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Learner Outcomes

As a result of this Continuing Education Activity, participants will be able to:

1. Describe physical characteristics (or facial features) of patients with 22Q.
2. List common medical problems associated with 22Q.
3. Discuss common speech characteristics associated with 22Q.
4. Compare and contrast when surgical intervention is indicated versus speech therapy based on case history and speech samples.
5. Explain what apraxia is and how it may affect velopharyngeal closure for speech.
DEFINING 22Q

Diagnosing 22Q

- FISH (fluorescence in situ hybridization) probe
- Looks for the specific deletion on chromosome 22, on the q (long) arm, in region 11.2
- Autosomal dominant
Syndrome Identification
Importance for patient care:
• Can anticipate problems through natural history of syndrome
• Allows providers and caregivers to plan appropriate treatment and have realistic goals
• Important for genetic counseling for family planning

What is VCFS/22Q?
• **Velo:** velopharyngeal dysfunction
• **Cardio:** minor cardiac, vascular anomalies
• **Facial:** dysmorphic facial features
• **Other:** small stature, long fingers, learning disabilities, oral motor dysfunction, psychological concerns, other medical problems
Velo Characteristics

Velopharyngeal Dysfunction
- Cleft or submucous cleft of the soft palate (often associated with Pierre Robin sequence)*
- Occult submucous cleft
- Pharyngeal hypotonia

Cardio Characteristics

Cardiac Anomalies
- Ventricular septal defect (VSD)
- Atrial septal defect (ASD)
- Patent ductus arteriosus (PDA)
- Pulmonary stenosis

Vascular Anomalies
- Right-sided aortic arch
- Tortuosity of retinal blood vessels
- Medially displaced internal carotid arteries
Facial Characteristics

- Long, narrow face with vertical maxillary excess
- Narrow palpebral fissures
- Flattened malar eminences
- Broad nasal bridge
- Narrow alar base and bulbous nasal tip

Facial Characteristics

- Minor auricular anomalies
- Thin upper lip
- Micrognathia, often with Class II malocclusion
- Microcephaly
Physical Features

- Small size and stature, usually below the 10th percentile in weight and height
- Long, slender fingers
- Hyper-extensibility of the joints

Common Medical Problems

- 22Q can effect multiple body systems, including:
  - Immune system
  - Endocrine system
  - Neurological system
- Pierre Robin sequence
  - Micrognathia (small mandible)
  - Cleft or submucous cleft
  - Glossoptosis (posterior tongue position) with airway obstruction
- Laryngeal web
- Umbilical or inguinal hernias
Common Functional Problems

- Early feeding problems
- Gross and fine motor dysfunction
- Conductive hearing loss due to chronic otitis media
- Language and learning disabilities
- Hypernasality
- Verbal apraxia
- High-pitched voice

Team Approach
Effects on Feeding, Speech, Resonance, Development, and Cognition

Early Feeding Problems

Due to:
• velopharyngeal insufficiency (VPI)
• hypotonia
• cardiac issues
• poor oral-motor coordination
• anatomical abnormalities
• gastro-intestinal difficulties
Feeding Adaptations

• Special bottles or nipples
• Positioning
• External pacing
• Medical interventions
• Failure to Thrive, may require extra calories
• Tube feeding
• Work with SLP, OT, ENT, GI and/or dietician
Structures Involved in Velopharyngeal Closure

- Velum (soft palate)
- Lateral pharyngeal walls (side walls of the throat)
- Posterior pharyngeal wall (back wall of the throat)
Velum during Speech

Kummer, 2009

Lateral Pharyngeal Walls

Kummer, 2009
Function of VP Valve

• Closes off nose from mouth.
• Allows air and sound to enter the mouth for speech.
• Important for the production of vowels and oral consonants.

Normal VP Closure
(Nasopharyngoscopy)
Video 1: Nasopharyngoscopy of Normal VP Closure

VPI and VPI

**Velopharyngeal insufficiency:** An anatomical or structural defect that precludes adequate velopharyngeal closure by causing the velum to be short relative to the posterior pharyngeal wall.

**Velopharyngeal incompetence:** A neuromotor or physiological disorder that results in poor movement of the velopharyngeal structures.
Velopharyngeal Insufficiency (VPI): Velum is too short

Video 2: VP Insufficiency (VPI)
Video 3: Medialized Internal Carotid during Nasopharyngoscopy

Evaluation Process

• Qualitative evaluation (resonance, airflow, articulation, voice quality)
• Quantitative evaluation (nasometry)
• Nasopharyngoscopy
• Team Format: SLP, ENT/plastic surgeon, geneticist (craniofacial team)
Resonance

• Hypernasality
• Hyponasality
• Cul-de-sac Resonance
• Mixed Resonance
Nasal Emission

• Inaudible nasal emission
• Audible nasal emission
• Nasal rustle
• Phoneme-specific nasal emission (PSNE)

Speech Characteristics

• Obligatory distortions
• Compensatory errors
• Apraxia
Video 4: Speech Sample of Child with 22q

Nasometer
Childhood Apraxia of Speech

• A neuromotor disorder.

• Problems with coordination and planning of the movements needed to make a sound or word.

Characteristics of Apraxia of Speech

• inconsistent speech sound errors
• difficulty moving from one speech sound to another or one syllable to another
• abnormal rhythm, stress and intonation during speech
• limited repertoire of vowels – little to no differentiation between vowel sounds
• variability in accurate speech production based on context
• more difficulty with volitional, self-initiated speech than modeled, more ‘rote’ speech
Characteristics of Apraxia of Speech

- use of only vowel sounds, grunts, or single syllables to communicate
- more errors on longer sentences or longer words than with single sounds or syllables
- difficulty and struggle when trying to find the right mouth position to make a sound
- normal receptive language (understanding) skills, but limited expressive language (talking) skills
- difficulty imitating mouth movements (in severe cases)
- voicing voiceless sounds

Apraxia and VPI

- Apraxia can also affect the coordination and timing of velopharyngeal closure for resonance.
- Velum is also an ‘articulator.’
- Importance of differential diagnosis.
- Bouncing of the velum during nasopharyngoscopy.
- Difficulty maintaining coordination during succession of nasal words of increasing complexity.
Video 5: Qualitative Evaluation

Video 6: Nasopharyngoscopy of child with Apraxia
VPI and the 22Q diagnosis

• Prognosis for total correction of VPI may be guarded due to pharyngeal hypotonia and oral-motor difficulties.
• Often have poor lateral pharyngeal wall movement.
• Most still require speech therapy following surgery to learn to use the changed structures.

Surgery for VPI – Pharyngeal Flap

[Diagram showing pharyngeal flap with labels: Velum, Flap, Posterior pharyngeal wall]
Treatment

• Treatment based on cause of speech/resonance disorder.

• Appropriate evaluation of speech and resonance is paramount to recommending the right treatment.

• Referral to craniofacial team for evaluation by specialized professionals.

• Surgery vs. therapy.
Language and 22Q

- Language difficulties
- Learning disabilities
- Reading difficulties
- Difficulty with abstraction
- Socialization (pragmatic language) difficulties

Cognition and 22Q

- Intelligence in the low to normal range.
Psychiatric Profile

Increased incidence of:
• Behavior difficulties
• Schizophrenia and schizo-affective disorder
• Depression
• Bipolar disorder

General Treatment Guidelines

• Predictability, routines
• Concrete learners – concrete examples
• Use visual schedules, timer
• Verbal cues
• Repetition
• Appropriate expectations
Case Study 1

- 4yo 22Q with history of nasal rustle and speech sound disorder
History

- **Diagnoses:** 784.59 Other speech disturbance, 758.32 Velo-Cardio-Facial Syndrome with 22q11.2 deletion, 781.3 Muscular incoordination

- **Medical issues:** astigmatism, heart complications, delayed development in speech and language

- **Surgeries:** bilateral medial rectus recession (eye surgery), ventral septal defect repair, ventricular septal defect closure, pulmonary artery stenosis repair, transesophageal echocardiogram

- **Hearing status:** WNL

- **Oral Mechanism:** high palate with no other defects of palate/uvula, oral mechanism appropriate

- **Speech-language therapy:** KY First Steps to age 3 and school and private

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Current Speech/Language Goals

**LTG:** To improve early expressive speech/language skills

- **STG:** To demonstrate use of /sh/ at word level with 75% accuracy given fading cues.

- **STG:** To demonstrate use of /k/ in all positions of words given fading cues with 80% accuracy.

- **STG:** To demonstrate use of s-blends at word level with 80% accuracy (sp-, sn-, st-, sw-, sm-, sk-; errors ok with str-) given fading cues.

- **STG:** Tell how an object is used when shown a picture or real object for 10 of 12 items.

- **STG:** To demonstrate use of plurals when describing items or pictures in 8 of 10 occurrences during 1 session given fading cues

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**CONTINUED**
## Nasometry

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### Oral Nasalance Score

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<th>Oral Nasalance Score</th>
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<tr>
<td>Over 56%</td>
<td>Severely high</td>
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Language Assessment at age 3:6

- Preschool Language Scale
  - Receptive language standard score 112
  - Expressive language standard score 82
  - Total language standard score 97

- Patient demonstrated age appropriate understanding with difficulty using grammatical markers (i.e., present progressive, plurals, possessives)

Articulation Proficiency

- GFTA-3
  - Standard Score: 70
  - Percentile Rank: 2

- Articulation errors: t/k, d/g, b/v, t/ch, f/th (voiceless), d/th (voiced), y/l, w/r, L-blends, R-blends, f/s-blends

- No evidence of apraxia
Recommendations

• Continue monitoring language skills
• Continue addressing speech targets using:
  • Frequent repetition of targets
  • Segmenting and blending of sounds
  • Multisensory feedback (visual, tactile, auditory)
• Monitor velopharyngeal function

Case Study 2

6 year old with 22Q and Apraxia
History

- **Diagnoses:** 22Q
- **Medical issues:** Obstructive sleep apnea (OSA), feeding problems, pulmonary anomalies
- **Hearing status:** WNL
- **Speech:** Apraxia of speech, VPI, language deficits

Speech & Language Sample Pre-flap
### SNAP test, Age 5-5

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### Nasal Passage Picture Cued Subtest

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### Language Assessment at age 6:5

- **CELF-P2**

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Articulation Proficiency

- Obligatory distortions include nasalized consonants
- Compensatory errors include glottal stops and pharyngeal fricatives
- Correct production of /p/, /b/, /d/, /t/ in single words and short phrases with her nares occluded.
- Characteristics of Apraxia

Recommendations

- Pharyngeal flap to remediate VPI
- Pharyngeal flap was delayed due to medical issues
- Continue speech and language therapy
Speech – Language Therapy

Goals of treatment

• To imitate CV sequences (i.e., CV, CVCV, VCV) using "h, n, m, p/b, t/d" with 80% accuracy given fading cues.
• Morelia will demonstrate use of /f/ in final position of one to two syllable words given fading cues with 75% accuracy.
• To demonstrate understanding of spatial concepts (under, in back of, next to, in front of) with 70% accuracy given fading cues in structured tasks.

Language Sample Post-flap
Case Study 3

- 16 y.o. submucous cleft palate (SMCP) & 22Q

History

- **Diagnoses:** 22Q w/ SMCP
- **Medical issues:** muscle weakness & tightness, limited range of motion, scoliosis, pulmonary concerns
- **Speech Diagnosis:** resonance disorder and articulation disorder
- **Intelligibility Rating:** mild
- **Cognitive Issues:** mild-moderate delay
- **Language Level:** mild-moderate delay
- **Speech-language therapy:** discharged based on progress
## Resonance Assessment at age 16

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## Pre-surgery Video, Age 16y4

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## Resonance Assessment post-flap

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### Oral Nasalance Score vs. Severity Rating

- <25%: Normal
- 26-40%: Mildly high
- 41-55%: Moderately high
- Over 56%: Severely high

## Video Post-flap, Age 16-11
Speech – Language Therapy

Goals of treatment
• To produce /f/ and /v/ at the word level with 100% success and in sentences with 90% success.
• To produce initial /k/ and /g/ at the word level with 90% success.
  • To produce /k/ and /g/ at the sentence level and conversational level with 90% success.
  • To produce /k/ correctly during oral reading with 90% success
• To produce /th/ at the word, phrase, and sentence levels with 90% success.

References
References


References


Questions?

• Jennifer Marshall, M.A., CCC-SLP
  Jennifer.marshall@cchmc.org
• Margaret Wilson, M.A., CCC-SLP
  Margaret.Wilson@cchmc.org

Resource

Sign up now for updates and SLP tools from Cincinnati Children’s Division of Speech-Language Pathology

Link: https://viablesynergy.wufoo.com/forms/s3q62e1k51n5v/