Unit 7: CLEFT AND CRANIOFACIAL ANOMALIES

MEMBERS:

- Camus
- Castillo
- Galo
- Pascual

CLEFT

DEFINITION

- A cleft is an abnormal opening or fissure in an anatomical structure that is normally closed. Clefting is caused by a failure of the palate to fuse during fetal development at 7-12 weeks gestation. This congenital condition manifests as cleft palate and cleft lip, where the normal structure of the hard and soft palates and the lips is interrupted by a gap, dividing the roof of the mouth and/or the upper lip sagittally into separated left and right portions.

Developmental Stage	Time Frame	Description
Formation of Neural Crest	Early gestation	Critical for facial development; precursor to facial structures including the lips and palate
Lip and Alveolus Formation	6 to 7 weeks gestation	Development begins at incisive foramen; closure of lip and alveolar ridge occurs
Tongue Position	7 to 8 weeks gestation	Tongue drops down as mandible moves forward, bringing it into position for palate formation
Palatal Shelf Position	8 to 9 weeks gestation	Palatal shelves are vertical and positioned on either side of the tongue
Palatal Fusion	~7 to 8 weeks gestation	Shelves move to horizontal position and begin to fuse, first with premaxilla and then with each other
Closure of Hard Palate	~9 weeks gestation	Completion of hard palate formation; velum also closes
Completion of Palate	~12 weeks	Finalization of palate closure

ETIOLOGY

There are several causes that lead to a failure of the regular median fusion of the embryo's oral-facial structures between the seventh and twelfth weeks of gestation. Clefting does not have a single cause; instead, it is a clinical outcome of various possible diseases (Shprintzen, 1995):

- Mechanical Interference
  - Crowding in utero
- Exogenous Factors
  - Environmental teratogens
  - Viruses
  - Maternal nutrition
  - Maternal obesity
- Endogenous Factors
  - Chromosomal disorders
  - Genetic disorders
- Multifactorial inheritance
  - Both endogenous and exogenous factors

PREVALENCE & INCIDENCE

Locally	*Internationally*
● St. Luke's Medical Center (2017): 1 out of 1,000 Filipino live births, a cleft lip or palate patient is born. ● Muntz (2013): 1 in 750 people, with only 16% anticipated to be repaired under current conditions ● PubMed (1997): Clefts associated with multiple anomalies was 21% at birth	● United States (2023): 1 in every 1,600 babies is born with CLP;1 in every 2,800 babies is born with CL without CP; 1 in every 1,700 babies is born with CP. ● Talarova (2022): Orofacial cleft is approximately 1 in every 500-550 births. ● Minnesota Department of Health: About 20% have CL only, 30% have CP only, and 50% of children with a CL will also have a CP. About 400 syndromes have cleft lip and palate as part of the affected systems. About 13% of babies with cleft lip and palate have other birth defects.
Fortunately, facial clefts are very rare. The exact incidence of facial clefts is unknown, and estimates vary greatly because of the rarity of their occurrence and the lack of standard methods of data collection.	

SIGNS, SYMPTOMS, PATHOMECHANICS

Manifestations that the **Physician/Allied Health Professional** Perceive	● Crooked, poorly shaped or missing teeth ● Misalignment of teeth and jaw ● Deformities of the upper jaw (maxilla) ● Speech problems ● Unrepaired oronasal fistulae, which is a hole between the mouth and nose cavity. ● Alveolar clefts, which are defects in the bone that supports the teeth.
Manifestations that the **Parents/Significant Others** Perceive	● Feeding difficulties ● Infants may be unable to suck properly. ● Ear infections due to a dysfunction of the eustachian tube that connects the middle ear and the throat that could then lead to hearing loss. ● Speech and language delay ● Dental problems as a result of the abnormalities.
Manifestations that the **Patient** Experiences	● Immediate concern after birth is feeding. ● While most babies with cleft lip can breast-feed, a cleft palate may make sucking difficult. ● Babies are especially at risk of developing middle ear fluid and hearing loss. ● If the cleft extends through the upper gum, tooth development may be affected. ● Speech difficulties ● Speech may sound too nasal. ● Facing social, emotional and behavioral problems due to differences in appearance and the stress of intensive treatment
Pathomechanics and Pathophysiology	The pathophysiology of clefting involves disruptions in the normal processes of embryonic development. During the first trimester of pregnancy, the tissues of the face and mouth region undergo complex growth and fusion. Any disturbance in this process can lead to cleft lip, cleft palate, or both. Research suggests that genetic mutations, maternal smoking, alcohol consumption, certain medications, and nutritional deficiencies can increase the risk of clefting. Additionally, there may be multifactorial inheritance patterns, meaning that both genetic predisposition and environmental factors play a role.

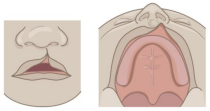
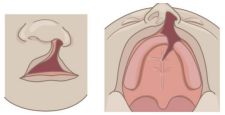
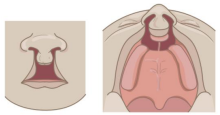
	The pathomechanics of clefting involve the physical consequences of the malformation. In cleft lip, the separation typically occurs along the upper lip, extending from the lip into the nostril. In cleft palate, the gap may vary in size and location along the roof of the mouth, affecting the hard palate, soft palate, or both. These structural abnormalities can impact feeding, speech development, dental health, and facial aesthetics. They can also contribute to middle ear problems and hearing loss due to dysfunction of the Eustachian tube.
Structural & Anatomical Changes	● Cleft lip involves a separation or gap in the upper lip, which can vary in severity from a small notch to a complete division extending into the nostril. ● Cleft palate results from the incomplete fusion of the palatal shelves during embryonic development. The failure of fusion leads to a gap in the palate, affecting functions such as feeding, speech, and the closure of the nasal cavity during swallowing. ● Combined cleft lip and palate involve both a gap in the upper lip and an opening in the palate, which can vary in severity and extent.

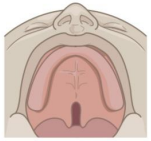
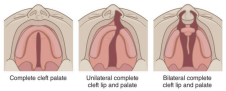
POSSIBLE SPEECH-LANGUAGE PROBLEMS ASSOCIATED WITH CLEFT

Speech-Language Areas	Speech-Language Problems
Hearing	● This is important if the child has frequent ear infections or has extra fluid in the ears (this happens a lot to people who have a cleft palate). An audiologist may monitor the hearing status of a person with cleft lip and palate, especially from birth to age 6.
Speech	● Cleft lip may not create any speech problems, but a person with cleft palate may need help learning to speak more clearly. ● Overall delay of speech sound acquisition ● Cleft lip and palate can impact the ability to produce specific speech sounds accurately due to structural differences in the oral cavity. This may result in distorted speech sounds.

Feeding and Swallowing	● Babies with a cleft lip probably won't have trouble feeding, but babies with a cleft palate might. Babies with a cleft palate may have trouble breastfeeding, and they may need special bottles or nipples if they bottle-feed. SLPs can help you find the best bottle-and-nipple combination.
Resonance	● Individuals with cleft palate may experience hypernasality or nasal air emission due to the opening between the oral and nasal cavities, affecting the quality of their speech. ● characterized by a pronounced nasality of all the vowel sounds, an inability to pronounce the dentals t and d, the labials p and b, the palatals k, hard g and hard c and an incorrect pronunciation of the labiodentals f, v, l, r, and the sibilants s and z.
Language	● Cleft lip and palate can also impact language development, including vocabulary acquisition, grammar, and understanding of language concepts. ● There have been several reports that children with cleft lip and palate show delayed expressive language, evidenced by slower acquisition of sounds and words and restricted inventory of sounds in early infancy. It was also believed that they catch up by the age of three, usually after palate repair.
Fluency	● Some individuals with cleft lip and palate may experience stuttering or other fluency disorders. ● Even with early cleft repair, some children exhibit 'cleft palate speech' characterized by atypical consonant productions, abnormal nasal resonance, abnormal nasal airflow, altered laryngeal voice quality, and nasal or facial grimaces.
Pragmatics	● Social communication skills, such as turn-taking, topic maintenance, and understanding non-verbal cues, may be affected in individuals with cleft lip and palate. ● Children with cleft lip and palate may show a less assertive style of conversational participation, at least during the preschool years.

TYPES, COURSE, & PROGNOSIS

TYPES	DESCRIPTION	COURSE	PROGNOSIS
PRIMARY CLEFT PALATE			
Unilateral Incomplete 	One-sided incomplete cleft, often occurring on the left side.	May require surgical repair for functional and aesthetic reasons.	Favorable with proper surgical intervention and post-operative care.
Bilateral Incomplete	Both sides have incomplete clefts.	Similar to unilateral incomplete cleft.	Prognosis depends on the extent and severity of the cleft.
Unilateral Complete 	One-sided complete cleft, extending through the entire lip, nostril sill, and alveolar ridge to the incisive foramen.	Requires comprehensive surgical correction.	Good prognosis with timely surgical intervention and multidisciplinary care.
Bilateral Complete 	Both sides have complete clefts, isolating the prolabium and premaxilla bone.	Requires extensive surgical intervention.	Prognosis depends on the surgical approach and post-operative management.
Microform Cleft 	Very mild form of incomplete cleft, such as forme fruste or Simonart's band.	Often managed conservatively or with minor surgical procedures.	Generally good prognosis, may require minimal intervention for cosmetic purposes.
Nasolabial Fistula	Residual fistula left open	Follow-up care to monitor closure	Good prognosis with proper management

	during initial repair, later closed by bone graft.	and prevent complications.	and follow-up care.
SECONDARY CLEFT PALATE			
Incomplete  Incomplete cleft palate	Does not extend all the way to the incisive foramen. Can be minor, like forme fruste or Simonart's band.	May require surgical correction based on functional impact.	Prognosis varies depending on the extent of the cleft and associated complications.
Complete  Complete cleft palate Unilateral complete cleft lip and palate Bilateral complete cleft lip and palate	Extends to the incisive foramen, affecting the hard palate, velum, and uvula.	Often requires surgical repair for speech and swallowing issues.	Prognosis depends on the extent of correction achieved and post-operative care.
Nasolabial Fistula	Residual fistula left open during initial repair, later closed by bone graft.	Follow-up care to monitor closure and prevent complications.	Good prognosis with proper management and follow-up care.
SUBMUCOUS CLEFT PALATE			
Overt	Visible on the oral surface with characteristic features.	Can be incomplete (hypoplastic uvula) or complete (extends to incisive foramen).	May cause velopharyngeal insufficiency and hypernasal speech.
Occult	Hidden on the oral surface, detectable through nasopharyngoscopy.		
FACIAL CLEFTS			
Midline	Involve the central facial structures, such as the nose and upper lip, and may be associated with	Varies widely, from mild notching to severe anomalies affecting multiple structures.	Variable, depends on the extent and associated anomalies. May require multiple surgeries for correction.

	midline anomalies like bifid nose or hypertelorism.		
Paramedian	Occur adjacent to the midline and can affect the nasal structures, lips, and possibly the palate, often accompanied by asymmetry of the face.		
Orbital	Impact the eye area, including the eyelids, orbits, and sometimes extend to the nasal region, potentially causing visual and facial deformities.		
Oblique	Extend diagonally from the mouth towards the cheek, involving facial bones, nasal structures, orbits, and ears, leading to complex craniofacial anomalies.		

Factor	Description
Severity	Varies from narrow to very wide clefts, affecting structures like the lip, alveolar ridge, and nasal ala.
Embryological Fusion Lines	Follows primary and secondary palate suture lines, affecting structures anterior and posterior to the incisive foramen.
Treatment	Surgical repair often leaves a nasolabial fistula open temporarily to aid maxillary growth, later closed by bone graft.

Impact on Nasal Formation	Clefts affect nasal ala, columella, and overall nose appearance, especially when extensive or wide.
Visual Classification	Kernahan's striped-Y diagram visually represents type and extent of cleft, aiding in treatment planning and prognosis assessment.

MANAGEMENT

MEDICAL/SURGICAL MANAGEMENT

1. **Healthcare Resources**
   - Patients with cleft conditions require access to a range of healthcare resources, including:
     - Pediatric surgeons for corrective surgeries

CLEFT LIP REPAIR

- typically performed when the infant is around 3 to 6 months old, depending on the individual's health and surgeon's recommendation.

UNILATERAL

- **Millard Technique (Rotation Advancement Flap)**

Description: The Millard technique is a commonly used method for repairing unilateral cleft lips, accounting for about 80% of cases. It is considered one of the most anatomical repairs.
Procedure: The lip is repaired in three layers from inside out: mucosa, muscle, and skin. A "patch" of tissue is inserted to lengthen the shortened philtral ridge.
Philtral Ridge Lengthening: The patch of tissue is inserted at the top of the lip, just beneath the nose. This brings the shortened philtral ridge down to match the unaffected side, improving symmetry and aesthetics.

- **Tennison-Randall Technique (or Modification Thereof):**

Description: The Tennison-Randall technique, or a modification of it, is another approach for repairing unilateral cleft lips.
Procedure: Similar to the Millard technique, the lip is repaired in three layers. The shortened philtral ridge is lengthened by inserting a tissue patch just above the vermilion border of the lip.
Philtral Ridge Positioning: This technique positions the tissue patch slightly differently than the Millard technique, aiming to achieve symmetry and a natural appearance.

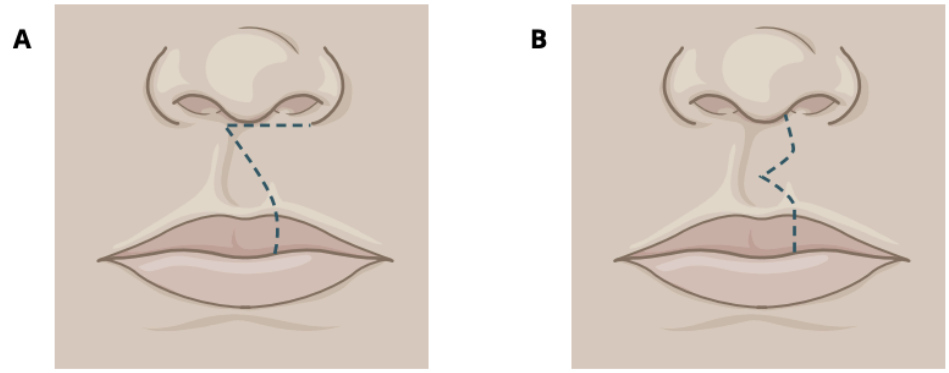


FIGURE 17-1 Unilateral cleft lip repairs. **(A)** Millard repair. **(B)** Tennison-Randall repair.

BILATERAL

- **Modified Manchester Repair**

Description: The Modified Manchester Repair is one of the major methods used for repairing bilateral cleft lips.
Procedure: The surgery includes repair of the mucosa, muscle, and then the skin layers of the lip. The prolabium, located just in front of the premaxilla, is a key focus area.
Muscle Realignment: Due to the discontinuity of the orbicularis oris muscle in bilateral clefts, the muscles insert abnormally on either side of the piriform aperture at the alar bases. During surgery, both muscles are detached from their abnormal insertions and realigned to create the orbicularis oris sphincter.

- **Millard Bilateral Cleft Repair**

Description: The Millard Bilateral Cleft Repair is another major technique used for repairing bilateral cleft lips.
Procedure: Similar to the unilateral repair, the surgery involves repairing the mucosa, muscle, and skin layers of both sides of the cleft lip.
Muscle Realignment: In cases of bilateral clefts, the orbicularis oris muscles may also need to be realigned to create a functional sphincter for improved lip function and aesthetics.

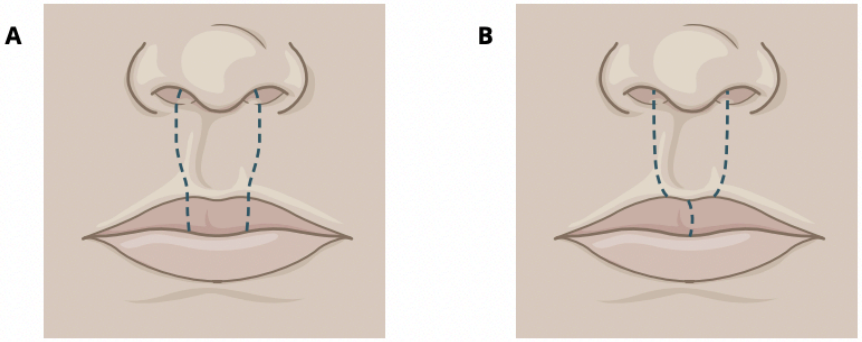


FIGURE 17-4 Bilateral cleft lip repairs. **(A)** Modified Manchester repair. **(B)** Millard bilateral cleft repair.

CLEFT PALATE REPAIR

- usually performed between 9 to 18 months of age, once the palate is large enough for surgical repair but before significant speech development occurs.

PALATOPLASTY

- The goals of palatoplasty are to:
  - Close the opening between the nose and mouth.
  - Help create a palate that works well for speech.
  - Prevent food and liquid from leaking out of the nose.

PROCEDURE: The surgeon may need to make two incisions (cuts) on each side of the palate behind the gums to ease tension on the palate repair. These are called “relaxing incisions.” In some cases, the surgeon may also borrow some tissue from the inner surface of the cheeks to help complete the repair.

CLEFT PALATE REPAIR TECHNIQUES

- **von Langenbeck**

Description: The von Langenbeck repair is one of the oldest and most successful means of palatal closure and is still popular today.

Procedure: An incision is made just inside the gum line, starting behind the area of the molars and extending up to the area of

the canine tooth. The mucoperiosteum is carefully raised off the bone and, in conjunction with the velum, separated in one large layer. The cleft margin is incised, and the raw edges are brought together and sewn down the middle. The incisions along the gum line are usually left open and fill in naturally with time.

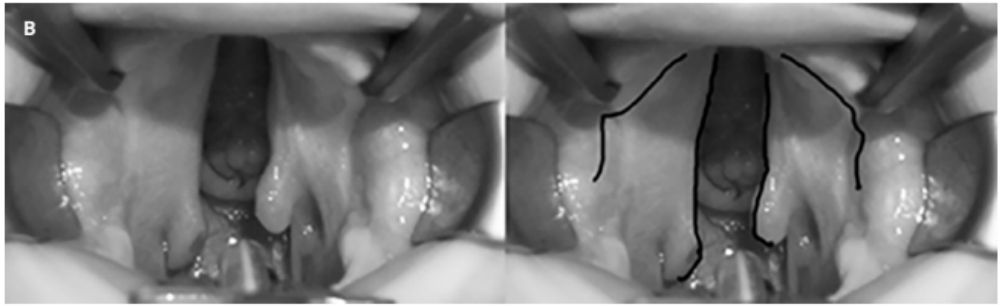


FIGURE 17-5 von Langenbeck repair. **(A)** Note lateral relaxing incisions, which are left open. **(B)** Photograph of the palate marked for von Langenbeck repair. Anterior palate remains attached, forming a bipedicle flap.

- **Wardill–Kilner V–Y Pushback**

Description: This procedure frees up the mucoperiosteum of the whole palate and allows it to be pushed back in an attempt to lengthen it.

Procedure: The initial incisions are similar to those of the von Langenbeck procedure except instead of leaving the mucoperiosteum attached in the front of the mouth, it is cut across as a V. The resulting open area is Y shaped.

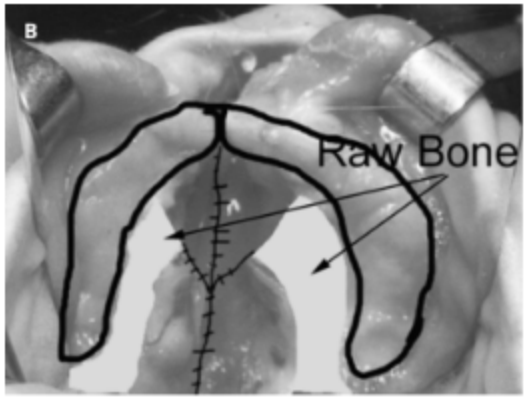


FIGURE 17-6 Wardill–Kilner repair. **(A)** The palate is lengthened by "pushing" back the mucoperiosteum. The raw area is allowed to fill by secondary healing (scarring). **(B)** Photograph of the Wardill–Kilner repair.

- **Intravelar Veloplasty (IVVP)**

Description: Normalizes the velopharyngeal sling and can be done in conjunction with any type of palate repair.
Procedure: Under general anesthesia, the surgeon exposes the cleft palate area, makes incisions along its edges, and repositions the soft tissues, including muscles and other structures, to close the gap. The tissues are then sutured together, and the incision site is closed.

- **Furlow Z-Palatoplasty**

Description: a plastic surgery technique that is used to lengthen tissue.
Procedure: Involves reconstruction of the levator sling and lengthening the velum by closing it with opposing Z-plasties.

    - Otolaryngologists for ear, nose, and throat evaluation
    - Orthodontists for dental alignment
    - Speech-language pathologists (SLPs) for speech and language therapy
    - Nurses for post-operative care and monitoring
- Rehabilitation services such as feeding specialists for addressing swallowing difficulties

2. Under SLP Therapy

Areas for Evaluation	• **Speech Sound Production** • **Nasal Emission** • **Resonance** • **Phonation** • **Visual Detection** • **Tactile Detection** • **Auditory Detection**
Evaluation Materials	• **Diagnostic Interview** • **Language Screening** • **Speech Assessment** • **Speech Samples**
Intervention Strategies	• **Articulation therapy techniques:** Targeting specific speech sounds affected by the cleft • **Language intervention:** Developing vocabulary, grammar, and communication skills • **Feeding therapy:** Techniques to improve oral motor function and safe swallowing

CRITICAL MEMBERS OF THE MANAGEMENT TEAM

1. **Otolaryngologist (ENT Specialist)**
   - Responsible for monitoring middle ear function and hearing
   - Treats middle ear disease, common in children with cleft anomalies
   - Manages upper airway obstruction, crucial for infants with Pierre Robin sequence
   - Assesses and treats structural aspects of the oral and nasal cavities, pharynx, and upper airway
   - Performs nasopharyngoscopy evaluations and surgeries for velopharyngeal insufficiency (VPI)
2. **Pediatrician**
   - Assesses overall medical health, growth, and development
   - Identifies other medical conditions that may impact surgical plans
   - Coordinates with specialists for comprehensive care
3. **Plastic Surgeon**
   - Performs surgical repair of cleft lip and palate

- Reconstructs facial and cranial anomalies
- Corrects velopharyngeal insufficiency (VPI) through surgery
- Performs bone grafts and orthognathic surgeries

4. **Prosthodontist**
   - Restores or replaces missing teeth
   - Develops devices for improving oral and facial structures
   - Assists with velopharyngeal closure using devices if surgery is not feasible
5. **Psychologist**
   - Assesses psychosocial needs of patients and families
   - Helps cope with emotional challenges related to anomalies and medical conditions
   - Assists in determining a patient's emotional readiness for surgeries
6. **Pulmonologist**
   - Evaluates airway issues and sleep problems
   - Monitors airway and orders sleep studies if obstructive sleep apnea (OSA) is suspected
7. **Social Worker**
   - Assists families in managing challenges related to the child's special needs
   - Coordinates appointments and assists with insurance and funding
   - Provides emotional support and stress management for families
8. **Speech-Language Pathologist (SLP)**
   - Counsels parents on communication development and home strategies
   - Evaluates feeding, swallowing, speech, language, resonance, and velopharyngeal function
   - Provides therapy for communication and feeding/swallowing disorders
   - Performs nasopharyngoscopy evaluations
9. **Team Coordinator**
   - Represents the team in interactions with parents and healthcare professionals
   - Schedules patients for team meetings and compiles recommendations for comprehensive reports
   - Counsels families on team recommendations and ensures follow-up on recommendations

MEDICAL PRECAUTIONS IN SPEECH-LANGUAGE THERAPY

1. **Safety Actions**
   - Sterilization of therapy tools and equipment to prevent infections
   - Maintaining a clean therapy environment to reduce exposure to allergens
   - Proper positioning and handling during therapy sessions to ensure client safety
2. **Preventive Measures**

Before SLP Therapy	**1. Health Assessment** ● Conduct a thorough pre-therapy health assessment, including respiratory status evaluation, to ensure the client is medically stable for therapy ● Screen for any allergies or sensitivities that may impact therapy (e.g., latex allergy) **2. Communication with Medical Team** ● Communicate with the client's medical team to understand any recent surgical procedures, medications, or medical concerns that may affect therapy. **3. Environment Preparation** ● Ensure the therapy environment is clean, free from allergens, and conducive to safe and effective therapy sessions. ● Set up appropriate seating and positioning equipment to support the client's comfort and safety during therapy.
During SLP Therapy	**1. Sterilization and Hygiene** ● Maintain strict hygiene protocols, including regular hand washing and sterilization of therapy tools and equipment, to prevent infections and cross-contamination. ● Use disposable items or clean reusable items thoroughly between sessions. **2. Safe Handling Techniques** ● Employ safe handling techniques when working with clients with cleft conditions, especially if they have undergone recent surgical procedures or have sensitive areas. ● Avoid excessive pressure on surgical sites or areas of healing. **3. Monitoring** ● Continuously monitor the client's comfort level and response during therapy, adjusting techniques or activities as needed to prevent discomfort or distress. ● Watch for signs of fatigue or respiratory challenges, and modify activities accordingly.
After SLP Therapy	**1. Post-therapy Care**

	● Provide post-therapy care instructions to the client and their caregivers, including guidance on home exercises, strategies, and follow-up appointments. ● Educate caregivers on proper feeding techniques and swallowing precautions if relevant to the client's condition. **2. Documentation and Communication** ● Document the session outcomes, progress, and any concerns in the client's medical record or therapy notes for future reference. ● Communicate any notable changes or observations to the client's medical team for ongoing care coordination. **3. Follow-up Assessment** ● Schedule regular follow-up assessments to track progress, reassess therapy goals, and address any new challenges or needs that may arise.

SUPPORT SYSTEMS FOR PEOPLE WITH CLEFT

PHILIPPINE BAND OF MERCY	The Philippine Band of Mercy offers free surgical repair for patients with cleft lip and palate, as well as other craniofacial anomalies. They also provide rehabilitation and follow-up care.
NOORDHOOF CRANIOFACIAL FOUNDATION, INC	provides comprehensive cleft care services, including surgery, speech therapy, dental care, and psychosocial support for individuals and families affected by cleft lip and palate.
SMILE TRAIN PHILIPPINES/INTERNATIONAL	Smile Train provides free cleft surgery and comprehensive cleft care to children in the Philippines. They partner with local hospitals and healthcare professionals to provide sustainable solutions.

OPERATION SMILE PHILIPPINES/INTERNATIONAL	Operation Smile conducts medical missions and provides free surgeries for children and young adults born with cleft lip and cleft palate in various countries, including the Philippines. They also offer comprehensive care and support services.
ABOUTFACE (CANADA)	AboutFace is a Canadian organization that provides support, resources, and advocacy for individuals with facial differences and their families. While based in Canada, they offer online resources and support that can be accessed internationally.
CDU SLP CARES CLINIC	provides comprehensive care to patients with cleft lip and palate and other craniofacial anomalies.

CRANIOFACIAL ANOMALIES

DEFINITION

- Craniofacial conditions refer to a group of disorders or anomalies affecting the head and face, including the skull, facial bones, and soft tissues, which may lead to functional and aesthetic challenges (Kummer, 2018).

ETIOLOGY

- The causes of craniofacial conditions vary and can include genetic factors, environmental influences, or a combination of both. Genetic syndromes are a significant contributor, with over 400 distinct syndromes associated with facial clefts alone (Gorlin, Cohen, & Hennekam, 2001). These syndromes are diagnosed based on the pattern of major and minor malformations, and they may involve a range of genetic abnormalities affecting facial development (Jones & Jones, 2009).

PREVALENCE & INCIDENCE

Locally	*Internationally*
● Down Syndrome: 1 in every 800 babies born (PNA, 2023) ● Over 100,000 Filipino households are living with a person with Down syndrome, based on the data released by the Down Syndrome Association of the Philippines Inc. (DSAPI) in 2014. ● At one hospital in Bacolod City, medical records of infants born with clefts of the lip and/or palate and other major anomalies were reviewed and findings include a birth prevalence of 1.94 per 1000 live births for cleft lip with/without palate (PubMed, 1997). ● Pascasio (2023): 20 to 25 patients during a medical mission they conducted, a significant majority (70 to 80%) of their patients with cleft and craniofacial anomalies hailed from Mindanao.	● Demke and Tatum (2021): overall incidence is considered to be 0.2 to 0.5 per 1000 births. ● Robin Sequence: 1 in 8500–1 in 20,000 births (2023) ● Tavares and Moody (2022): 35% of babies are born with craniofacial abnormalities of the skull, jaws, ears, and/or teeth.

SIGNS, SYMPTOMS, PATHOMECHANICS

Manifestations that the **Physician/Allied Health Professional** Perceive	● Physicians and allied health professionals often observe craniofacial anomalies through clinical examination, imaging studies (such as X-rays, CT scans, or MRIs), and sometimes genetic testing. ● Manifestations may include abnormal facial features such as cleft lip or palate, asymmetry of facial structures, abnormal skull shape (craniosynostosis), or malformations of the ears, eyes, or nose. ● Functional issues such as difficulty breathing, eating, or speaking may also be noted. ● Additionally, healthcare providers may assess developmental delays or neurological symptoms associated with certain craniofacial anomalies.
Manifestations that the **Parents/Significant Others** Perceive	● They may notice physical differences in their child's appearance compared to other children, leading to concerns about social acceptance and potential stigmatization. ● Parents may also observe functional challenges in their child's faces, such as difficulty feeding, breathing, or speaking, which can impact daily life and require specialized care.
Manifestations that the **Patient** Experiences	● Experience physical discomfort, pain, or functional limitations ● Difficulties in breathing, eating, or speaking. ● They may also face psychosocial challenges, including low self-esteem, bullying, and social isolation due to their appearance. ● Emotional distress, depression, and anxiety are particularly common during adolescence when body image becomes more salient. ● Patients may also have concerns about surgical procedures, recovery, and long-term outcomes, which can impact their overall well-being and quality of life
PATHOMECHANICS AND PATHOPHYSIOLOGY	
EMBRYOLOGICAL DEVELOPMENT	During embryonic development, the craniofacial structures undergo intricate processes of growth, fusion, and differentiation. Disruptions in these processes can occur due to genetic mutations, environmental factors, or a combination of both. For example, anomalies such as cleft lip and palate

	result from failure of fusion of facial prominences during early gestation.
GENETIC FACTORS	Many craniofacial anomalies have a genetic basis, although the inheritance patterns can vary. Some anomalies are caused by mutations in single genes, while others may result from chromosomal abnormalities or multifactorial inheritance involving the interaction of multiple genes with environmental factors.
ENVIRONMENTAL INFLUENCES	Factors such as maternal nutrition, exposure to teratogenic substances (e.g., alcohol, certain medications), infections, and maternal illnesses can influence craniofacial development. Maternal smoking, for instance, has been associated with an increased risk of orofacial clefts.
STRUCTURAL ABNORMALITIES	Craniofacial anomalies can manifest as structural abnormalities affecting the bones, muscles, and soft tissues of the skull and face. These abnormalities can range from mild to severe and may involve asymmetry, hypoplasia (underdevelopment), hyperplasia (overdevelopment), or agenesis (absence) of craniofacial structures.
STRUCTURAL ABNORMALITIES	Craniofacial anomalies can manifest as structural abnormalities affecting the bones, muscles, and soft tissues of the skull and face. These abnormalities can range from mild to severe and may involve asymmetry, hypoplasia (underdevelopment), hyperplasia (overdevelopment), or agenesis (absence) of craniofacial structures.

STRUCTURAL AND ANATOMICAL CHANGES

Cleft Lip and Palate	● Separation or gap in the upper lip, which may extend into the nostril (unilateral or bilateral) ● Incomplete fusion of the facial structures during embryonic development, leading to gaps in the lip and/or palate.
Craniosynostosis	● Abnormal skull shape and restricted skull growth ● Premature suture fusion affects skull growth patterns, resulting in characteristic deformities such as a long, narrow head (scaphocephaly), asymmetric forehead and orbits (plagiocephaly), triangular-shaped forehead (trigonocephaly), or flattening of the back of the head (brachycephaly)

Microtia	● Underdevelopment or absence of the external ear (pinna)
Anotia	● Complete absence of the external ear

POSSIBLE SPEECH-LANGUAGE PROBLEMS ASSOCIATED WITH THE CONDITION

- Articulation
  - Structural differences in the oral cavity, such as cleft lip and palate or malocclusion, can affect the ability to produce speech sounds accurately, leading to articulation disorders.

- Resonance
  - Conditions like velopharyngeal dysfunction (VPD), often associated with cleft palate, can lead to hypernasality or nasal air escape, affecting the resonance and quality of speech.

- Voice
  - Anomalies affecting the larynx or vocal folds can result in changes in voice quality, pitch, and volume. SLPs may work with individuals to improve vocal production and address voice disorders.

- Language
  - Craniofacial anomalies may impact language development, including vocabulary acquisition, grammar, and language comprehension. SLPs assess and address language delays or disorders through therapy and intervention.

- Fluency
  - Some individuals with craniofacial anomalies may experience fluency disorders such as stuttering or cluttering, which SLPs can assess and treat through various therapeutic approaches.

- Feeding and Swallowing
  - Craniofacial anomalies can affect oral motor function, leading to difficulties with feeding and swallowing. SLPs play a crucial role in evaluating swallowing function and providing interventions to improve safety and efficiency of swallowing.

- Social Communication:
  - Individuals with craniofacial anomalies may experience challenges in social communication skills, such as pragmatic language difficulties or social anxiety related to their condition. SLPs provide support and strategies to improve social interaction and communication.

TYPES, COURSE, & PROGNOSIS

Types	● **Beckwith–Wiedemann Syndrome:** Genetic disorder that results in prenatal and postnatal overgrowth characterized by large body size, abdominal wall defects, and increased risk of certain tumors. ● **CHARGE Syndrome:** Acronym for coloboma, heart defect, choanal atresia, retarded growth and/or development, genitourinary anomalies, and ear anomalies and/or deafness. It is a genetic condition causing a range of developmental and physical challenges, including heart defects and sensory impairments. [Cleft Lip (with or without Cleft Palate) & Cleft Palate Only] ● **Down Syndrome (Trisomy 21):** A genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21 causing intellectual disability, distinctive facial features, and various health issues. ● **Fetal Alcohol Syndrome:** Caused by maternal alcohol consumption during pregnancy, leading to developmental delays, facial abnormalities, and cognitive impairments. [Cleft Lip (with or without Cleft Palate)] ● **Hemifacial Microsomia:** Condition where one side of the face is underdeveloped, affecting the ear, jaw, and cheekbone. [Cleft Lip (with or without Cleft Palate) & Cleft Palate Only] ● **Kabuki Syndrome:** Genetic disorder causing distinctive facial features like wide palpebral fissures with eversion (turning out) of the lateral portion of the lower lid, external ear anomalies, arched eyebrows, and a broad nasal tip, and developmental delays, and skeletal abnormalities. [Cleft Palate Only] ● **Moebius Syndrome:** Rare neurological condition resulting in facial paralysis and difficulties with eye movements and facial expressions. ● **Neurofibromatosis Type 1:** Genetic disorder causing tumors to grow on nerves, leading to skin changes, bone deformities, and other complications. ● **Opitz G Syndrome:** Rare genetic disorder affecting facial features, intellectual development, and various organ systems. [Cleft Lip (with or without Cleft Palate)] ● **Oral-Facial-Digital Syndrome Type I:** X-linked dominant disorder affecting the development of the mouth, face, and digits, leading to abnormalities in these areas. [Cleft Lip (with or without Cleft Palate)] ● **Pierre Robin Sequence:** Congenital condition involving a small lower jaw, cleft palate, and difficulty breathing. ● **Stickler Syndrome:** Genetic disorder causing distinctive facial features, joint problems, and eye abnormalities such as cataracts. [Cleft Palate Only]

<table>
<tr><td></td><td>

Treacher Collins Syndrome: Genetic disorder causing facial deformities, hearing loss, and dental abnormalities.
Trisomy 13: Born with an extra copy of chromosome 13 causing severe developmental abnormalities, often leading to early death. [Cleft Lip (with or without Cleft Palate)]
Van der Woude Syndrome: Genetic condition causing cleft lip and/or cleft palate, along with other features like lip pits. [Cleft Lip (with or without Cleft Palate) & Cleft Palate Only]
Velocardiofacial/22q11.2 Deletion Syndrome: Genetic disorder causing a range of physical, developmental, and behavioral challenges, including cleft palate, heart defects, and learning disabilities. [Cleft Palate Only]
Wolf-Hirschhorn Syndrome (4p Syndrome): Deletion or missing portion of the short arm of chromosome 4. Genetic disorder characterized by distinctive facial features, intellectual disability, and developmental delays. [Cleft Lip (with or without Cleft Palate)

</td></tr>
<tr><td>Most Common in the Philippines</td><td>
<table>
<tr><th>TYPES</th><th>DESCRIPTION</th><th>COURSE</th><th>PROGNOSIS</th></tr>
<tr><td>DOWN SYNDROME (TRISOMY 21)</td><td>Most common chromosome disorder with an incidence of 1 in 700 live births. A genetic disorder caused when abnormal cell division results in an extra full or partial copy of chromosome 21. Down syndrome is primarily categorized on the severity of intellectual disability, ranging from mild to moderate.</td><td>Down syndrome is a lifelong condition that manifests from birth and continues throughout the individual’s life.</td><td>Varies among individuals and is influenced by factors such as the severity of associated health conditions, access to medical care, and early intervention services. Developmental milestones, including motor skills, speech and language development, and cognitive abilities, may be delayed compared to typically developing peers.</td></tr>
</table>
</td></tr>
</table>

	Neurofibromatosis Type 1	Also known as von Recklinghausen disease, is a genetic disorder characterized by the development of tumors along nerves throughout the body. It is caused by mutations in the NF1 gene, which is involved in regulating cell growth and division. NF1 is a multisystem disorder that can affect various organs and systems, including the skin, nervous system, bones, and eyes. Neurofibromatosis Type 1 (NF1) is primarily diagnosed based on clinical evaluation and the presence of specific features, including café au lait macules, neurofibromas, freckling in certain areas, optic pathway tumors, Lisch nodules, osseous lesions, and family history of NF1.	Symptoms may present in childhood or adulthood and may change over time. Neurofibromas typically appear during adolescence or early adulthood and may increase in number and size over the years. While most neurofibromas are benign, some individuals with NF1 may develop malignant peripheral nerve sheath tumors (MPNSTs), a rare but serious complication.	Untreated NF1 can lead to progressive neurofibroma growth, potential malignancy in some cases, and neurological complications like velopharyngeal incompetence.While NF1 is a chronic condition that requires ongoing management, many individuals with NF1 lead productive lives with appropriate medical intervention and support. Early diagnosis and proactive management of symptoms and associated complications are key to optimizing outcomes for individuals with NF1. Genetic counseling may be beneficial for individuals and families affected by NF1 to understand inheritance patterns, assess risks, and make informed decisions about family planning.

Pierre Robin Sequence	Pierre Robin Sequence is primarily characterized by micrognathia, which is associated with several craniofacial syndromes such as Stickler syndrome, Treacher Collins syndrome, velocardiofacial/22q11.2 deletion syndrome (VCFS/22q deletion syndrome), and fetal alcohol syndrome.	If left untreated, Pierre Robin Sequence can lead to severe upper airway obstruction in newborns due to glossoptosis, which may require interventions like prone positioning, nasopharyngeal airway tubes, or even tracheostomy. The presence of a tracheostomy tube can interfere with vocalizations and speech development.	The prognosis varies depending on the severity of the condition and the effectiveness of interventions. Early management strategies like mandibular distraction can lead to normal respiration and feeding, but speech issues related to the cleft palate may persist, especially if tracheostomy is required for an extended period.
Treacher Collins Syndrome	Treacher Collins syndrome, also known as mandibulofacial dysostosis, is primarily classified as an autosomal dominant condition with variable expressivity, making it challenging to predict the outcomes in offspring of affected individuals. However, there are no distinct subtypes or variations	The condition typically progresses with classic features such as downward-slanting palpebral fissures, colobomas of the lower eyelids, micrognathia (underdeveloped jaw), hypoplasia of the maxilla and zygomatic arches, macrostomia (large mouth), microtia (small or dysplastic ear), and atresia of the	Despite the physical challenges presented by Treacher Collins syndrome, intelligence is typically normal in affected individuals. However, speech disorders are common due to hearing loss and micrognathia, which can be further complicated by the presence of cleft palate or airway obstruction. The overall

		within the syndrome itself.	external auditory canal. Conductive hearing loss is common due to middle ear anomalies, often leading to speech disorders. Some individuals may also experience Pierre Robin sequence, although clefts are not universally present.	prognosis can vary depending on the severity of associated complications and the individual's access to medical interventions and supportive care.

MANAGEMENT

MEDICAL/SURGICAL
1. Healthcare Resources: a. Comprehensive medical evaluation by a pediatrician or geneticist. b. Surgical interventions for specific anomalies (e.g., craniofacial surgeries, airway management). c. Rehabilitation services (e.g., physical therapy, occupational therapy).
-same as those mentioned above (cleft portion)

THE HEALTHCARE TEAM FOR DENTAL CONDITIONS AND DENTAL HYGIENE

Dentists/Dental Surgeons	● Primary care providers for oral health

	● Diagnose and treat a wide range of dental conditions, including cavities, gum disease, tooth decay, and root canals ● Perform preventive care procedures like cleanings, fluoride treatments, and sealants, as well as restorative procedures like fillings, crowns, and bridges ● Oral surgeons are dentists with additional training to perform surgeries in the mouth and jaw, such as wisdom teeth removal, dental implant placement, and corrective jaw surgery
Orthodontists	● Dentists who specialize in straightening teeth and correcting misaligned bites ● Use braces, retainers, and other corrective appliances to move teeth into their proper positions
Oral and Maxillofacial Surgeons	● Surgeons who specialize in treating conditions of the mouth, face, and jaws ● Perform a wide range of surgeries, including complex tooth extractions, jaw reconstruction, facial trauma repair, cleft lip & palate repair, and tumor removal
Speech-Language Pathologists	● Can collaborate with dentists and orthodontists to address speech difficulties caused by dental problems, such as missing teeth or cleft lip and palate
Periodontists	● Dentists who specialize in the treatment of gum disease ● Provide deep cleanings (scaling and root planing), gum surgery (flap surgery), and bone grafting procedures to treat gum disease and support teeth
Implant Dentists	● Dentists with advanced training in placing dental implants—artificial tooth roots surgically placed in the jawbone to support crowns, bridges, or dentures
Prosthodontics	● Dentists who specialize in restoring and replacing missing teeth ● Create dentures, bridges, crowns, and other dental prosthetics to improve a person's smile, function, and appearance

Oral Rehabilitative Professionals	● A team of dental specialists who work together to restore a patient's oral health and function
Physicians	● Play a role in oral health by identifying and managing systemic conditions that can affect oral health, such as diabetes and HIV/AIDS ● May also prescribe medication to treat oral infections or manage pain
Psychologists	● Can help address dental anxiety and phobia, which can interfere with a person's ability to seek dental care ● Can provide therapy to help patients develop coping mechanisms for dental fear and anxiety
Physiotherapists	● Can help treat temporomandibular joint disorder (TMJ), which can cause pain and discomfort in the jaw joint ● May use massage, exercises, and other techniques to improve jaw mobility and reduce pain.

SUPPORT SYSTEMS FOR PEOPLE WITH CRANIOFACIAL ANOMALIES

PHILIPPINE BAND OF MERCY	The Philippine Band of Mercy offers free surgical repair for patients with cleft lip and palate, as well as other craniofacial anomalies. They also provide rehabilitation and follow-up care.
NOORDHOOF CRANIOFACIAL FOUNDATION, INC	provides comprehensive cleft care services, including surgery, speech therapy, dental care, and psychosocial support for individuals and families affected by cleft lip and palate.
SMILE TRAIN	Smile Train provides free cleft surgery and comprehensive cleft care to children in the Philippines. They

PHILIPPINES/INTERNATIONAL	partner with local hospitals and healthcare professionals to provide sustainable solutions.
OPERATION SMILE PHILIPPINES/INTERNATIONAL	Operation Smile conducts medical missions and provides free surgeries for children and young adults born with cleft lip and cleft palate in various countries, including the Philippines. They also offer comprehensive care and support services.
ABOUTFACE (CANADA)	AboutFace is a Canadian organization that provides support, resources, and advocacy for individuals with facial differences and their families. While based in Canada, they offer online resources and support that can be accessed internationally.
CDU SLP CARES CLINIC	provides comprehensive care to patients with cleft lip and palate and other craniofacial anomalies.

REFERENCES

Alasti, F., Van Camp, G., & Smith, R. J. H. (2008). The Genetics of Microtia and Associated Syndromes. Journal of Medical Genetics, 45(11), 669–678. https://doi.org/10.1136/jmg.2008.058123

Bauman-Waengler, J. (2019). *Articulation and Phonology in Speech Sound Disorders: A Clinical Focus*. Pearson.

Berkowitz, S. (2009). Cleft Lip and Palate: Diagnosis and Management (3rd ed.). Springer.

Broder, H. L., & Smith, F. B. (2005). Relations between malocclusion and adolescents' judgments of facial attractiveness. Community Dentistry and Oral Epidemiology, 33(5), 399–404. https://doi.org/10.1111/j.1600-0528.2005.00243.x

Chapman, K., Graham, K. T., Gooch, J. L., & Visconti, C. F. (1998, November 1). Conversational Skills of Preschool and School-Age Children with Cleft Lip and Palate. ˜the œCleft Palate-craniofacial Journal (Print). https://doi.org/10.1597/1545-1569_1998_035_0503_csopas_2.3.co_2

Cleft lip and palate. (2017, March 27). https://www.stlukes.com.ph/health-library/health-articles/cleft-lip-and-palate

Cleft lip and palate. (n.d.). Johns Hopkins Medicine. https://www.hopkinsmedicine.org/health/conditions-and-diseases/cleft-lip-and-palate

Cleft Lip and Palate - MN Dept. of Health. (n.d.). https://www.health.state.mn.us/diseases/cy/cleftlippalate.html

Dirmann, J. (2021, June 14). ,. , - YouTube. Retrieved April 5, 2024, from https://www.asha.org/public/speech/disorders/cleftlip/

Dixon, M. J., Marazita, M. L., Beaty, T. H., & Murray, J. C. (2011). Cleft lip and palate: understanding genetic and environmental influences. Nature Reviews Genetics, 12(3), 167–178. https://doi.org/10.1038/nrg2933

DSc, M. M. T. M. P. (n.d.). Pediatric Cleft Lip and Palate: Practice Essentials, Pathophysiology, Etiology. https://emedicine.medscape.com/article/995535-overview?form=fpf

Facts about Cleft Lip and Cleft Palate | CDC. (2023, June 28). Centers for Disease Control and Prevention. https://www.cdc.gov/ncbddd/birthdefects/cleftlip.html

Giustina, A., Allora, A., Frara, S., Spina, A., & Mortini, P. (2022). The hypothalamus. In *Elsevier eBooks* (pp. 301–340). https://doi.org/10.1016/b978-0-323-99899-4.00002-0

Gorlin, R. J., Cohen, M. M., & Hennekam, R. C. (2001). Syndromes of the Head and Neck (Oxford Monographs on Medical Genetics). Oxford University Press.

Home | Smile Train. (n.d.). https://www.smiletrain.ph

Home| Operation Smile Philippines. (n.d.). https://operationsmile.org.ph/

Hunt, O., Burden, D., Hepper, P., & Johnston, C. (2006). The psychosocial effects of cleft lip and palate: a systematic review. European Journal of Orthodontics, 28(3), 274–285. https://doi.org/10.1093/ejo/cji103

Kaban, L. B., & Troulis, M. J. (Eds.). (2018). Pediatric Oral and Maxillofacial Surgery. Elsevier.

Kummer, A. W. (2018). *Cleft Palate and Craniofacial Conditions: A Comprehensive Guide to Clinical Management: A Comprehensive Guide to Clinical Management*. Jones & Bartlett Learning.

McCarthy, J. G., & Grayson, B. H. (Eds.). (2012). Craniofacial Surgery (3rd ed.). Thieme.

Muntz, H. R., & Meier, J. D. (2013). The financial impact of unrepaired cleft lip and palate in the Philippines. International

Journal of Pediatric Otorhinolaryngology, 77(12), 1925–1928. https://doi.org/10.1016/j.ijporl.2013.08.023

Murray, J. C., Daack-Hirsch, S., Buetow, K. H., Munger, R. G., Espina, L., Paglinawan, N., Villanueva, E., Rary, J., Magee, K., & Magee, W. P. (1997). Clinical and epidemiologic studies of Cleft Up and Palate in the Philippines. *The Cleft Palate Craniofacial Journal*, *34*(1), 7–10. https://doi.org/10.1597/1545-1569_1997_034_0007_caesoc_2.3.co_2

PCO pushes for inclusion of people with Down syndrome. (n.d.). Philippine News Agency. https://www.pna.gov.ph/articles/1194405#:~:text=Statistics%20showed%20that%20one%20in,of%20the%20typical%2046%20chromosomes.

Pope, A. W., Snyder, H. T., & Mood, L. (2005). Psychosocial Adjustment in Children and Adolescents with Craniofacial Anomalies: A Review. Journal of Pediatric Psychology, 30(8), 647–662. https://doi.org/10.1093/jpepsy/jsi046

Rojean Grace G. Patumbon, & Rojean Grace G. Patumbon. (2023, November 20). *Free craniofacial surgeries now available in Davao*. SunStar Publishing Inc. https://www.sunstar.com.ph/davao/free-craniofacial-surgeries-now-available-in-davao

Stock, N. M., Feragen, K. B., & Rumsey, N. (2015). "We're not hiding anything, but …": Facial difference and its meaning in the everyday lives of British Pakistani women living in the United Kingdom. Body Image, 14, 1–10. https://doi.org/10.1016/j.bodyim.2015.02.005

Surg, I. (2009, Oct). Communication disorders in individuals with cleft lip and palate: An overview. Retrieved from: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2825064/#:~:text=There%20have%20been%20several%20reports,three%2C%20usually%20after%20palate%20repair.

Tavares, A. L. P., & Moody, S. A. (2022). Advances in understanding the pathogenesis of craniofacial birth defects. *Journal of Developmental Biology*, *10*(3), 27. https://doi.org/10.3390/jdb10030027