Cleft Palate Speech and Feeding

*Train the Trainer*
Module 5: Syndromes

- Treacher Collins Syndrome
- 22q11.2 Deletion Syndrome
- Pierre Robin Sequence
- Moebius Syndrome

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Genetics of cleft lip
(with or without cleft palate)

● Substantial underlying genetic pathogenesis

● Most cases are isolated
  ● No associated syndromes or other birth defects
Genetics of cleft palate only

- A substantial number are caused by
  - multiple congenital syndromes or
  - multiple congenital anomaly disorder
Treacher Collins Syndrome

- Underdeveloped facial bones (especially impacted are the cheekbones).
- Cleft palate (possibly)
- Small jaw and chin and skull shape is atypical.
- Tiny nose, mouth, and throat airways.
- Potential eye, dental, and hearing issues.
- Typically, no delays in cognition or language.
Treacher Collins Syndrome
22q11.2 is a Common Genetic Disorder

- According to the International 22q11.2 Foundation Inc., 22q11.2 occurs 1 in every 2000-4000 births
- Second leading cause of congenital heart defects
- No differences in regards to gender (impacts males and females at equal rates and severities)
Areas Affected by 22q11.2
Significant Variability across Affected Individuals

- Heart abnormalities
- Cognitive deficits
- Language deficits
- Immune dysfunction
- Growth disturbances
- Developmental abnormalities
- Psychiatric abnormalities
- Learning difficulty
22q11.2 previously known as...

- DeGeorge Syndrome
- Velocardiofacial Syndrome
- Shprintzen Syndrome

Now all called 22q11.2 deletion syndrome. All are caused by the same chromosomal deletion.
Underdiagnosis of Africans and Others From The African Diaspora with 22q11.2

- No race differences in terms of prevalence
  
  BUT

- There has been shown to be an underdiagnosis in Africans and those from the African diaspora due to similar facial features associated with 22q11.2
How is 22q11.2 Diagnosed?

- Diagnosed with a blood test
- FISH test used to diagnose (finds the 22q11.2 deletion)
- Microarray analysis
  - More sensitive. Identifies 22q deletion and 22q duplications
Velopharyngeal Dysfunction and 22q11.2

Only 5-10% of those with 22q11.2 have a visible cleft palate that requires surgical repair.

Majority of patients with 22q11.2 have hypernasal speech with either:

- A submucous cleft; or
- A palate that appears adequate, but does not function appropriately for speech due to velopharyngeal disproportion, that is the posterior pharyngeal wall is set back.
Velopharyngeal Dysfunction and 22q11.2

Palatopharyngeal disproportion
- The palate (although may appear typical) is unable to touch the posterior pharyngeal wall
  - This is very typical in children with 22q11.2
- The palate structure and palate length is normal, however, the posterior pharyngeal wall is set further back (deep pharynx) in patients who have 22q11.2
The Role of Speech and Language Therapy in 22q11.2

- Approximately 90-95% of those with 22q11.2 have a speech and or language delay such as:
  - Articulation deficits in 75% of patients, with poor speech intelligibility;
  - Difficulty producing speech sounds;
  - Delayed onset of first words, with 70% nonverbal at age 2;
  - Delayed expressive skills;
  - Comprehension issues, but generally stronger than expressive; and/or
  - Social language skills.
Role of speech language therapy in 22q11.2

- **Hypernasal speech**, due to palatal dysfunction
- **Voice disorders**
  - Hoarse, Breathy, Low volume
- **Vocal fold paralysis and/or laryngeal web**
- **Motor speech disorders**
  - Dysarthria
  - Cranial nerve abnormalities
  - Apraxia
Pierre Robin Sequence

1. Wide, bell-shaped cleft palate

2. Micrognathia
   • Small jaw or mandible, also known as \textit{retrognathia}

3. Glossoptosis
   • Atypical placement in the back of the tongue
     (tongue typically is downward and back)
Pierre Robin Sequence

● **Cause**
  
  ● Small mandible makes the tongue stay high in the oral cavity
  
  ● This position of the tongue interfere with the closing of the velum
Pierre Robin Sequence: Breathing Complications

- Following birth, glossoptosis (tongue typically is downward and back) can block the upper airway
- This may cause severe respiratory distress
Prevalence of Pierre Robin Sequence

- 1 in 14,000 live births
  - 66% classic U-shaped cleft palate
  - 33% had other malformations
  - Stickler Syndrome 6 in 10

Printzlau and Andersen 2007 Cleft Palate Craniofacial J. 2004 41(1) 47-52. Study in Denmark. But they had to have a cleft to be included—so this study probably understates the problem.
Moebius Syndrome

- Predominantly affects the 6th-Abduces and 7th-Facial cranial nerves, but other cranial nerves can be affected
- Often cannot smile, frown, or blink eyes
- Difficulty with lateral eye movements
- Generally normal intelligence and cognition
- Prevalence estimated at 2-20 per 1 million people.
Moebius Syndrome: Physical Appearance

- Small chin (micrognathia)
- Small mouth (microstomia)
- Short or atypical shape/movement of tongue
- Cleft palate
- Could have high arch in the oral cavity
- Missing teeth
- May have limb disturbances
Moebius Syndrome Impairments

- Respiratory issues
- Speech, feeding, and swallowing issues
- Impaired vision
- Sensory integration deficits
- Problems sleeping
- Upper body strength weakness
Credits

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All Cleft Palate Speech Therapy Resources Available for FREE download LEADERSProject.org

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Speech Sound Assessment and Stimulability
Acevedo Spoke
Therapy Word Games
Therapy Books for Phrases and Sentences

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References


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