

Early Childhood Intervention And Velocardiofacial Syndrome

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VCFS and ECI

- **Learning Objectives**

- 1. Understand the importance of early identification of VCFS and begin to consider the implications of the new law, including how it impacts their role with families.
- 2. Understand the prevalence of VCFS in the general population, and within the population with a history of cardiac disorder, and /or cleft palate,
- 3. Be familiar with the common characteristics of VCFS have an understanding of the possible co-existing diagnosis associated with VCFS.
- 4. Have an understanding of resources available about VCFS

Cayler Syndrome

Takao Syndrome

Velocardiofacial Syndrome

22q11.2 Deletion Syndrome

Catch 22 Syndrome

Shprintzen Syndrome

Conotruncal Anomalies Face
Syndrome

DiGeorge Syndrome

Sedlackova Syndrome

VCFS and ECI

- 1:2,000 - General Population
- 1:1,800 – Live Births
- Autosomal Dominant
- “New Mutations”
- FISH Study vs. Chromosome
- 90% with 3-Mb deletion
spanning 3 million base pairs of
DNA
- Late Diagnosis

VCFS and ECI

VCFS and ECI

- Neonate - Infancy
 - Presence of Hyptonia
 - Global
 - Laryngeal/Pharyngeal
 - Feeding Complications
 - Nasal Regurgitation
 - Presence of midline cardiac defects
 - VSD, ASD, Pulmonic Artresia or Stenosis, TOF, Truncus Arteriosus, Vascular Ring

VCFS and ECI

- Feeding
 - If cleft palate, this is NOT driving force for feeding disorder
 - Pharyngeal Hypotonia affects feeding
 - Pharyngeal hypotonia
 - Decreased oral and pharyngeal sensation
 - Decreased oral and pharyngeal motor
 - GI motility problems
 - VPD
 - Nasal Regurgitation during feeding
 - O₂/Heart rate decrease during feeding
 - Cardiac complications and feeding
 - Alternate method of feeding

VCFS and ECI

- Neonate - Infancy
 - Oral Facial
 - Cleft Palate
 - Pierre Robin Sequence
 - VPD
 - Asymmetrical Crying Facies
 - Craniosynostosis
 - Presence of Endocrine Disorders
 - Hypocalcemia
 - Hypothyroidism
 - Absent/Hypoplastic Thymus

VCFS and ECI

- Toddler/Early Childhood
 - Language Developmental Delay
 - Expressive > Receptive
 - Articulation Errors
 - Early use of
 - Glottal stops
 - Pharyngeal fricatives
 - Overuse of /m/, /n/, /ng/
 - Poorly differentiated vowels
 - Resonance Imbalance
 - Hypernasality

VCFS and ECI

- Toddler/Early Childhood
 - Motor Developmental Delay
 - Laryngeal Findings
 - Upper airway obstruction
 - Vocal cord paresis/paralysis
 - Ear/Hearing
 - Recurrent OME
 - Ear findings

VCFS and ECI

- Toddler/Early Childhood
 - Short stature/small for age
 - Eye Findings
 - Suborbital congestion (“allergic shiners”)
 - Narrow palpebral fissures
 - Oral/Facial Findings
 - Enamel hypoplasia
 - Flaccid/hypotonic facies
 - Long face
 - Prominent nasal bridge

VCFS and ECI

- Why is Early Identification So Important?
 - Medical
 - Evaluation of “at risk” systems
 - Cardiac
 - Immunology
 - Endocrinology
 - Velopharyngeal Function/Speech

VCFS and ECI

- Why is Early Identification So Important?
 - Educational
 - Transfer into School District
 - OHI Classification
 - Early identification and resources for possible learning differences
 - Other Resources
 - » OT

VCFS and ECI

- Questions?????