Velo-Cardio-Facial Syndrome: Early Development and Intervention

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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
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
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%)
Typical Facies
- Thin, down slanting palpebral fissures
- Limited facial expression
- Asymmetric crying facies
- Wide nasal bridge
- Flat malar region
- Retrognathia
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
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
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
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.
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.
Similarities to cleft palate in type of errors
Difference in severity of articulation and resonance deficits
Glottal stops
Hypernasality
Decreased oral consonant inventory
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
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
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
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.vcfsccenter.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


