Velo-Cardio-Facial Syndrome: Early Development and Intervention

Valerie E. Boyer, PhD, CCC-SLP
Southern Illinois University
Carbondale, IL
Leah Fullman, MS, CCC-SLP
Children's Rehab Center
Grand Island, NE
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Disclosure Statement

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Outcomes

- Discuss relationship between early medical concerns and early feeding development
- Describe early speech and language development in children with VCFS
- Integrate specific developmental information regarding VCFS into treatment planning in EI

Introduction

- VCFS ≠ Cleft Palate
- Most common syndrome of cleft palate.
- VCFS is a complex, genetic disorder
  - Significant range in phenotype
  - Pervasive
  - Much more research about effects than intervention

Overview of VCFS

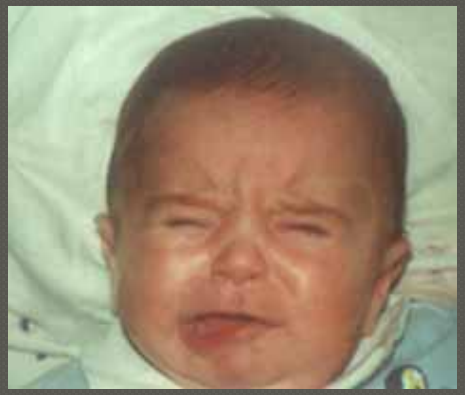
- Most common microdeletion syndrome
- Second most common chromosomal defect
- Prevalence of 1 in 2000 births in U.S.
- Also called 22q11 Deletion syndrome, and Shprintzen syndrome
- DiGeorge syndrome, CATCH 22 syndrome
- First described in 1978

Phenotype

- Diversity – more than 180 clinical findings, but not all are present in any one individual with VCFS
- Most common findings:
  - Velopharyngeal Insufficiency in 90%
  - Cleft palate in 98%, submucous in 70%
  - Cardiac anomalies in 82%
  - Typical facies (in all patients to varying degrees)
  - Learning difficulties (99%)

Phenotype

- Typical Facies
  - Thin, down slanting palpebral fissures
  - Limited facial expression
  - Assymetric crying facies
  - Wide nasal bridge
  - Flat malar region
  - Retrognathia



Etiology

- Hemizygous gene deletion – 2-3 million base pairs of DNA across 30-40 genes on chromosome 22 are missing
- Majority de novo – not inherited
- 6-10% of cases hereditary
- Autosomal dominant

Diagnosis of VCFS

- Cardiac anomaly + cleft palate typically results in referrals
- Hypocalcemia or asymmetric crying facies can also lead to early referral
- Flourescence in situ hybridization (FISH) testing
- Echocardiogram (EKG)
- Blood tests (immune system dysfunction)

Medical Issues in Infancy

- Cardiac anomalies include tetralogy of Fallot and ventricular septal defect
- Cleft Palate/VPI
- Hypotonia
- Hypocalcemia
- Cardiovascular anomalies (vascular ring)
- Gastroesophageal Reflux

All can have negative impact on feeding

Feeding

- CP, VPI, hypotonia, cardiovascular defects
  - Nasal regurgitation and reflux
  - Decreased intraoral pressure
  - Poor coordination of suck-swallow-breath
  - Slow peristalsis and constipation
  - Obstructed esophagus
  - Fatigue
  - Slower, weaker oral and pharyngeal stages of swallow
  - Aspiration

Implications for Practice

- Failure to Thrive
- Medical management
- Adaptations
- Pacing
- Oral sensory exercises
- Pre-chaining

Language Development

- Delays in language development reported in first three years of life
- Expressive language deficit > Receptive deficit
- Degree of deficit increases between first and third years of life

Prevalence of Language Delay

- Emergence of language delayed in all 40 children with VCFS (Gerdes et al., 1999)
- Gerdes et al. (2001) administered PLS-3 to 50 kids with VCFS.
  - 34% scored 2 standard deviations (SDs) below the mean on total language score and
  - 46% were between 1 and 2 SD's below the mean.
  - 20% were within normal limits.

Language Profile

- Scherer et al. (1999) compared four children over time between 6 months and 3 years of age with three other groups
  - (a) typical peers
  - (b) kids with cleft lip + palate (CLP) and
  - (c) kids with isolated cleft palate (ICP)
- By 12 months, significant difference in receptive and expressive language for VCFS group compared with typical peers
- By 24 months, significant differences in receptive and expressive language for VCFS group compared with CLP
- By 30 months, significant differences in receptive and expressive for VCFS group compared with ICP group

Oral Speech Challenges

- Similarities to cleft palate in type of errors
- Difference in severity of articulation and resonance deficits
- Glottal stops
- Hypernasality
- Decreased oral consonant inventory

Articulation Development

- Scherer et al. (1999) reported VCFS group significantly fewer consonant types than kids with CLP and ICP by 24 months of age.
- D'Antonio et al. (2001) described children under seven with VCFS with severe speech impairment but children older than 7 a mild speech impairment.
  - Children in the younger group with VCFS (less than 7 years of age) produced fewer consonants than either older kids with VCFS and children without VCFS
  - Younger kids with VCFS produced all manner of consonants, but fewer types in each manner category
  - More restrictions in place preferring front and glottal placement
  - Preferred voiceless to voiced across manner and placement
  - Children with VCFS used glottal stops more

Hypernasality

- Severe hypernasality present in 70-75% of cases (Shprintzen, 2000; 2005)
  - can be trigger for referral to craniofacial team
- Scherer et al. (1999) 100% VCFS group had hypernasality or nasal emission compared with 50% in CLP & ICP group
- D'Antonio et al. (2001) documented absence of velar motion and lateral wall movement in endoscopic evaluations

Social Development

- Early separation anxiety
- Less emotional expression
- Less imaginative play
- Poor initiations and reciprocity with peers – often play independently of other kids
- Reports of less compliance with directions and less motivation by praise
- Strengths - resiliency

Co-Morbidity

- Autism – Fine et al. (2005) 11 out of 98 children with VCFS appropriate diagnosis of ASD
- Distinguishing between VCFS + ASD and VCFS alone can be challenging
- Kates et al. (2007) – capacity for make-believe, stereotypies, and nonverbal social interaction deficits may be key for differential diagnosis

Implications for Practice

- Early Intervention recommended
- Focus on language in conjunction with social skills and articulation
- Encourage families with information regarding improvement typically in later preschool years
- Deficits become less severe with age
- Surgical management for VPI + speech therapy

Implications for Practice

- Sign language as AAC strategy to facilitate expressive language development
- Receptive language aspects to focus on
  - Directions
  - Concepts
- Traditional approach to oral speech development – teach place and manner
- Early opportunities to play with peers
- Teaching responsiveness
- Promote reinforcement of oral sounds not glottal sounds

Implications for Practice

- Repetition and structure are often helpful - rote memory is a strength
- Build on strengths including resiliency, concrete learners, and music/computers
- Lots of repetition and intensity
- Individuality of families

Resources

- www.vcfsef.org – National educational foundation for VCFS
- www.vcfscenter.com – Virtual center for VCFS in which you can connect with experts in the area of VCFS
- www.friendsofquinn.com – Blogs and Q & A website focused on learning disabilities

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