CRANIOFACIAL ANOMALIES

**Definition:**

Craniofacial anomalies consist of a variety of congenital deformities in the growth of the head and facial bones.

**Possible Causes:**

**Unknown** - in most cases but most scientists believe clefts are due to a combination of genetic and environmental factors.

**Genes** - a child may receive a particular combination of genes from one or both parents. There could also be a change in the genes at the time of conception.

**Environment** - exposure to chemicals or viruses to the fetus in utero. Some medications taken by the mother may also harm the fetus include: anti-seizure/anticonvulsant medications, acne medications containing Accutane, and Methotrexate (a drug commonly used for treating cancer, arthritis, and psoriasis). Congenital abnormalities have also been linked to maternal hypoxia, caused by maternal smoking, maternal alcohol abuse or some forms of maternal hypertension treatment. Other environmental factors that have been studied include: seasonal causes (such as pesticide exposure), retinoids (members of the vitamin A family), nitrate compounds, organic solvents, parental exposure to lead, and illegal drugs (cocaine, crack cocaine, heroin, etc.).

**Deficiency in Folic acid** - women who have an insufficient amount of folic acid (vitamin B found in leafy green vegetables, orange juice and grains) in their diet may be at a higher risk of having a baby with certain congenital anomalies.

**Common Types:**

**Cleft lip** - lip does not completely form and can be mild (notch in vermillion) to severe (up the lip through alveolar ridge and through incisive foramen).

**Cleft palate** - when the roof of the mouth does not completely close, leaving an opening that can extend into the nasal cavity. It may be unilateral (does not extend into the nose) or bilateral and can run through the hard palate to the end of the uvula.

**Craniosynostosis** - when the sutures (soft spots) in the skull of an infant close too early, causing problems with normal brain and skull growth. Premature closure of the sutures may also cause the pressure inside of the head to increase and the skull or facial bones to change from a normal, symmetrical appearance.
**Hemifacial microsomia** - when the tissues on one side of the face are underdeveloped, affecting primarily the aural, oral, and mandibular areas. Sometimes, both sides of the face can be affected and may involve the skull, as well as the face. Also known as Goldenhar syndrome, brachial arch syndrome, facio-auriculo-vertebral syndrome (FAV), oculo-auriculo-vertebral spectrum (OAV), or lateral facial dysplasia.

**Vascular malformation** - a congenital birthmark or growth made up of blood vessels that can cause functional or aesthetic problems and may involve multiple systems of the body. Also known as lymphangiomas, arteriovenous malformations, and vascular gigantism.

**Hemangioma** - a type of birthmark which may look like a faint red mark at birth or appear in the first few months of life. It is the most common benign tumor of the skin. Also known as a port wine stain, strawberry hemangioma, and salmon patch.

**Plagiocephaly** - asymmetrical shape of the head due to repeated pressure to the same area.

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**Cleft Lip and Cleft Palate**

**Statistics:**

- The 4th most common birth defect in the US and affects 1/700 births each year.

- Less common among Africans and African Americans (about 1/1500) and more common among Asians (about 1/4000), also common among Latinos and Native Americans.

- Affects twice as many boys than girls who are born with a cleft lip, both with and without a cleft palate. And most babies born with a cleft are otherwise healthy with no other birth abnormalities.

- Cleft palate occurring alone is more common in girls than boys.

- Unilateral cleft are more common on the left than the right.

- Bifid/split uvula occurs in 1/1200 people in the general population.

- Higher incidence when more people in the family have a cleft.

**Growth of Palatal Plates in Fetus:**

Cleft of hard palate occurs between week 6 and 10.
Cleft of soft palate occurs between week 10 and 12.

Bifid uvula occurs by week 12.

**Associated with a Syndrome, such as:**

-Van der Woude Syndrome

-Siderius X-linked mental retardation, and in addition to cleft lip and/or palate, symptoms include facial dysmorphism and mild mental retardation.

**Associated with Syndromes, which also cause other problems:**

-Stickler's Syndrome can cause cleft lip and palate, joint pain, and myopia.

-Loeys-Dietz syndrome can cause cleft palate or bifid uvula, hypertelorism, and aortic aneurysm.

-Hardikar syndrome can cause cleft lip and palate, hydronephrosis, intestinal obstruction and other symptoms.

**Associated with Many Different Chromosome Disorders:**

-Patau Syndrome (Trisomy 13)

-Treacher Collins Syndrome

-Velo-Cardio-Facial Syndrome

-Pierre-Robin Sequence

**Surgery:**

A nasoalveolar mold placed before surgery can help improve long term nasal symmetry with unilateral cleft lip/palate patients when compared to only having surgery.

A prosthetic device which fits into the roof of the mouth for a cleft palate is called the palatal obturator.
Cleft lip is usually repaired by 4-12 weeks around the country. It is repaired at 6-8 weeks in Utah. Surgery can be performed soon after birth, but the preferred age is at approximately 10 weeks of age. This follows the "rule of 10s" created by surgeons Wilhelmsen and Musgrave in 1969 (the child is at least 10 weeks of age; weighs at least 10 pounds, and has at least 10g hemoglobin).

Cleft palate can be repaired between 6 and 12 months. If the cleft is bilateral and extensive, two surgeries may be required to close the cleft, one side first, and the second side a few weeks later.

An incomplete cleft lip requires the same surgery as complete cleft since the group of muscles required to purse the lips run through the upper lip and a full incision is made. Then the surgeon tries to line up the scar with the natural lines in the upper lip (the edges of the philtrum) and tuck away stitches as far up the nose as possible.

About 10-20% only need one surgery for normal, non-hyper nasal speech. While others need a combination of surgeries and procedures as they grow.

The Latham appliance may be surgically placed between the 4th and 5th month to help with future lip or cleft repairs. Pins are used to keep it in place and the parents turn a screw every day to bring the cleft together.

Bone tissue taken from the child's own hip, chin or rib may also help fill in the cleft extending into the maxillary alveolar ridge.

Orthodontics (beginning around age 7 up to age 18) and/or further cosmetic corrections, including jawbone surgery (ages 15-18).

**Post-Surgery:**

There will be stitches on the palate where the cleft was repaired which will dissolve several days after surgery. Some children go home with packing placed on the palate.

Bloody drainage coming from the mouth and nose will become less after the first day.

Swelling will also go down, within the first week.

The child will usually go home with a prescription for pain. They may also be given a prescription for nasal congestion, which may include decreased appetite, mouth breathing and nasal snorting.

To prevent infection, antibiotics may be prescribed also. And a small amount of water can be used to gently clean the area several times daily.

No straws or pacifiers should be used as they could damage the repair.

For the first 7-10 days, a soft diet is recommended.
**Feeding Issues:**

The severity of the cleft affect the child's ability to suck. Babies born with a cleft lip, a small cleft or sub mucosal cleft can be successful breast feeders and may also need assisted milk delivery. Babies with:

-Cleft lip... need a nipple with a wide base. Place the nipple on the unaffected side. Breast and bottle are fine.

-Cleft palate... use a soft nipple with a larger hole (preemie nipple) for bilateral. The NUK nipple can be placed on regular bottles with disposable bags. The Mead Johnson Nurser set comes with a soft nipple with a crosscut end which allows the baby to bite down to release liquids. The nipple is slightly longer than most which places the nipple farther back on the tongue. The Haberman Feeder has a slicecut hole in the nipple and the liquid stays in the nipple once it has been sucked. This bottle system is designed with a valve to help control the amount the baby drinks and to prevent milk from going back into the bottle.

There are common breast feeding problems that may arise with this population. If there is milk leaking from the baby's nose, try using a semi-upright feeding position. If the baby is unable to hold the nipple in his mouth, have the mother use her finger to hold his lips together or put the nipple in the side of his mouth. If the baby gets tired too quickly, feed him more often (every 3 hours) and for shorter periods of time (20 mins). If the baby does not empty the breast, be sure to have mother express her milk to maintain her milk production. When the baby's suck is weak the milk "letdown" is difficult. Make sure the mother is getting enough rest and she may need to use a breast pump to express her milk after the baby nurses.

Even though palates are not always repaired by 6 months, it is appropriate to introduce strained/lumpy foods. Place only small amounts of food on the spoon and allow for the child to taste it and set the pace. Present these foods slowly. The poorly swallowed food will come up through the nose and can be very uncomfortable. Some learn where to put their tongue to redirect the food better than others. Citrus fruits and tomatoes are acidic and should be avoided until the child has learned better control.

**Hearing Loss:**

Tympanostomy tubes are usually placed into the eardrums to aerate the middle ear at 6-12 months of age.

Hearing impairment is prevalent in children with cleft palate. The tensor muscle fibers that open the Eustachian tubes are impaired and don't function properly. Contaminates from the pharynx go up into the Eustachian tube (next to the adenoids) because of the improper closure of the tube. It then swells up and a middle ear infection is created.
**Speech Therapy:**

Velopharyngeal inadequacy (inability of the soft palate to close the opening from the throat to the nasal cavity) typically resolves after palate repair. Speech errors may develop as the result of an attempt to compensate for the inability to produce the target phoneme. These compensatory articulations are usually sounds that are non-existent in normal English phonology and often do not resolve automatically after palatal repair causing a decrease in speech intelligibility.

The 16 pressure consonants affected are: plosives (6) and fricatives (10). The sounds /N/ and /L/ are not produced in error unless related to a malocclusion and can be produced with a recessed upper lip instead of premaxilla, behind teeth.

The wider the opening between the oral and nasal cavities the more nasality is noted. Start therapy on the low vowels (easiest) then on the high vowels (more difficult).

Denasality is a medical problem and can’t improve with therapy. It may be caused by a pharyngeal flap being too large, deviated nasal septum (common in unilateral), large adenoids or nasal polyps, obturator bulb too big, nasal congestion or scarring in nasopharynx due to pre- or post-surgical infection.