anomalies

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Abstract:

Craniofacial anomalies (CFA) are a diverse group of deformities in the growth of the head and facial bones. Anomaly is a medical term meaning "irregularity" or "different from normal." These abnormalities are present at birth (congenital) and there are numerous variations. Some craniofacial anomalies are associated with anomalies elsewhere in the body, which can be serious. Cranio facial malformations include a variety of anomalies, including cleft lip with or without cleft palate, craniosynostosis, hemangioma, vascular malformation, and hemifacialmicrosomia. Congenital craniofacial abnormalities frequently require ophthalmic evaluation and surgical management. The term craniofacial anomalies include all congenital deformities of cranium and face. More specifically it refers to the deformities of the head that interfere with physical and mental well-being of an individual. It mainly deals with the understanding when, where and how the genes are expressed with specific pattern of morphogenesis.

Keywords: Craniofacial anomalies, Cleft lip, Cleft palate, Craniosynostosis, Hemangioma, Vascular malformation.

Introduction:

A craniofacial malformation is an anomaly of embryonic development that results in a serious impairment of the normal anatomy of skull, jaws and adjacent soft tissues. Most of the malformations diagnosed at birth fall in the category "craniofacial congenital anomalies". Children with craniofacial anomalies require a very detailed and unique medical support. Hence, geneticists, surgeons, paediatricians, neurosurgeons, orthodontists, ophthalmologists, speech therapists and many others who will take care of these patients should all have a very specific expertise in the field, because the problems of these patients often differ substantially from those of normal patients. The orthodontist as one of the many specialists within the craniofacial team plays a major role in stabilization and optimization of the craniofacial defects from the initial birth stages until skeletal growth maturation. Approximately 1% of these new-borns have syndromes or multiple anomalies. Syndromes are composed of multiple malformations thought to be etiologically and pathogenetically related. Syndromes that have cleft lip or cleft palate as one of the features are of in the quest for etiologic and pathogenetic factors, and it is estimated that 30% of cleft cases are

syndromic. Therefore, 70% are non-syndromic, thus craniofacial anomalies are among the most common birth defects and are associated with increased mortality and, in many cases, the need for lifelong treatment. ^[1]

The Common Types of Craniofacial anomalies:

* Cleft lip, an abnormality in which the lip does not completely form. The degree of the cleft lip can vary greatly, from mild (notching of the lip) to severe (large opening from the lip up through the nose).

* Cleft palate happens when the roof of the mouth does not completely close, leaving an opening that can extend into the nasal cavity. The cleft may involve either side of the palate. It can extend from the hard palate to soft palate. The cleft may also include the lip.

* Craniosynostosis is a condition in which the sutures (soft spots) in the skull of an infant close too early which interrupt the development of the brain and skull. Premature closure of the sutures leads to increased pressure in the cranial cavity resulting in the change in the shape of skull or facial bones.

* Hemifacialmicrosomia is a condition in which the tissues on one side of the face are underdeveloped. This mostly affects the ear (aural), mouth (oral) and jaw (mandibular) areas. Sometimes, both sides of the face can be affected and may involve the skull and the face. Hemifacialmicrosomia is also known as Goldenhar syndrome, brachial arch syndrome, facio-auriculo-vertebral syndrome, oculo-auriculo-vertebral spectrum, or lateral facial dysplasia.

* Vascular malformation is a birthmark or growth, present at birth, which is composed of blood vessels. It can cause functional or aesthetic problems. Vascular malformations may involve multiple body systems. There are four major categories of vascular malformations based on their flow characteristics: slow-flow (capillary malformation, venous malformation, lymphatic malformation) and fast-flow (arteriovenous malformation). These lesions often have components of multiple malformations, such as a mixed lymphatico-venous malformation, further adding to the confusion with respect to proper nomenclature ^[2].

Etiology:

Most medical professionals agree that there is no single factor that causes these types of abnormalities. A child may receive a particular combination of genes from one or both parents. Or, there may be a change in the genes at the time of conception. This results in a craniofacial anomaly. Studies have shown that women who do not take sufficient folic acid during pregnancy, or have a diet lacking in folic acid, may have a higher risk of having a baby with certain congenital anomalies.

Thus, it also includes:

- ✓ Chromosomal disorders
- ✓ Single gene disorders
- ✓ Multifactorial inheritance
- ✓ Maternal infections in pregnancy
- ✓ Maternal use of medications
- ✓ Radiation exposure

Cleft Lip and Cleft Palate

Cleft lip is defined as a congenital deformity that occurs in the primary palate which is located anteriorly to incisive foramen. Its occurrence may be unilateral, bilateral, complete or incomplete. Cleft palate is defined as a congenital abnormality that occurs in the secondary palate (soft and hard palate). Its occurrence may be unilateral, bilateral, complete or incomplete^[3,4]

Cleft lip and palate are congenital structural anomalies caused by atypical embryological development. Craniofacial differences are a result of interruption in embryologic growth between the 4th and 10th week of the developing embryo and fetus. Cleft lip and cleft palate (Fig.1 & 2) are openings or splits in the upper lip, the roof of the mouth (palate) or both. Cleft lip and cleft palate result when facial structures that are developing in a fetus don't close completely. Cleft lip and cleft palate are among the most common birth defects. Symptoms usually, a split (cleft) in the lip or palate is immediately identifiable at birth. Cleft lip and cleft palate may appear as: 1. A split in the lip and roof of the mouth (palate) that affects one or both sides of the face 2. A split in the lip that appears as only a small notch in the lip or extends from the lip through the upper gum and palate into the bottom of the nose 3. A split in the roof of the mouth that doesn't affect the appearance of the face .

*Causes: Cleft lip and cleft palate occur when maxillary prominence and medial nasal process don't fuse properly. Normally, they fuse together in the second and third months of pregnancy. But in babies with cleft lip and cleft palate, the fusion never takes place or occurs only part way, leaving an opening (cleft),

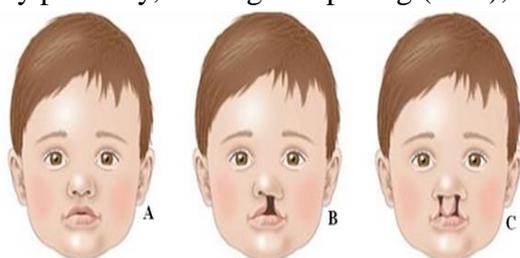


Fig. 1 Cleft lip
A. Normal B. Unilateral cleft lip C. Bilateral cleft lip

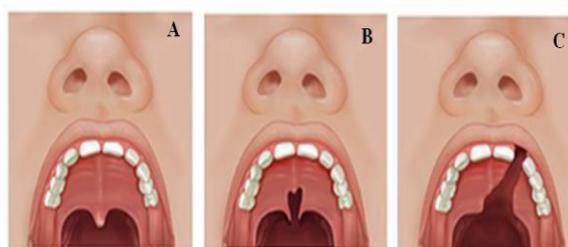


Fig. 2 Cleft palate
A. Normal B. Partial cleft palate C. Complete cleft palate

*Treatment: Children with a cleft lip or palate will need several treatments and assessments as they grow up. A cleft is usually treated with surgery. Other treatments, such as speech therapy and dental care, may also be needed.

Hemangioma:

The Greek suffix “oma” means cellular proliferation of a tumor and thus the term hemangioma is erroneous when used for malformations.^[5] They occur most frequently in head and neck region (60%), followed by the trunk (25%) and the extremities (15%), which are grouped into Infantile Hemangiomas (IHs) and Congenital Hemangiomas (CHs)^[6]

A hemangioma is a bright red birthmark that shows up at birth or in the first or second week of life. It looks like a rubbery bump and is made up of extra blood vessels in the skin. A hemangioma can occur anywhere on the body, but most commonly appears on the face, scalp, or chest (Fig.3). A hemangioma is made up of extra blood vessels that group together into a dense clump. What causes the vessels to clump isn't known.



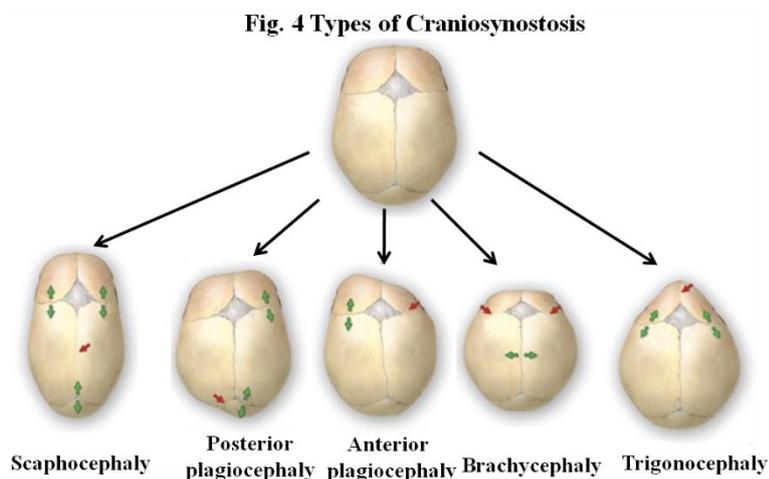
Fig. 3 Hemangioma
A. On face B. On scalp C. On lip

*Symptoms: Hemangiomas are usually painless, red to blue coloured lesions on the skin, lips, or inside the mouth. Superficial lesions may bleed or turn into sores, particularly if bumped or injured. Deep hemangioma in muscle may cause pain, as well as swelling around the it that increases with activity.

*Treatment:

Topical medicine on the skin rubbed onto the hemangioma. Topical beta blockers may help lighten the hemangioma and slow its growth. This works best on smaller, superficial hemangioma. Topical antibiotic can be applied when there are open sores with concern for infection. Oral medicine Propranolol is now the first-line treatment option for concerning hemangioma. Prednisone is used in select cases that may be resistant to other treatment, or if propranolol cannot be used. Surgery may be considered once the hemangioma has stopped growing or other treatments have failed. Laser may lighten the appearance of blood vessels left over from hemangioma.

Craniosynostosis:



It is a birth defect in which one or more of the fibrous joints between the bones of a new born baby's skull (cranial sutures) close prematurely (fuse), before the brain is fully formed. Brain growth continues, giving the head a misshapen appearance. Normally, during infancy the sutures remain flexible, giving the baby's brain time to grow. In the front of the skull, the sutures meet in the large soft spot (fontanel) on top of the head. The anterior fontanel is the soft spot you feel just behind the baby's forehead. The major complications associated with

uncorrected craniosynostosis include increased intracranial pressure, asymmetry of the face, and malocclusion. Asymmetry of the orbits leads to strabismus.^[7]

Types: As shown in (fig4),

*Scaphocephaly: It is a type of cephalic disorder which occurs when there is a premature fusion of the sagittal suture, it is the most common craniosynostosis condition characterised by long narrow head

*Posterior plagiocephaly: It is also known as unilateral lambdoid synostosis results in flattening of the back of the head on the affected side as well as compensatory growth of the mastoid process on the same side (ipsilateral mastoid bulge). This leads to a characteristic and unique “tilt” in the cranial base.

*Anterior plagiocephaly: It is the Premature fusion of a single coronal suture leads to a head shape called anterior plagiocephaly. This results in restricted anterior growth of the skull, involving the top of the skull as well as the cranial base

*Brachycephaly: It is caused by symmetrical flattening of the occipital bone area at the back, lower part of the skull. These infants have little or no rounding on the back of the head and a disproportionately wide head when viewed from the front.

*Trigonocephaly: It's a congenital condition of premature fusion of the metopic suture (from Greek metopon, "forehead"), leading to a triangular forehead. The merging of the two frontal bones leads to transverse growth restriction and parallel growth expansion. When the suture fuses prematurely the frontal bone and forehead cannot grow in response to the growth of the brain.

*Treatment:

Surgery is not performed in patients without increase ICP (Intra Cranial Pressure) until the shape of head does not improve by age, 2-4 months; it is unlikely to resolve with age.

Cosmetic surgery is performed in infants aged 3-6 months.

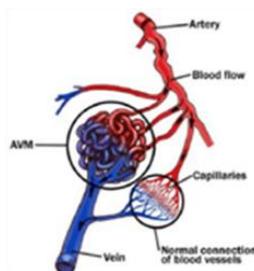


Fig 6. AVM

Vascular malformation:

It is a general term that includes congenital vascular anomalies of only veins, only lymph vessels, both veins and lymph vessels, or both arteries and veins (Fig 5). Vascular malformations are classified by the predominant vessel type within the lesion (capillary, venous, arterial, and lymphatic)^[8]

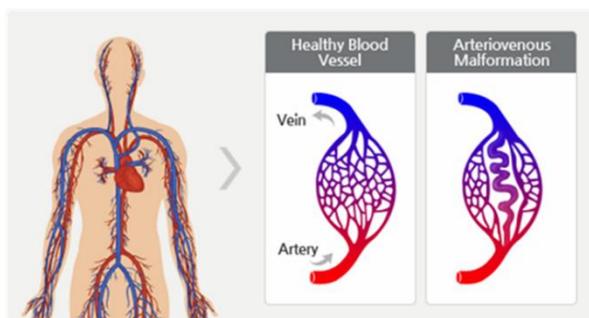


Fig 5. Vascular Malformation

TYPES:

Only veins: Venous Malformation (VM)

Only lymph vessels: Lymphatic Malformation (LM)

Both veins and lymph vessels: Veno-Lymphatic Malformation (VLM)

Arteries connected directly to veins without any capillaries in between: Arterio-Venous malformation (AVM) ,as in Fig 6.

* Causes: Vascular malformation are caused by development of abnormal direct connections between arteries and veins, but experts don't understand why this happens. Certain genetic changes may play a role, but most types are not usually inherited.

*Symptoms: Bleeding, headache, nausea and vomiting, seizures, loss of consciousness, back pain

*Treatment:

The most common techniques are embolization is minimally invasive procedure that closes the abnormal blood vessels from the inside using “glues” or particles. Laser Treatment can be effective for treating superficial venous malformation or the superficial component of a deep lesion. Sclerotherapy helps us treat venous malformations and lymphatic malformations.

Conclusion:

Orthodontic management for patients with craniofacial anomalies tends to be more complex, takes more time and clinical resources and should be based on a precise coordination with multiple dental, surgical and medical providers to achieve the best long-term esthetic and functional results. As the orthodontic management is commonly needed prior to most surgical procedures associated with craniofacial anomalies, management protocols should be based on a precise understanding of the exact nature of the anomalies as certain mechanics may be provided efficiently, safely and with acceptable durability, while at the same time, other techniques might be not effectively applied with some complications.

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