CLEFT LIP AND/OR PALATE (CLAP)

Definition

- **Cleft** is an abnormal opening or a fissure in an anatomical structure that is normally closed. Cleft lip and palate is the ***fourth*** *most common birth defect* and the most common congenital defect of the ***face***. However, oral clefts don't have to happen together—an individual can have one without the other (Kummer, 2018).
- **Cleft lip** is the result of failure of parts of the lip to come together early in the life of a fetus.
- **Cleft palate** occurs when the sections of the roof of the mouth do not fuse normally during the fetal development, leaving an opening between the oral cavity and the nasal cavity.

Embryological Development

- Cleft lip and palate occur when certain facial structures don't form correctly during development in the womb. This is due to problems with ***neural crest cells***, which are special cells that help form the face and skull. If these cells don't migrate or move correctly, it can lead to clefts and other facial abnormalities (Kummer, 2018).
- **Lip**: Starts around **6-7 weeks**, begins at the front and moves towards the sides.
- **Palate**: Starts around **8-9 weeks**, begins at the front and moves towards the back.

Etiology

The development of cleft lip and palate occurs due to the failure of normal median fusion of the oral-facial structures during the **seventh to twelfth weeks of gestation** (Kummer, 2018). This failure can be influenced by a range of causes rather than a single factor, leading to the clinical presentation of clefting. Several key causes include:

Exogenous Factors - *external* factors that may affect the clefting

- Environmental Teratogens - These are substances that can cause congenital malformations.
  - Cigarette Smoke
  - Certain drugs like phenytoin, valium, and corticosteroids
  - Lead pollution
  - Viruses like rubella and influenza
  - Maternal nutritional deficiencies, particularly lack of vitamin B-6
  - Maternal obesity and underweight (Kutbi, 2014)
  - Especially those with lower education levels, had a higher risk of having children with orofacial clefts. Additionally, mothers with diabetes (gestational or pre-gestational) and hypertension are also at higher risk. The combination of these factors further increases the risk. Metabolic syndrome, a cluster of health risks, can also increase the risk of orofacial clefts in children.
- Mechanical Interference
  - This can occur in utero when the developing fetus is crowded, leading to abnormal positioning of the head and tongue.
  - This can interfere with palate formation, resulting in a cleft palate, often with a wide, bell-shaped appearance.

Endogenous Factors - *internal* factors that may affect the clefting

- Chromosomal Disorders
  - These are abnormalities in the number or structure of chromosomes.
  - Examples: Down syndrome and Turner syndrome
- Genetic Disorders
  - These are inherited conditions that can affect the development of the face and

mouth.
    - Examples: Stickler syndrome and Treacher Collins syndrome.
- Older Parental Age
    - A study Father's aged 40 years or more have and increased risk of having a child with CLAP (De Carvalho et al., 2016).

Multifactorial Inheritance - combination of factors that may affect the clefting.

- In most cases, the cause of a cleft is not due to a single factor but rather a combination of genetic and environmental factors. This is known as multifactorial inheritance.

Prevalence & Incidence

Locally:

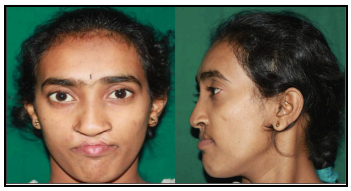
- 1 out of 1,000 Filipino live births, a cleft lip or palate patient is born. (Cleft Lip and Palate, 2017)

Internationally:

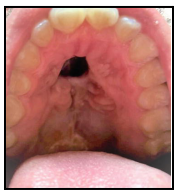
- According to the World Health Organization's (WHO) Global Oral Health Status Report (2022), oral diseases are estimated to affect around 3.5 billion people globally. Globally, orofacial clefts were estimated to affect 1 in 1000–1500 babies.

Manifestations of the Disease that the Physician/Allied Health Medical Professional Perceives

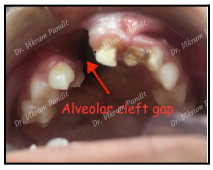
- Crooked, poorly shaped or missing teeth
- Dental problems: misaligned teeth and jaw
- Ear infections and hearing loss due to a dysfunction of the eustachian tube that connects the middle ear and the throat that could then lead to hearing loss.
- Deformed/misaligned upper jaw (maxilla)



- Unrepaired oronasal fistulae - hole between the mouth and nose



- Alveolar clefts - defects in the bone that supports the teeth



Manifestations the Patient Experiences

- Abnormal nasal resonance
    - Caused by a combination of velopharyngeal dysfunction (VPD) and anterior nasal complications leading to an inability to effectively manage the air stream for continuous speech
- Difficulty swallowing/feeding
    - Potential for liquids or foods to come out the nose
    - Unable to suck properly
- Abnormal speech and language delay
    - Evidenced by slower acquisition of sounds and words and restricted inventory of sounds in early infancy.
- Muscle function may be decreased, leading to abnormal speech
- Psychological challenges of coping with CLAP
    - Greater behavioral problems, more episodes of depression and low self-esteem
    - Complex psycho-social effects due to un-operated condition

Structural and Anatomical Changes Related to the Condition

Cleft Lip

- Short Philtrum With One Or Both Columns Affected
- Pattern Of Nasal Deformities (Nasal Septum, Short Columella, Nasal Cartilage)
- Alveolar Ridge Misaligned—May Cause Gap In The Upper Gum Line

Muscles affected:

- Orbicularis Oris muscle is the primary muscle of the lip.
  - Deep component - works with other oropharyngeal muscles in swallowing and serves as a sphincter.
  - Superficial component - a muscle for facial expression and connects to the anterior nasal spine, the base of the nostrils (sill), the sides of the nostrils (alar base), and the skin. This muscle helps form the philtral ridges, the vertical lines above the upper lip.
- *Complete cleft lip* - the main lip muscle (orbicularis oris) is split by the cleft and ends on either side of the gap instead of going around the mouth. The outer part of this muscle curves upward along the edges of the cleft and stops below the nostrils.
- *Incomplete cleft lip* (gap is less than two-thirds the height of the lip) - muscle fibers follow the edges of the cleft and then run across the top, mixed with connective tissue.
- *Bilateral cleft lip* - structure depends on how complete the cleft is. The middle part of the lip (prolabium) is usually pulled back, doesn't have muscle fibers, and can look like it's attached to the nose.

Bones affected:

- Premaxilla - bone that holds the front teeth (incisors) and the alveolus
- Maxilla - upper jawbone; The cleft can cause the maxillary arches to collapse or the premaxilla to protrude if left unrestrained.
- Alveolar Ridge - bony ridge that holds the teeth, and when affected by a cleft, it can disrupt dental alignment and development.

<u>Cleft Palate</u>

- Incomplete Fusion Of The Palate—Opening Between The Oral Cavity And The Nasal Cavity Causing
- Difficulty With Coordination Of Sucking, Swallowing, And Respiration During Feeding
- Abnormal Uvula
- Eustachian Tube Complications

Muscles affected:

- Levator veli palatini - Key muscle for speech and velopharyngeal competence; normally forms a sling to elevate the soft palate.
- Palatopharyngeus - Helps move the soft palate backward.
- Superior pharyngeal constrictor - Aids in closing the velopharyngeal opening.
- Tensor veli palatini - Functions to dilate the eustachian tube and maintain its integrity.
- Uvular muscle - Minimal contribution to speech.
- Velar musculature - Often underdeveloped (hypoplasia) in cleft palate, with abnormal muscle insertions along the cleft.

Bones affected:

- Incisive foramen - Key landmark in the bony palate.
- Premaxilla - Located in front of the incisive foramen; can vary in size and composition.
- Alveolus and incisors - Part of the premaxilla.
- Maxillary arches - May collapse and exclude the premaxilla if unrestrained.
- Hard palate - Formed by the palatine processes of the maxilla and palatine bones.
- Palatine shelves - Collapse posterior to the premaxilla in complete bilateral clefts.

<u>Cleft Lip And Palate</u>

- Facial Deformities
- Dental Conditions

Muscles Affected:

- Orbicularis oris (for cleft lip) - The muscle around the mouth is split by the cleft, disrupting both its deep (swallowing) and superficial (facial expression) components.
- Levator veli palatini (for cleft palate) - Essential for speech and soft palate elevation, often improperly inserted along the cleft.
- Tensor veli palatini, palatopharyngeus, superior pharyngeal constrictor, and uvular muscle - Muscles involved in speech and swallowing may be abnormally positioned, contributing to velopharyngeal dysfunction and feeding issues.

Bones Affected:

- Premaxilla and alveolus - The bone holding the upper teeth may protrude or fail to fuse properly with the rest of the maxilla, resulting in gaps.

- Maxilla and palatine bones - The upper jaw and hard palate are often misaligned, causing collapse or protrusion of the premaxilla, disrupting the integrity of the palate.

Possible SLP Areas Affected and Their Characteristics

Cognition	• May be at risk for reading delays and may have higher incidence of learning disabilities who demonstrate average intelligence and a slightly lower verbal IQ (Hardin-Jones & Chapman, 2011).
Hearing	• Cleft palate-related hearing impairments usually go away by the time a child is six or eight years old, but they can sometimes result in more serious hearing problems in the long run. ◦ **Conductive hearing loss** is most commonly associated with CLAP, this is most likely due to: ▪ More than 90% of newborns with cleft palates have **Otitis Media with Effusion (OME)** associated with eustachian tube dysfunction, which causes middle ear abnormalities that result in lifelong hearing loss in adults. This results in more than 50% of the adult CLAP population having permanent hearing loss.
Speech	• Continuous mispronunciation of sounds as a result of compensatory speech patterns or residual structural abnormalities. • Errors in speech production are noticed due to the abnormalities in oronasal structure / function, orofacial structure and growth, learned neuromotor patterns during early infancy, and / or disturbed psychosocial development • **Obligatory Errors** ◦ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ◦ The most common obligatory error is *glottal stop substitution*. ▪ occurs when a consonant is substituted with a sound produced in the back of the throat called a glottal stop . ▪ Ex. the sharp, middle sound in "uh-oh". ◦ Error is due to *structural differences* • **Compensatory Errors** ◦ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD ◦ Error is a *learned or* adapted less effective way to produce sounds • **Velopharyngeal dysfunction (VPD)** is the inability to separate the oral and nasal cavities adequately during speech production through the actions of the velum and pharynx. This can be caused due to either a: ◦ **Velopharyngeal insufficiency** - lack of tissue ◦ **Velopharyngeal incompetence -** lack of proper movement of the walls.

<table>
<tr>
<td>Feeding and Swallowing</td>
<td>
○ Difficulty in swallowing food or liquids.
<table>
<tr><th>Adults with unrepaired CLAP</th><th>Adults with repaired CLAP</th></tr>
<tr>
<td>● The cleft would make it difficult to create the necessary suction to pull food and drinks into the mouth.
● There is a risk for aspiration as food and liquid may enter the lungs, which could lead to aspiration pneumonia.</td>
<td>● There could be some dental issues from misaligned teeth or missing teeth, which could complicate feeding and swallowing.
● The lips, palate, and tongue might become immobile and lose their ability to function due to scar tissue from prior procedures.</td>
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<td>Pragmatics</td>
<td>
● Difficulties in social contexts with understanding and using language.
○ This could be due to cognitive impairments resulting from:
■ Early life experiences that had a lasting impact on cognitive development can include emotional difficulties, social isolation, hearing loss, and other challenges. These effects can persist even after successful surgery.
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<td>Resonance</td>
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● Hypernasality
○ Excessive nasal resonance caused by inadequate velopharyngeal closure.
○ The nasal cavity also resonates sounds that are primarily intended to be produced in the oral cavity.
○ Using "sea" or "know" as an example, they might sound more like "shee" or "no-oh."
● Hyponasality
○ Reduced nasal resonance caused by obstruction to the nasal passages.
○ Perceived when there is reduced nasal resonance on vowels, sonorants, and nasal consonants.
○ In more extreme cases, hyponasality co-occurs with denasalization of nasal consonants (/m/, /n/, and /ŋ/), making them sound more oral in quality (e.g., /b/ for /m/, /d/ for /n/, and /g/ for /ŋ/)
● Mixed Resonance
○ Has both qualities of Hypernasality and Hyponasality.
● Cul-de-sac Resonance
○ An obstruction at the outflow of one of the vocal tract's cavities (nasal, oral, or pharyngeal) causes sound to be muted and low in volume (Kummer, 2020).
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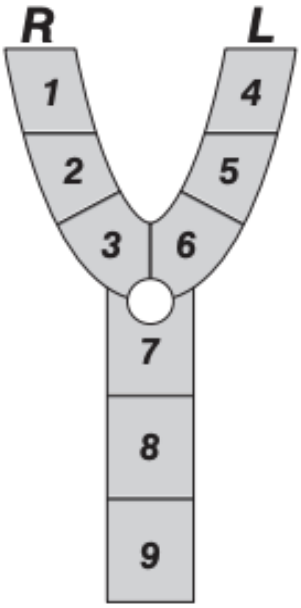
Classification

➔ ***Primary Palate*** - structures that are **anterior** to the incisive foramen. It includes the *alveolus and the lip.* These are the structures that fuse around 7 weeks of gestation.

➔ ***Secondary Palate*** - structures that are **posterior** to the incisive foramen. It includes the *hard palate and the velum*. These are the structures that fuse around 9 weeks of gestation.

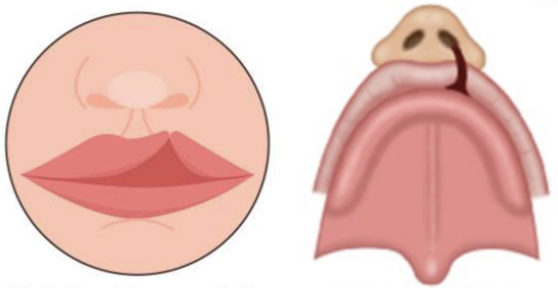
★ The **Kernahan and Stark Classification System** is a visual tool used to describe the extent of a cleft lip and palate. It uses a "Y"-shaped diagram with numbered segments to represent different parts of the mouth.

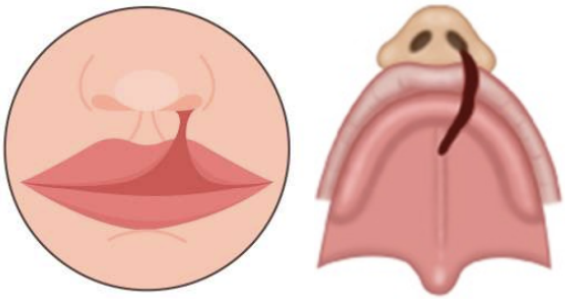
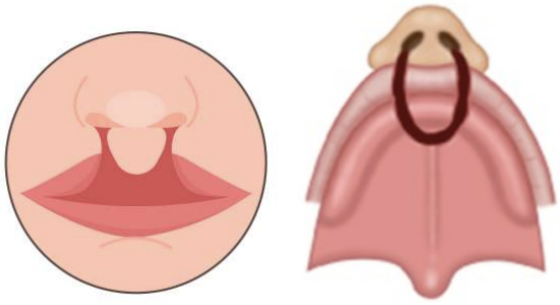
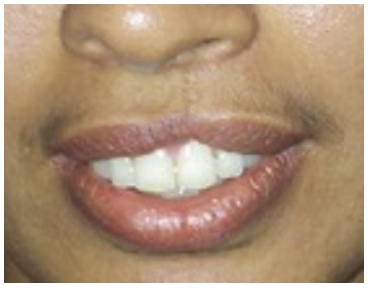
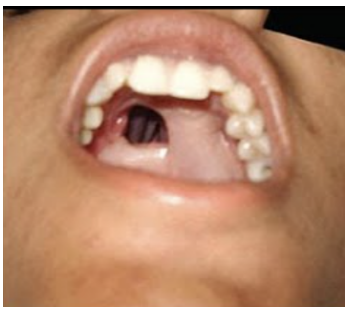
- The upper arms of the Y represent the *primary palate* (lip and alveolar ridge), and the base represents the *secondary palate* (hard and soft palate).
- Each segment is numbered from 1 to 9.
  - **1-3**: Right side of the primary palate
  - **4-6**: Left side of the primary palate
  - **7-9**: Secondary palate
- Darkened segments indicate the affected areas of the cleft.
- Crosshatch marks indicate a submucous cleft (hidden cleft).
- **How to use the system**:
  - ***Identify the affected segments*:** Look at the diagram and darken the segments that are affected by the cleft.
  - ***Describe the cleft*:** Use the numbers to describe the extent of the cleft. For example, a cleft lip and palate might be described as "1, 2, 3, 4, 5, 6, 7, 8, 9."
  - ***Indicate submucous clefts*:** Use crosshatch marks to show hidden clefts.



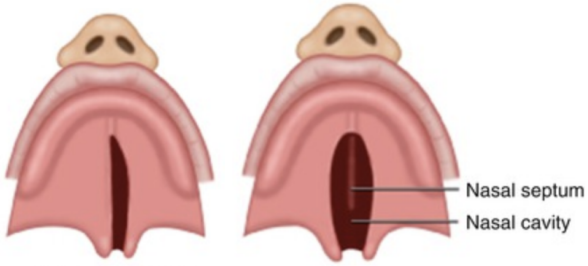
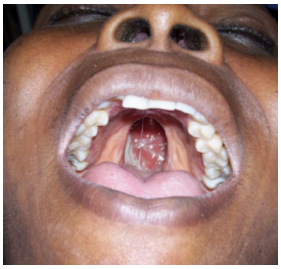
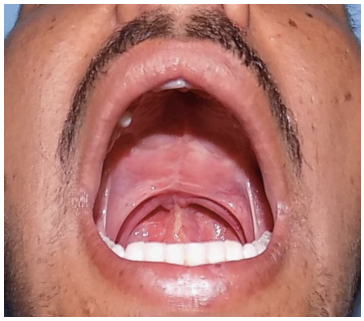
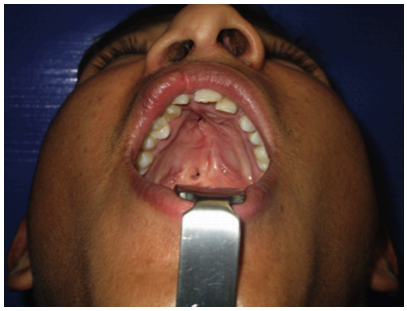
Clefts of the Primary Palate

Incomplete - Does not extend to the incisive foramen; it can be as minor as a small subcutaneous notch in the vermilion, or it may involve the entire lip and part of the alveolus

Unilateral Incomplete Incomplete, one-sided cleft of the primary palate; often occurs on the left side	

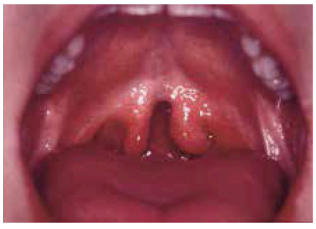
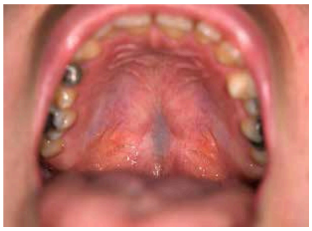
Bilateral Incomplete Incomplete, both left and right sides are affected	
Complete - Extends through the entire lip, nostril sill, and alveolus to the incisive foramen	
Unilateral Complete Complete, one-sided cleft of the primary palate	
Bilateral Complete Complete, both left and right sides are affected	
Form Fruste or **Microform Cleft** Rare; It is a partial or arrested form of a cleft lip	
Nasolabial Fistula Residual that patients who have had a cleft of the primary palate surgery/repair; located in the alveolus just under the upper lip	

Clefts of the Primary Palate

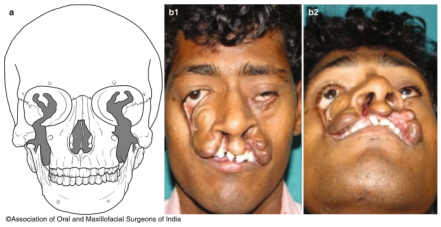
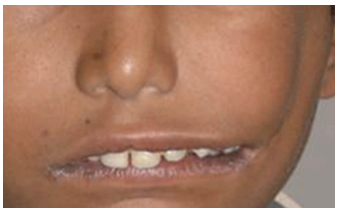
Complete Goes through the uvula and the velum, and then follows the median palatine suture line through the hard palate all the way to the incisive foramen	 
Incomplete May be as slight as a line in the midline of the uvula or a bifid uvula; a more severe cleft palate may extend into the velum or into part of the hard palate	
Isolated Cleft Palate	Solely cleft palate; has no involvement of the lip
Palatal Fistula Residual (hole) that patients who have had a cleft of the secondary palate surgery/repair	

Submucous Cleft Palate

A congenital defect that affects the underlying structure of the palate, whereas the oral surface mucosa is intact

Overt Seen on the oral surface and therefore, can be identified through an intraoral examination	
Occult Defect in the velum that is not apparent on the oral surface; it can only be seen by viewing the nasal surface of the velum through nasopharyngoscopy; occult means "hidden" or "not revealed"	

Facial Clefts

Midline (Medial**)** It can be very mild so that there is only a notch in the midline of the vermilion or a slight cleft of the upper lip	
Paramedian Most cleft cases; One or both sides of the face is affected; located near the midline	
Orbital Impacts the eye area; Involves the eyes/orbit	
Oblique Can be unilateral or bilateral; It begins at the mouth and then courses laterally, horizontally, and upward	

Progression of the Condition	**Outcome if Left Treated and/or Untreated**
• Since cleft lip and palate are birth defects, it doesn't progress on its own	• If left untreated, it can lead to several challenges related to feeding, speech, hearing, dentition, and socioemotional difficulties.

Medical/Surgical Management

Healthcare Resources

Medical Care

- Otolaryngology (ENT) Services - Manage ear infections, hearing issues, and breathing difficulties often associated with CLAP. They can perform surgical interventions to improve function and quality of life.
- Dental Health Care - Helps manage dental abnormalities, assists with proper tooth alignment, and supports overall oral health. This can improve speech, eating, and appearance.

Surgical Care:

Plastic Surgery - Corrects cleft lip and palate, improves facial symmetry, and enhances overall appearance.

- Cheiloplasty - Primary Cleft Lip Repair
- Palatoplasty - Cleft Palate Repair
- Z-Plasty or Pharyngeal Flap - Lengthen the soft palate and decrease nasal escape of speech sounds
- Fistula Repair - Repair abnormal opening after palatoplasty

- Alveolar Bone Graft - Uses bone marrow from the hip to fill a gap in the gum line
- Maxillary Distraction Osteogenesis - Surgically freeing the upper jaw and moving it forward slowly over several weeks using a rigid external device called a halo.
- Lip and/or Nasal Revision - Lip and/or nasal revisions can be done at any age to improve appearance, but delaying treatment until facial growth is complete (around 16 or older) can reduce the need for future revisions.

Specific Surgical Procedures for Adults

- Millard Unilateral Cleft Lip Repair - for incomplete cleft lip
- Mohler's modification of Millard's Repair - for complete cleft lip
- Primary Nose Correction - for positioning lower lateral cartilage more medially and superiorly with dome to dome suturing to the opposite
- Two Flap technique w/ Intravelar Veloplasty - for complete cleft palate repair (Sommerlad, 2003)
- Von Langenback technique w/ Intravelar Veloplasty - for incomplete incomplete cleft palate
- Alveolar Extended Palatal Flap - to avoid post-alveolar fistula
- Mulliken Modification of Millard's technique - for bilateral cleft lip to symmetrical and aesthetically pleasing result, while also restoring the function of the lip and nose.
- Primary Abbe Flap - for patients with Median Facial Dysplasia to address tissue deficiency in the prolabium.
- Hole-in-One Procedure - for simultaneously repairing both the cleft lip and cleft palate in a single surgical intervention; for very wide clefts, an extended periosteal flap can be used to address the alveolar region
- Primary Pharyngeal Flap - address velopharyngeal insufficiency (VPI), a condition where the soft palate fails to close off the nasal cavity during speech, resulting in hypernasality.

Orthognathic Surgery - Corrects jaw misalignments and facial skeletal deformities.

POST-SURGERY CARE & MANAGEMENT

	CLEFT LIP	**CLEFT PALATE**
Recovery Period	Can take up to four weeks to recover from surgery	
Appearance	• After cleft lip surgery, there will be stitches and swelling around the lip and nose. • Care for the stitches as instructed by nurses. • The stitches will dissolve in about 7-10 days. • The scar may appear "pulled up" at first, but it will settle over time. • Gentle massage can help with this.	• After cleft palate surgery, there will have stitches in the mouth that will dissolve in 7-10 days.
Pain Management	• Patients may be a little unsettled for 1–3 weeks and require extra attention. • Give paracetamol and/or codeine as required for pain relief, usually only for a few days. • Contact the hospital for any concerns.	• Patients may be a little unsettled for 1–3 weeks and require extra attention. • Give paracetamol or codeine for pain regularly is recommended for the first few days, especially before feeds • Contact the hospital for any concerns.

Diet	• Food should be thinned down or eaten with a cup.	• Use a spoon carefully for any puréed solids. • Avoid hard foods such as toast, biscuits or rusks during healing.
Precautions	• Avoid aspirin as it may cause bleeding. • Straws are generally not safe to use. • Avoid bumping the lip after repair.	• Avoid aspirin as it may cause bleeding. • Avoid placing any objects such as straws into the mouth. • Avoid bumping the palate after repair. • You need to keep the mouth as clean as possible and should rinse the mouth with cooled boiled water after feeding.

Obturators (Prosthetic Management):

- This helps individuals with cleft lip and palate (CLAP) achieve better feeding, speech, and overall development.
- Typically made of acrylic resin and often include soft materials for comfort. They can be held in place by extraoral devices or by engaging the palate's undercuts.

Kinds of Obturators

- Palatal Obturators - to seal the cleft in the palate, separating the oral and nasal cavities
- Speech Bulb Obturators - to close off a portion of the pharynx when the velum is very short
- Palatal Lifts - to elevate the velum when there is velopharyngeal insufficiency (VPI)

Steps in Fabricating a Obturator

Traditional Method

- Material Selection: Using auto- or heat-polymerized acrylic resin for the main body of the obturator, and soft, elastic material for the undercut lining.
- Mold Creation: Taking a detailed impression of the palate to create a mold for the obturator.
- Fabrication: Constructing the obturator using the mold, ensuring proper fit and function.
- Retention: Attaching the obturator to extraoral devices or using soft material to engage the undercuts for secure placement.

Vacuum-Formed Method

- Material Selection: Using ethylene-vinyl soft materials commonly used for athletic mouthguards.
- Mold Creation: Taking a detailed impression of the palate.
- Vacuum Forming: Heating the material and drawing it over the mold to create the obturator.
- Finishing: Trimming and adjusting the obturator to ensure proper fit and function

Rehabilitation Care

- Speech-Language Therapy - Address speech sound disorders, articulation difficulties, and swallowing problems.
- Aural Habilitation & Rehabilitation - Provide hearing aids, auditory training, and counseling to help individuals with hearing loss communicate effectively.

SLP Therapy

Areas	Strategies	Assessment & Materials	Approaches
Speech To enhance articulation,	• Target specific error sounds, especially those	• Oral Peripheral Mechanism Test • Bauman-Waengler	• Traditional articulation therapy

phonology, clarity and intelligibility.	affected by velopharyngeal dysfunction • Use visual feedback (e.g., mirrors, video recordings) • Implement oral motor exercises to improve muscle strength and coordination • Identify articulation errors characterized as *obligatory errors* (due to structural differences) or as *compensatory* (learned) errors	• Speech-Motor Screening • Frenchy Dysarthria Assessment • Goldman Fristoe Test of Articulation 3rd Edition (GFTA-3) • Passage and Conversation • Any articulation screener with sounds in different placements	• Phonological process approach • Cycles approach for multiple sound errors
Resonance Improve overall voice quality and speech naturalness.	• Focus on reducing hypernasality or hyponasality • Teach proper oral-nasal balance	• Speech Screening • Assessment for velopharyngeal dysfunction ○ Oral inspection ○ Acoustic nasometry ○ Mirror tests ○ Nasoendoscopy with a photo or video recording. ○ Videofluoroscopy for velum function. ○ MRI • Comprehensive Assessment	• Continuous positive airway pressure (CPAP) therapy • Biofeedback techniques (e.g., nasometry) • Resonance therapy exercises
Voice To address issues such as hoarseness or breathiness and achieve a clearer, more efficient voice production.	• Address compensatory articulation patterns • Improve vocal quality and pitch control • Assess for voice quality and signs of vocal fold pathology (e.g., hoarseness, breathiness).	• Auditory-perceptual assessment • Instrumental assessment • Assessment of voice handicap • Referral for ear, nose, and throat assessment	• Vocal Range and Flexibility Exercises • Voice Therapy Programs • Vocal Hygiene • Breath Support Training

Feeding and Swallowing	• Improve oral motor control • Address any structural issues affecting swallowing	• Eating and drinking abilities (e.g. oral containment, sucking, mastication) • Secretion management • Oral hygiene practices • Sensory status • Acceptance of food • Diet diversity • Oral medication management • Caregiver behaviors during feeding • Psychosocial impact on family and child dynamics	• Obturator Fitting • Oral Motor Exercises • Swallowing Maneuvers
Language and Pragmatics	• Address any language delays or disorders resulting from hearing loss or social factors • Improve social communication skills by increasing interactions for more opportunities for language growth	• Comprehensive Assessment of Spoken Language 2nd Edition (CASL-2) • Clinical Evaluation of Language Fundamentals Fifth Edition (CELF-5) • Western Aphasia Battery (WAB) Test • Structured and Unstructured Conversations	• Cognitive-linguistic Therapy • Social Skills Training • Script Training or Narrative Interventions • Pragmatic Language Intervention • Augmentative and Alternative Communication (AAC) ○ If speech is severely impaired, AAC strategies may be used to support communication.
Hearing	• Assess detection, discrimination, identification, and comprehension	• Ling- 6 & Ling-10 Test • Early Listening Function (ELF) • Glendonald Auditory Screening Procedure (GASP)	• Listening and Spoken Language Therapy • Auditory-Oral or Auditory-Based Speech Therapy • Total Communication Approach

Cognition	• Target specific cognitive domains (e.g., attention, memory, executive function) • Implement compensatory strategies • Provide environmental modifications	• Western Aphasia Battery (WAB) Test • Montreal Cognitive Assessment (MoCA) • Mini Mental State Examination (MMSE)	• Cognitive Stimulation Therapy • Attention Process Training • Memory Strategy Training

Educational Management

- **Comprehensive Education**
  - Long-term care plans and education help families manage cleft/craniofacial anomalies, improving adaptation and reducing complications.
- **Targeted Interventions**
  - Raising awareness of nutrition, dental issues, and speech abnormalities helps alleviate mental stress and improve disease management.
- **Preventive Education**
  - Increased awareness prevents further complications, improving quality of life for patients and families.
- **Caregiver and Parental Support**
  - Educating and equipping parents and caregivers with the skills to manage stress and carry out treatment significantly improves patient outcomes.
- **Inclusive Education and Awareness**
  - Collaboration between medical and educational professionals increases community awareness of cleft lip and palate, promoting better care practices and support for families (Alidadi et al., 2024).

Critical Members of the Management Team

Primary Care Clinician	Coordinate comprehensive care across medical, surgical, and rehabilitative services
Otolaryngologist/Surgeon (ENT)	Perform necessary surgical interventions and manage ongoing ENT-related issues associated with CLAP
Nutritionist	Address and manage nutritional needs and feeding challenges specific to individuals with CLAP
Audiologist	Assess and manage hearing issues (e.g. OME), which are common in CLAP patients
Psychologist	Offer therapeutic support for emotional and psychological challenges related to CLAP
Dentist	Provide routine dental care, assess oral health, educate patients on oral hygiene, and collaborate with orthodontists and prosthodontists with the use of orthodontic devices and prosthetic appliances.
Orthodontist	Straighten and reposition teeth using braces and other devices,

	implement presurgical nasoalveolar molding (NAM; a custom-made appliance gently molds the gum tissues and aligns the cleft segments to enhance their position prior to surgical repair), and perform palatal expansion to improve dental alignment and function in patients with cleft lip and palate.
Prosthodontist	Prosthodontists create artificial teeth and dental appliances, construct obturators to close gaps in the palate, and design speech bulbs to enhance speech production for individuals with cleft lip and palate.
Respiratory Therapist	Manage any respiratory issues that may arise due to CLAP
Speech-Language Pathologist	• Assess and provide therapy for communication challenges, including feeding, speech, and language development. • Advocate to patients and their families, ensuring access to appropriate care and resources. • Counseling by offering emotional support to families. ***Roles of SLPs in Surgical Programs:*** • **Parental education** - Inform about the cleft condition and its impact on communication and feeding. • Inform on the **preventive measures** in surgery: ○ Hot foods and drink are prohibited ○ No sucking through straw due to increased intraoral pressure, which can affect healing. ○ Eat a soft diet only for safe swallowing and minimize trauma to the surgical site. • **Obturator fitting/therapy** ○ Provide guidance on techniques and strategies to prevent the development of compensatory speech patterns and promote effective communication.
Occupational Therapist	Enhance overall quality of life and independence by providing opportunities for social participation, developing self-care and play abilities, and educating families.
Genetic Counselors	Help parents and adult patients understand the chances of having future children with a cleft palate or cleft lip.

Medical Precautions Regarding Speech-Language Therapy

Before	During	After
• Review Client History ○ Assess the client's medical history, including any surgeries related to cleft repair, allergies, and previous interventions. • Ensure a Safe Environment ○ Maintain a clean, hazard-free therapy space that is sanitized	• Explain Procedures ○ Clearly explain each step of the therapy session, ensuring the client and their caregivers understand the purpose, especially in addressing speech or feeding difficulties. • Set Boundaries and Obtain Consent	• Share Results and Progress ○ Communicate therapy results and progress with the client and their significant other. • Provide Post-Therapy Instructions ○ Offer clear post-therapy care instructions and

and free of hazards to avoid infection risks, particularly important for clients with surgical repairs. ● Plan for Emergencies ○ Develop and review a plan for handling medical emergencies or unexpected health issues such as airway obstructions or sudden health issues during the session.	○ Establish clear boundaries and obtain explicit consent for any physical contact or intervention. ● Monitor Comfort and Reactions ○ Continuously assess the client's comfort and physical reactions, especially when working around sensitive areas like the palate or jaw. ● Maintain Hygiene ○ Strictly follow hygiene protocols, including sterilizing equipment to prevent infection, particularly in clients with recent surgeries or oral complications. ● Utilize Appropriate Communication ○ Use clear, accessible language tailored to the client and check in regularly to ensure they understand instructions and feedback.	suggest carry-over techniques for continued practice at home. ● Document and Follow-Up ○ Accurately document session outcomes and any concerns, and schedule follow-up appointments as needed. ● Review Therapy Goals ○ Regularly review and adjust therapy goals based on the client's progress and feedback to ensure that therapy remains relevant and effective.

SUPPORT SYSTEMS

Local

Department of Health (DOH)

- Despite its primary focus on public health, the DOH provides healthcare services and programs that can help people with CLAP.

Philippine Association of Speech-Language Pathologists (PASP)

- Offers services and support related to speech, language, and hearing, including for adults with CLAP

Operation Smile Philippines (OSP)

- Offers complete treatment for people in the Philippines who have CLAP, including preoperative evaluations, surgical procedures, post-operative care, community outreach, family support, and follow-up services. Along with advocating for laws that benefit people in the Philippines who have cleft lip and palate, they also provide training to local medical professionals and work to lower stigma and ensure long-term care.

Smile Train PH

- Provides free cleft lip and palate surgery, comprehensive care, and support to CLAP patients in the Philippines. They partner with local medical professionals and raise awareness about CLAP to improve access to care and support for affected children and families.

International

Smile Train

- Although they specialize in pediatric surgery, they also provide tools and support programs for adult patients and their families.

American Cleft Palate Craniofacial Association (ACPA)

- Provides information, encouragement, and advocacy to people with cleft problems and their families.

Operation Smile

- They identify people in need, offer healthcare, and develop the capacity of various national healthcare systems in collaboration with local partners. Furthermore, Operation Smile International promotes laws and initiatives that help people with cleft lip and palate all around the world.

CRANIOFACIAL ANOMALIES

Definition

- **Craniofacial anomalies** are deformities that affect an individual's head and facial bones and can range from mild to severe (Kummer, 2018).
- **Syndrome** is a pattern of multiple anomalies that are pathogenically related, and therefore have a common known or suspected cause; A recognizable pattern of signs or symptoms that "run together" (Kummer, 2018).
- An **association** is a nonrandom occurrence of a pattern of multiple anomalies in two or more individuals that are not a known syndrome or sequence; A group of malformations that occur together more than would be expected by chance alone (Kummer, 2018).
- In contrast to a syndrome or association, a **sequence** describes a series of anomalies that result from a single initiating event, anomaly, or mechanical factor; A pattern of deformations and malformations which is a consequence of a single malformation (Kummer, 2018).

Etiology

- **Genetic and Chromosomal Disorders**
  - specific gene mutations and atypical chromosomal arrangements
- **Environmental teratogens**
  - substances, cigarette smoking, drugs, viruses, maternal nutrition deficiencies, and pollution
- **Folic acid deficiency**
  - B vitamin that is found in green leafy vegetables, orange juice, fortified breakfast cereals, and enriched grain products.

Prevalence & Incidence

Generally

Locally:

- Surveillance of birth defects will generate data to help in making health policies to prevent and address them, but a surveillance system remains to be established
- Specifically for craniofacial anomalies, there is a lack of data in the Philippine context as this population is not that studied on so no data can be obtained

Internationally:

- Facial malformations are anatomical abnormalities that impact the face and are visible from birth and during pregnancy. They affect 2%–3% of the world's population and have multiple contributing factors (Slah-Ud-Din et al., 2023).

Specifically

Locally	Internationally
• In the Philippines, over 100,000 households are taking care of a family member who has **down syndrome**, and one in every 800 infants is affected.	• An estimated 1 in 1000 to 1 in 1100 live births is the incidence, according to the World Health Organization.14,15 Of these, approximately 95% of patients have **free trisomy 21**, 3% to 4% have an unbalanced

	Robertsonian **translocation** between chromosome 21 and its long arms, and 1% to 2% have **mosaic trisomy 21**. (Chen & Kanekar, 2022) ● **Apert syndrome** is thought to affect 1 in 65,000 people; it is inherited autosomally dominantly with incomplete penetrance and is linked to a mutation in the FGFR2 gene located at 10q26.(Chen & Kanekar, 2022) ● For **Crouzon Syndrome,** the FGFR2 gene mutations on 10q25–10q26 are linked to the autosomal dominant inheritance with variable penetrance; the frequency of this condition is estimated to be 1 in 25,000 live births.(Chen & Kanekar, 2022) ● **Treacher Collins** syndrome, another name for mandibulofacial dysostosis, is thought to affect 1 in 50,000 people. Of those cases, 40% are autosomal dominant and the remaining 60% are sporadic. A de novo mutation at 5q32 is linked to about 60% of cases, and expression varies within a family.(Chen & Kanekar, 2022) ● In roughly 40% to 65% of the cases, the **Pierre Robin sequence** is discovered isolated.(Chen & Kanekar, 2022)

Manifestations of the Disease that the Physician/Allied Health Medical Professional Perceives	**Manifestations the Patient Experiences**
Physical Deformities ● Altered facial and skull shape due to abnormal or premature fusion of skull sutures (e.g., Craniosynostosis, Crouzon Syndrome). ● Asymmetry in facial features (e.g., Hemifacial Microsomia) **Impaired Function** ● Hearing loss ● Trouble with breathing, chewing, swallowing, and speech. **Developmental Issues** ● Impaired cognitive or motor development **Syndrome-Specific Signs** ● Fusion of skull sutures leading to restricted head growth (e.g., Craniosynostosis). ● Abnormally wide-set eyes, beaked nose, and short upper jaw (e.g., Crouzon Syndrome). ● Fusion of fingers or toes, often associated with syndromic craniofacial anomalies (e.g., Apert Syndrome). **Diagnostic Testing** ● MRI, CT scan, X-rays	**Difficulty feeding** ● Unable to suck properly **Ear infections and hearing loss** ● Dysfunction of the eustachian tube that connects the middle ear and the throat **Speech and language delay** ● Muscle function may be decreased, which can lead to a delay in speech or abnormal speech **Dental problems** ● Teeth may not develop normally and orthodontic treatment may be required **Psychological challenges** ● Coping with the disorder ● Social acceptance ● Educational and career opportunities

Structural and Anatomical Changes Related to the Condition

Apert Syndrome
- Syndactyly (webbed fingers/toes), developmental disabilities.

Crouzon Syndrome
- Possible hydrocephalus, agenesis of the corpus callosum.

Down Syndrome
- Almond-shaped eyes with epicanthal folds, macroglossia, short stature, single palm crease, hypotonia, congenital heart defects.

EEC Syndrome
- Ectrodactyly (lobster-claw deformity), ectodermal dysplasia (dry skin, absent sweat glands), lacrimal duct defects.

Moebius Syndrome
- Mask-like facies, limb abnormalities, occasional hearing loss, chest wall defects.

Pierre Robin Sequence
- Glossoptosis (tongue far back), airway/feeding difficulties.

Stickler Syndrome
- Severe myopia, risk for retinal detachment, early-onset osteoarthritis.

Treacher Collins Syndrome
- Lower eyelid colobomas, microtia (small ears), hypoplastic zygomatic arches.

Van der Woude Syndrome
- Bilateral lip pits on lower lip, missing teeth.

Velocardiofacial Syndrome (VCFS)
- Long face, vertical maxillary excess, cardiac anomalies (VSD, ASD), psychosis risk, slender digits.

Possible SLP Areas Affected and Their Characteristics Specific to Each Syndrome

Apert Syndrome	**Articulation and Speech** ● Difficulty making spoken sounds as a result of abnormal facial characteristics and malocclusion. ● **Obligatory Errors** ○ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ● **Compensatory Errors** ○ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** ● Abnormal palate and facial muscles causes swallowing difficulties. **Cognition** ● Difficulty focusing and maintaining attention due to the impact of craniosynostosis on brain development.

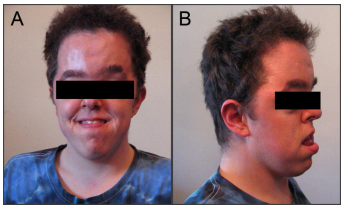
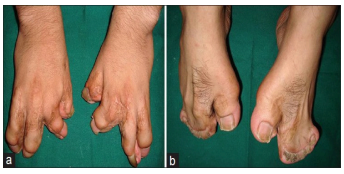
Crouzon Syndrome	**Articulation** ● Difficulty making spoken sounds as a result of abnormal facial characteristics and malocclusion. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** ● Abnormal palate and facial muscles causes swallowing difficulties. **Cognition** ● Problems with short-term and long-term memory due to cognitive impairments and the impact of craniosynostosis on brain development.
Down Syndrome	**Articulation** ● Delayed speech development, difficulty pronouncing complex sounds, and articulation issues. **Language** ● Delayed language acquisition, problems with language comprehension and usage, and pragmatic issues. **Cognition** ● Down syndrome is associated with intellectual disability, which can affect language development and comprehension. ● They often have varying levels of severity. ● Difficulties in reading, writing, and math because to developmental delays and cognitive disabilities. ● Difficulties with daily living skills, such as self-care and communication, due to cognitive impairments and motor delays. **Swallowing** ● Delayed oral-motor development and weakening of the muscles increase the risk of **aspiration**.
Ectrodactyly-Ectodermal Dysplasia Cleft Syndrome	**Articulation and Speech** ● Speech difficulties brought on by cleft palate and abnormal face characteristics. ● **Obligatory Errors** ○ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ● **Compensatory Errors** ○ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency. **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** ● Abnormal palate and facial muscles causes swallowing difficulties.

Moebius Syndrome	**Pragmatics and Speech** ● Inability to move facial muscles, affecting facial expressions and speech. **Articulation** ● Difficulty producing bilabial and labiodental sounds due to **paralysis** of the lips. **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Cognition** ● Due to the syndrome's related facial paralysis, problems with tasks requiring spatial awareness and visual perception may arise. ● The facial paralysis and possible muscle weakness, difficulties with fine motor abilities, such as handwriting and using utensils, may arise.
Pierre Robin Sequence	**Articulation** ● Difficulty producing speech sounds due to cleft palate and abnormal tongue position. ● **Obligatory Errors** ○ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ● **Compensatory Errors** ○ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency. **Swallowing** ● Atypical tongue posture and airway blockage increase the risk of aspiration.
Stickler Syndrome	**Hearing** ● Conductive or mixed hearing loss due to middle ear abnormalities. ● The hearing loss may be sensorineural, indicating that it arises from alterations in the inner ear , or conductive, meaning that it is caused by abnormalities of the middle ear. **Articulation** ● Difficulty producing speech sounds due to hearing loss and potential cleft palate. **Language** ● Delayed language acquisition, problems with language comprehension and usage, and pragmatic issues.

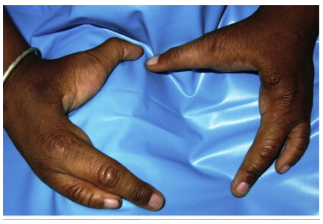
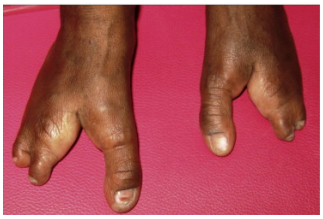
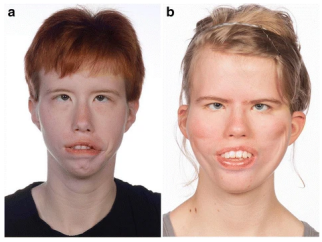
Treacher Collins Syndrome	**Articulation and Speech** ● Difficulty producing speech sounds due to abnormal facial features and micrognathia. ● **Obligatory Errors** ○ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ● **Compensatory Errors** ○ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency. **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** ● Abnormal palate and facial muscles causes swallowing difficulties.
Van Der Woude Syndrome	**Articulation and Speech** ● Difficulty producing speech sounds due to abnormal facial features and micrognathia. ● **Obligatory Errors** ○ Exist due to structural abnormalities that result in velopharyngeal insufficiency and oral structural deviations (e.g., oronasal fistulas, dental deviations, or malocclusions) ● **Compensatory Errors** ○ Actively learned maladaptive articulations that develop in response to abnormal structures found in VPD. **Resonance** ● Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency. **Voice** ● Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** ● Abnormal palate and facial muscles causes swallowing difficulties.

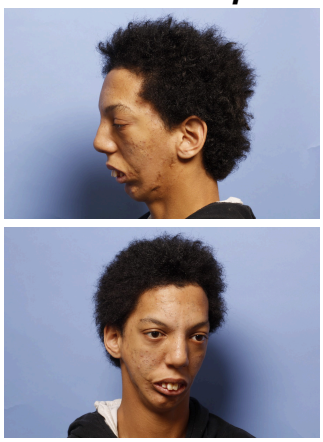
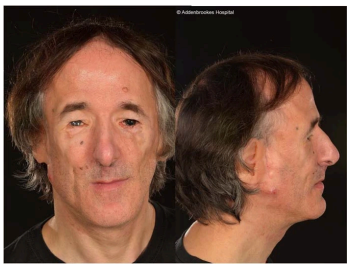
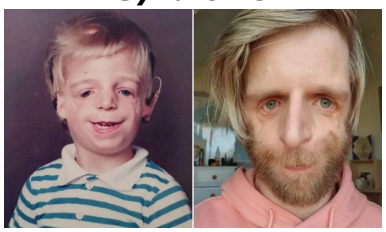
Velocardiofacial Syndrome	**Articulation** • Difficulty producing speech sounds due to abnormal facial features and micrognathia. **Resonance** • Hypernasality or Mixed Resonance caused by velopharyngeal insufficiency. **Voice** • Abnormal anatomy of the vocal folds causing hoarseness or breathiness. **Swallowing** • Abnormal palate and facial muscles causes swallowing difficulties. **Cognition** • Brain abnormalities, especially in areas related to language development and cognitive function, can result from the genetic deletion linked to VCF syndrome. • Challenges with reading, writing, and mathematics due to cognitive impairments and potential hearing loss. **Language** • Delayed language acquisition, problems with language comprehension and usage, and pragmatic issues.

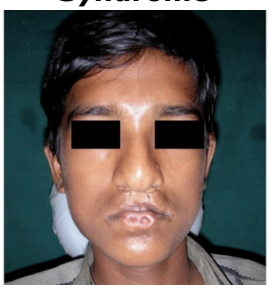
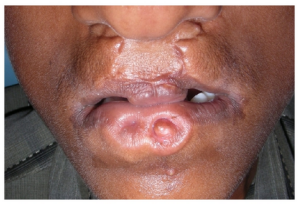
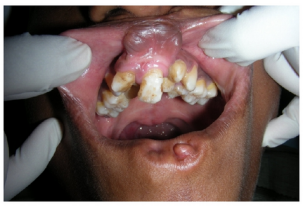
Types of Disorders

TYPE	ETIOLOGY & PHENOTYPIC FEATURES
Apert Syndome (aka: *Acrocephalosyndactyly Type I***)**   	**Etiology** • Mutation of a gene on the long arm of chromosome 10 (10q25.3-q26). FGFR2 (10q26) Causes premature closure of the coronal sutures so that the skull grows laterally, but not anteriorly **Phenotypic Features** • **Prominent forehead with a flat occiput** (back of the head/skull) • **Syndactyly or webbing of fingers and toes** • **Shallow Pharynx** • Exophthalmos (protrusion of the eyes) • Hypertelorism (abnormally large distance between the eyes) • Antimongoloid slant (opening between the eye lids and slanting up or down) • Strabismus (crossed eyes) • Midface Hypoplasia/Retrusion (underdevelopment of the middle part of the face), • Class III malocclusion • Hearing loss • Low set ears • Small Maxilla

Crouzon Syndrome  	**Etiology** • Mutation along the long arm of chromosome 10 (10q25.3-q26). FGFR2 (10q26) Causes premature closure of the coronal sutures so that the skull grows laterally, but not anteriorly **Phenotypic Features** • **Flattened head and nose bridge** • Prominent forehead with a flat occiput (back of the head/skull) • **Exophthalmos** (protrusion of the eyes) • **Hypertelorism** (abnormally large distance between the eyes) • Antimongoloid slant (opening between the eye lids and slanting up or down) • Strabismus (crossed eyes) • Midface Hypoplasia/Retrusion (underdevelopment of the middle part of the face), • Class III malocclusion • Hearing loss • Low set ears • Small Maxilla • Shorter nasopharyngeal space • Can have hydrocephalus or agenesis of the corpus callosum
Down Syndrome (aka: *Trisomy 21***)*** 	**Etiology** • Extra copy of chromosome 21 **Phenotypic Features** • **Almond-Shaped Eyes** - Upward slanting of the palpebral fissures with epicanthal folds on the inner corner of the eyes • **Macroglossia** and/or protruding tongue • **Hypotonia** • **Short neck** • **Mild to moderate intellectual disability** • Micrognathia (small mandible) • Broad face • Low set ears • Short limbs resulting in short stature • A crease across one or both palms • Congenital heart defects • Gastroesophageal reflux • Breathing issues and obstructive sleep apnea

Ectrodactyly-Ectodermal Dysplasia-Cleft Syndrome (*EEC Syndrome****)*** 	**Etiology** • Genetic defect on the long arm of chromosome 7 (7q11.2-q21.3) **Phenotypic Features** • **Cleft Lip and/or Palate** • **"Lobster-claw"** deformity or **Ectrodactyly** (deficiency or absence of one or more central digits of the hand or foot)   • **Ectodermal dysplasia** (dry skin and mucosa, dry, sparse hair) • **Photophobia** (eyes' sensitivity to light) → **Seizure** • **Ossicular anomalies → Conductive HL** • Partial anodontia or microdontia • Maxillary and malar hypoplasia • Defects of the lacrimal duct system
Moebius Syndrome 	**Etiology** • Absence or under development of abducens nerve (CN VI) and facial nerve (CN VII) **Phenotypic Features** • **Abnormalities in movement** of cheeks, lips, and eyes, resulting in a **flat**, "**mask-type**" **facies** (appearance or facial expression of an individual that is typical of a particular disease or condition) • **Strabismus** (crossed eyes) • Occasional hearing loss if cranial VIII is affected • Occasional breathing and/or in swallowing problems • Limb abnormalities, including clubbed feet, missing digits • Chest wall abnormalities.

Pierre Robin Sequence 	**Etiology** • Micrognathia that may be due to crowding in utero, or may be genetic as part of a syndrome (e.g., Stickler's syndrome or velocardiofacial syndrome). • Micrognathia interfere with the downward progression of the tongue and the closure of the velum. **Phenotypic Features** • A **wide bell-shaped cleft palate** • **Micrognathia** • **Glossoptosis** (tongue far back in their mouth) • Airway and feeding issues, particularly at birth.
Stickler Syndrome 	**Etiology** • Mutations on the short arm of chromosome 6 (6p21) • Most common cause of Pierre Robin sequence **Phenotypic Features** • **Most common syndromic causes of cleft palate** • Micrognathia in infancy • **Flat facial profile** • **Epicanthal folds** (a skin fold of the upper eyelid covering the inner corner of the eye) • **Small nose** • Flat nasal bridge • Midface hypoplasia • **High myopia** (severe nearsightedness, usually progressive) • **High risk for retinal detachments** • **High frequency hearing loss** • **Risk for early onset osteoarthritis**
Treacher Collins Syndrome 	**Etiology** • Mutations on the long arm of chromosome 5 (5q32-q33.3) • Cheekbones and jawbones do not develop well. **Phenotypic Features** • **Downward slanting** of the palpebral fissures (the area between the open eyelids) • **Colobomas** (part of the tissue that makes up the eye is missing) of the lower eyelids • **Microtia** or middle ear anomalies • **Hypoplastic zygomatic arches and malar hypoplasia** • **Macrostomia or microstomia** • Micrognathia • Glossoptosis

Van der Woude Syndrome 	**Etiology** • Mutation of interferon regulatory growth factor 6 (IRF6) gene on the long arm of chromosome (11q.32-41). **Phenotypic Features** • **Cleft lip and/or palate** • **Bilateral lip pits** on the lower lip • Missing teeth  

Velocardiofacial Syndrome (*VCFS;* aka: *DiGeorge syndrome or 22q11.2 Syndrome****)***
 
 
	Etiology
	• Deletion of part of chromosome 22
	• Second most common genetic syndrome next to Down Syndrome
	
	Phenotypic Features
	• **Velo: Velopharyngeal dysfunction causing hypernasality**, usually secondary to an occult submucous cleft or pharyngeal hypotonia.
	• **Cardio: Minor cardiac and vascular anomalies** including;
	  ○ Ventriculoseptal deviation (VSD)
	  ○ Atrial septal defect (ASD)
	  ○ Patent ductus arteriosis (PDA)
	  ○ Pulmonary stenosis
	  ○ Tetralogy of Fallot
	  ○ Right-sided aortic arch
	  ○ Medially displaced internal carotid arteries
	  ○ Tortuosity of the retinal arteries.
	  ○ Parents often report a history of heart murmur at birth.
	• **Facial:**
	  ○ **Microcephaly**
	  ○ **Long face with vertical maxillary excess**
	  ○ Micrognathia (small jaw) or retruded mandible, often with a Class II malocclusion
	  ○ **Nasal anomalies** including wide nasal bridge, narrow alar base and bulbous nasal tip
	  ○ **Narrow palpebral fissures** (slit-like eyes)
	  ○ Malar flatness
	  ○ Thin upper lip
	  ○ Minor auricular anomalies
	  ○ Abundant scalp hair
	• **Other:**
	  ○ **Long slender digits**
	  ○ **Hyperextensibility of the joints**
	  ○ Short stature, usually below the 10 percentile in weight and height
	  ○ Pierre Robin sequence (cleft palate, micrognathia, glossoptosis with airway obstruction)
	  ○ Umbilical and inguinal hernias

	○ **Laryngeal web** ○ Gross and fine motor delays ○ Various brain anomalies ○ Social disinhibition ○ Risk of onset of psychosis in adolescence ○ Learning disabilities and concrete thinking ○ **Mild to moderate intellectual disability**

Progression & Outcome if Left Treated and/or Untreated

Craniofacial anomalies are generally *non-progressive*. However, if left untreated, it can lead to several challenges related to feeding, speech, hearing, dentition, and socioemotional difficulties.

Medical/Surgical Management

Healthcare Resources

- **Craniofacial Surgery**
  - Procedures to correct developmental or congenital conditions affecting the face, jaw, and mouth
  - For traumatic injuries, birth defects, and quality-of-life reconstruction
  - Involves Plastic Surgery
- **Minimally Invasive Surgery**
  - Small incisions, reducing pain, complications, and recovery time
  - Surgeons use special tools and techniques to perform many procedures through these small openings
- **Rehabilitation Care**
  - Physical Therapy - Target motor impairments due to abnormal head shapes or limited mobility
  - Occupational Therapy - Provides comprehensive care by focusing on functional and cognitive development.
  - Speech-Language Therapy - Address speech sound disorders, articulation difficulties, and swallowing problems.
  - Aural Habilitation & Rehabilitation - Provide hearing aids, auditory training, and counseling to help individuals with hearing loss communicate effectively.

SLP Therapy

Areas	Strategies	Assessment & Materials	Approaches
Speech To enhance articulation, phonology, clarity and intelligibility.	• Target specific error sounds, especially those affected by velopharyngeal dysfunction • Use visual feedback (e.g., mirrors, video recordings) • Implement oral motor exercises to improve muscle strength and coordination • Identify articulation	• Oral Peripheral Mechanism Test • Bauman-Waengler • Speech-Motor Screening • Frenchy Dysarthria Assessment • Goldman Fristoe Test of Articulation 3rd Edition (GFTA-3) • Passage and Conversation • Any articulation screener with sounds	• Traditional articulation therapy • Phonological process approach • Cycles approach for multiple sound errors

	errors characterized as *obligatory errors* (due to structural differences) or as *compensatory* (learned) errors	in different placements	

Resonance
Improve overall voice quality and speech naturalness.
	• Focus on reducing hypernasality or hyponasality
	• Teach proper oral-nasal balance
		• Speech Screening
		• Assessment for velopharyngeal dysfunction
		  ○ Oral inspection
		  ○ Acoustic nasometry
		  ○ Mirror tests
		  ○ Nasoendoscopy with a photo or video recording.
		  ○ Videofluoroscopy for velum function.
		  ○ MRI
		• Comprehensive Assessment
			• Continuous positive airway pressure (CPAP) therapy
			• Biofeedback techniques (e.g., nasometry)
			• Resonance therapy exercises

Voice To address issues such as hoarseness or breathiness and achieve a clearer, more efficient voice production.	• Address compensatory articulation patterns • Improve vocal quality and pitch control • Assess for voice quality and signs of vocal fold pathology (e.g., hoarseness, breathiness).	• Auditory-perceptual assessment • Instrumental assessment • Assessment of voice handicap • Referral for ear, nose, and throat assessment	• Vocal Range and Flexibility Exercises • Voice Therapy Programs • Vocal Hygiene • Breath Support Training
Feeding and Swallowing	• Improve oral motor control • Address any structural issues affecting swallowing	• Eating and drinking abilities (e.g. oral containment, sucking, mastication) • Secretion management • Oral hygiene practices • Sensory status • Acceptance of food • Diet diversity • Oral medication management • Caregiver behaviors during feeding • Psychosocial impact on family and child dynamics	• Obturator Fitting • Oral Motor Exercises • Swallowing Maneuvers

Language and Pragmatics	• Address any language delays or disorders resulting from hearing loss or social factors • Improve social communication skills by increasing interactions for more opportunities for language growth	• Comprehensive Assessment of Spoken Language 2nd Edition (CASL-2) • Clinical Evaluation of Language Fundamentals Fifth Edition (CELF-5) • Western Aphasia Battery (WAB) Test • Structured and Unstructured Conversations	• Cognitive-linguistic Therapy • Social Skills Training • Script Training or Narrative Interventions • Pragmatic Language Intervention • Augmentative and Alternative Communication (AAC) ○ If speech is severely impaired, AAC strategies may be used to support communication
Hearing	• Assess detection, discrimination, identification, and comprehension	• Ling- 6 & Ling-10 Test • Early Listening Function (ELF) • Glendonald Auditory Screening Procedure (GASP)	• Listening and Spoken Language Therapy • Auditory-Oral or Auditory-Based Speech Therapy • Total Communication Approach
Cognition	• Target specific cognitive domains (e.g., attention, memory, executive function) • Implement compensatory strategies • Provide environmental modifications	• Western Aphasia Battery (WAB) Test • Montreal Cognitive Assessment (MoCA) • Mini Mental State Examination (MMSE)	• Cognitive Stimulation Therapy • Attention Process Training • Memory Strategy Training

<u>Educational Management</u>

Patient and Family Education

- Provide information about the condition, care management, and psychosocial support strategies to address psychological challenges.

Targeted Interventions

- Raising awareness of nutrition, dental issues, and speech abnormalities helps alleviate mental stress and improve disease management. Teach effective communication strategies and social skills training to enhance interactions while considering cognitive aspects. Develop individualized

education plans (IEPs) that accommodate cognitive challenges, offering tailored learning strategies and support.

Preventive Education

- Increased awareness prevents further complications, improving quality of life for patients and families.

Psychological Support

- Provide access to counseling and mental health resources to address psychological difficulties, such as anxiety or low self-esteem.

Transition Planning

- Offer guidance for transitions between educational levels and into adulthood, focusing on social integration, independence, and addressing potential psychological impacts.

Critical Members of the Management Team

Primary Care Physician	Coordinate comprehensive care across medical, surgical, and rehabilitative services.
Geneticist	Identifies and manages the underlying genetic causes of craniofacial anomalies, offering guidance on hereditary risks and implications.
Brain Surgeon	Addresses any neurological complications or structural brain abnormalities associated with craniofacial anomalies.
Plastic Surgeon	Performs reconstructive surgeries to correct facial deformities, improve function, and enhance appearance
Anesthesiologist	Manages anesthesia during surgical procedures.
Psychologist	Provides mental health support to patients and families, helping them cope with the emotional and psychological impacts of craniofacial anomalies.
Orthodontist	Manages dental and jaw alignment issues as well as provides devices to facilitate repositioning of teeth.
Physical Therapist	Aids in improving physical function and mobility, particularly in cases where craniofacial anomalies affect muscle development or posture.
Speech-Language Therapist	Assesses and treats speech and language difficulties resulting from structural anomalies in the oral cavity and facial region.
Respiratory Therapist	Addresses breathing difficulties that may arise from airway obstructions or other structural issues.
Occupational Therapist	Improves daily living, promoting independence and emotional well-being of individuals with craniofacial anomalies.
Special Education Teacher	Provides individualized learning plans, social skills support, and integrates communication and emotional regulation strategies in the classroom.
Nutritionist	Address and manage nutritional needs and feeding challenges.

Audiologist	Evaluates and treats hearing impairments that are common in patients with craniofacial anomalies.

Medical Precautions Regarding Speech-Language Therapy

Before	During	After
Review Client History ● Assess the client's medical history, including surgeries related to craniofacial anomalies, allergies, syndromic conditions, previous interventions, and any psychological, emotional, or cognitive challenges. **Ensure a Safe Environment** ● Maintain a clean, hazard-free therapy space that is sanitized and free of hazards to avoid infection risks, particularly important for clients with surgical repairs. **Plan for Emergencies** ● Develop and review a plan for handling medical emergencies or unexpected health issues such as airway obstructions or sudden health issues during the session.	**Explain Procedures** ● Clearly explain each step of the therapy session, ensuring the client and their caregivers understand the purpose, especially in addressing speech or feeding difficulties. **Set Boundaries and Obtain Consent** ● Establish clear boundaries and obtain explicit consent for any physical contact or intervention. **Monitor Comfort and Reactions** ● Continuously assess the client's comfort and physical reactions, especially when working around sensitive areas like the palate or jaw. **Maintain Hygiene** ● Strictly follow hygiene protocols, including sterilizing equipment to prevent infection, particularly in clients with recent surgeries or oral complications. **Utilize Appropriate Communication** ● Use clear, accessible language tailored to the client and check in regularly to ensure they understand instructions and feedback.	**Share Results and Progress** ● Communicate therapy results and progress with the client and their significant other. **Provide Post-Therapy Instructions** ● Offer clear post-therapy care instructions and suggest carry-over techniques for continued practice at home. **Document and Follow-Up** ● Accurately document session outcomes and any concerns, and schedule follow-up appointments as needed. **Review Therapy Goals** Regularly review and adjust therapy goals based on the client's progress and feedback to ensure that therapy remains relevant and effective.

SUPPORT SYSTEMS

Local

- **Noordhoff Craniofacial Foundation Philippines, Inc. (NCFPI)**
  - NCFPI provides comprehensive care, including surgery, speech therapy, and psychosocial support, despite being largely focused on children. Adults may be able to utilize some of their services.
- **Department of Health (DOH)**
  - The DOH offers healthcare services and programs that may also benefit individuals with craniofacial anomalies.
- **Philippine Association of Speech-Language Pathologist (PASP)**
  - Provides speech, language, and hearing services and support, including for persons with craniofacial abnormalities.

International

- **International Craniofacial Institute (ICI)**
  - This organization provides resources, support, and education for patients, families, and healthcare providers involved in craniofacial care.
- **The World Health Organization (WHO)**
  - The WHO adds to a larger global network of assistance for people with craniofacial anomalies through setting standards, gathering data, and fighting for the rights of those with disabilities

REFERENCES

About Cleft Lip & Cleft Palate (n.d.). https://operationsmile.org.ph/about-cleft-lip-cleft-palate/

About us. (n.d.). Operation Smile. https://www.operationsmile.org/about-us

Alcausin, M. M. L., Sur, A. L. D., Tumulak, M. J. R., & Padilla, C. D. (2017). AB122. Occurrence of birth defects at the Philippine General Hospital, 2013–2014: a prospective study. Annals of Translational Medicine, 5(S2), AB122. https://doi.org/10.21037/atm.2017.s122

Alirhayim, R. N., & Mohammed, A. J. (2022). Feeding obturator: Indications, types, and benefits. A Literature Review. Azerbaijan Medical Association Journal, 62(06), 2155-2162. https://www.researchgate.net/publication/377358963_Feeding_Obturator_Indications_Types_and_Benefits_A_Literature_Review

American Speech-Language-Hearing Association. (n.d.). Cleft lip and palate. Retrieved October 5, 2024, from https://www.asha.org/practice-portal/clinical-topics/cleft-lip-and-palate/#collapse_6

American Speech-Language-Hearing Association. (n.d.). Comprehensive assessment for cleft lip and palate and resonance. https://www.asha.org/practice-portal/resources/comprehensive-assessment-for-cleft-lip-and-palate-and-resonance/

Benacerraf, B. R., Bromley, B., & Jelin, A. C. (2019). Paramedian orofacial cleft. American Journal of Obsterics & Gynecology, 221(5), B8-B12. https://doi.org/10.1016/j.ajog.2019.08.050

Bernthal, J. E., Bankson, N. W., & Flipsen, P. J. (2017). Articulation and phonological disorders: Speech sound disorders in children. Pearson.

Boston Children's Hospital. (n.d.). Craniofacial anomalies. Retrieved August 18, 2024 from, https://www.childrenshospital.org/conditions/craniofacial-anomalies#:~:text=Craniofacial%20anomalies%20are%20deformities%20that,the%20lip%20and%20the%20palate

Chen, J., & Kanekar, S. (2022). Imaging of congenital craniofacial anomalies and syndromes. Clinics in Perinatology, 49(3), 771–790. https://doi.org/10.1016/j.clp.2022.04.005

Cleft Lip and Cleft Palate. (n.d.). Johns Hopkins Medicine. Retrieved August 16, 2024, from https://www.hopkinsmedicine.org/health/conditions-and-diseases/cleft-lip-and-palate

Cleft lip and palate related hearing problems. (n.d.). https://www.salusuhealth.com/news/2019/07/cleft-lip-and-palate-related-hearing-problems.html

Cleveland Clinic. (2022). Cleft lip & cleft palate surgery. https://my.clevelandclinic.org/health/treatments/23394-cleft-lip-cleft-palate-surgery

Cleveland Clinic. (2023). Minimally invasive surgery. https://my.clevelandclinic.org/health/procedures/minimally-invasive-surgery

Conrady CD, Patel BC, Sharma S. Apert Syndrome. (2023 Jun 1). In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2024 Jan. https://www.ncbi.nlm.nih.gov/books/NBK518993/

Free, sustainable, local cleft care on 5 continents | Smile Train. (n.d.). https://www.smiletrain.org/our-approach

Hardin-Jones, M., & Chapman, K. L. (2011). Cognitive and language issues associated with cleft lip and palate. Seminars in Speech and Language, 32(2), 127-140. http://dx.doi.org/10.1055/s-0031-1277715.

Healing Smile Foundation. (n.d.). Healing Smile Foundation. Retrieved October 5, 2024, from http://healingsmilefoundation.org.in/HSF/functionalanatomy

International Craniofacial Institute in Dallas, Plano TX. (2022, June 13). Craniofacial. https://www.craniofacial.net/

John Hopkins Medicine. (2024). Craniofacial abnormalities. Retrieved August 19, 2024 from, https://www.hopkinsmedicine.org/health/conditions-and-diseases/craniofacial-abnormalities

Johns Hopkins Medicine. (n.d.). Craniosynostosis. Johns Hopkins Medicine. Retrieved August 17, 2024, from https://www.hopkinsmedicine.org/health/conditions-and-diseases/craniosynostosis

Kummer, A. W. (2014). Cleft palate and craniofacial anomalies: Effects on speech and resonance (third ed.). Cengage Learning.

Kummer, A. W. (2018). Cleft Palate and Craniofacial Conditions: A Comprehensive Guide to Clinical Management. ResearchGate. https://www.researchgate.net/publication/325854806_Cleft_Palate_and_Craniofacial_Conditions_A_Comprehensive_Guide_to_Clinical_Management

Kutbi, H. A. (2014). The role of obesity, diabetes, and hypertension in cleft lip and cleft palate birth defects. All Graduate Theses and Dissertations, Spring 1920 to Summer 2023. 3081. https://digitalcommons.usu.edu/etd/3081

Mayo Clinic. (2024, January 13). DiGeorge syndrome (22q11.2 deletion syndrome). https://www.mayoclinic.org/diseases-conditions/digeorge-syndrome/symptoms-causes/syc-20353543

McGraw Hill. (n.d.). Chapter 17. Genetics. Access Pediatrics Retrieved September 17, 2024, from https://accesspediatrics.mhmedical.com/content.aspx?bookid=443&sectionid=42148736#:~:text=Syndrome%2C%20Association%2C%20and%20Sequence&text=Sequence%3A%20A%20pattern%20of%20deformations,be%20expected%20by%20chance%20alone.

Murthy, J. (2009). Management of cleft lip and palate in adults. Indian Journal of Plastic Surgery, 42, S116-S122. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2825062/#CIT15

Nagarajan, R., Savitha, V., & Subramaniyan, B. (2009). Communication disorders in individuals with cleft lip and palate: An overview. NCBI. Retrieved October 5, 2024, from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2825064/

National Library of Medicine. (n.d.). Orbital cleft. Retrieved August 18, 2024 from, https://www.ncbi.nlm.nih.gov/medgen/347491

NCFPI | Cleft and Craniofacial Care | Free cleft lip and palate surgeries. (2023, September 13). Noordhoff Craniofacial Foundation Philippines, Inc. (NCFPI). https://www.ncfphil.org/

Phalke, N., & Goldman, J. (2023, July 6). Cleft Palate - StatPearls. NCBI. Retrieved August 16, 2024, from https://www.ncbi.nlm.nih.gov/books/NBK563128/

Professional, C. C. M. (2024, August 15). Speech-Language pathologist. Cleveland Clinic. https://my.clevelandclinic.org/health/articles/24602-speech-language-pathologist

Putri, F. A., Pattamatta, M., Anita, S. E. S., & Maulina, T. (2024). The global occurrences of cleft lip and palate in pediatric patients and their association with demographic factors: A narrative review. Children, 11(3), 322. https://doi.org/10.3390/children11030322

The Royal Children's Hospital Melbourne. (2015). Cleft lip repair: post-operative information. https://www.rch.org.au/uploadedFiles/Main/Content/plastic/Cleft%20lip%20repair%20post operative%20information.pdf

The Royal Children's Hospital Melbourne. (2015). Cleft palate repair: post-operative information. https://www.rch.org.au/uploadedFiles/Main/Content/plastic/Cleft%20palate%20repair%20 postoperative%20information.pdf

The University of Iowa. (2012, February 10). Cleft palate surgical procedures. Retrieved September 17, 2024, from https://uihc.org/childrens/health-topics/cleft-palate-surgical-procedures#:~:text=Primary %20Cleft%20Lip%20Repair%20(cheiloplasty,Fistula%20Repair

TheraPlatform. (2024). Speech therapy assessments. https://www.theraplatform.com/blog/773/speech-therapy-assessments

Tiongco, G. G. (2017, March 27). Cleft lip and palate. St. Luke's Medical Center. https://www.stlukes.com.ph/health-library/health-articles/cleft-lip-and-palate#:~:text=Cle ft%20lip%20and%20palate%20is,(roof%20of%20the%20mouth)

U.S. Centers for Disease Control and Prevention. (2024, May 15). Cleft lip/cleft palate. https://www.cdc.gov/birth-defects/about/cleft-lip-cleft-palate.html#:~:text=Key%20points ,mouth%20don't%20form%20properly.

University of California San Francisco. (n.d.). Cleft Lip and Palate | Conditions. UCSF Health. Retrieved August 16, 2024, from https://www.ucsfhealth.org/conditions/cleft-lip-palate

University of Pittsburgh Schools of the Health Sciences. (n.d.). Craniofacial Malformation Surgery, Symptoms and Treatment. UPMC. Retrieved August 17, 2024, from https://www.upmc.com/services/neurosurgery/brain/conditions/additional-conditions/crani ofacial-malformation#symptoms

Vanderbilt University Medical Center. (2024). Adult cleft and craniofacial program: Academic studies and treatment solutions. Retrieved August 19, 2024 from, https://www.vumc.org/plasticsurgdept/adult-cleft-and-craniofacial-program

Walker, N. (2023). Cleft Lip - StatPearls. NCBI. Retrieved August 16, 2024, from https://www.ncbi.nlm.nih.gov/books/NBK482262/

Yale Medicine. (2024). Craniofacial surgery. https://www.yalemedicine.org/conditions/craniofacial-surgery

Young, A., & Spinner, A. (2023, January 12). Velopharyngeal insufficiency. StatPearls - NCBI Bookshelf. https://www.ncbi.nlm.nih.gov/books/NBK563149/#:~:text=When%20the%20soft%20palate%20and,oral%20cavities%20leads%20to%20hypernasal/