Pediatric Syndromes of Head and Neck

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More than 3,000 syndromes classified
Optimal growth, development, and learning requires early recognition and intervention

Team Approach:
- Parents
- Pediatrician
- Otolaryngologist
- Cardiologist
- Nephrologist
- Geneticist
- Speech Therapist
- Teachers
- Others
The Sydromal Child

• History
  - Parental factors (age)
  - Consanguinity
  - Abortions
  - Teratogen exposure
  - Medical Pedigree
Physical Exam

- Major and Minor Anomalies
  - Airway
  - Skull
  - Ears
  - Facial skeleton
- Comparison to Family Members
- Reference Material
Down Syndrome
Velocardiofacial Syndrome
Branchio-Otorenal Syndrome
Treacher-Collins Syndrome
Crouzon and Apert Syndrome
Pierre Robin Sequence
CHARGE Association
Down Syndrome
Described by John Landon Down in 1866

Etiology: nondisjunction mutation resulting in Trisomy 21

Prevalence 1:700

- Most common chromosomal anomaly

Associated with Maternal age > 35
Down Syndrome

- Facial Characteristics
  - Macroglossia
  - Micrognathia
  - Midface hypoplasia
  - Flat occiput
  - Flat nasal bridge
  - Epicanthal folds
  - Up-slanting palpebral fissures
  - Progressive enlargement of lips
Down Syndrome

Airway Concerns

Due to midface hypoplasia, the nasopharynx and oropharynx dimensions are smaller.

- Slight adenoid hypertrophy can cause upper airway obstruction.
- Congenital mild-moderate subglottic narrowing not uncommon.
- Post-extubation stridor.

Down Syndrome
• Obstructive Sleep Apnea
  – Prevalence 54-100% in DS patients
  – Combination of anatomic and functional mechanisms
    • Midface hypoplasia, macroglossia, etc
    • Hypotonia of pharyngeal muscles
Obstructive Sleep Apnea

Management:

- Polysomnography to confirm
- Medical interventions:
  - CPAP
  - Weight Loss
  - Medications to stimulate respiratory drive
Obstructive Sleep Apnea

Management:

- Surgical
  - Adenoidectomy and Tonsillectomy
    » Controversial
  - UPPP
  - Partial tongue resection
  - Tracheotomy

Down Syndrome
Otologic Concerns
- Small pinna, Stenotic EAC
  - Cerumen impaction
- CHL
  - ETD: PE tubes
  - Ossicular fixation: surgical correction
- SNHL
  - Progressive ossification along outflow pathway of basal spiral tract
• Cardiovascular anomalies (40%)
  – ASD, VSD, Tetralogy of Fallot, PDA
• GI anomalies (10-18%)
  – Pyloric stenosis, duodenal atresia, TE fistula
• Malignancy
  – 20 fold higher incidence of ALL
  – Gonadal tumors

Down Syndrome
Velocardiofacial Syndrome
First described by Shprintzen et al. in 1978

Not uncommon
- Prevalence 1 in every 4,000 newborns
- 8% of all cleft palate patients

Autosomal Dominant inheritance
- Hemizygous microdeletion shared with DiGeorge Sequence at 22q11.2 locus

Features
- Cleft palate
- Congenital heart disease
- Characteristic facies
- Hypernasal speech
- Learning disabilities
Oropharyngeal Findings:

- Apparent cleft palate (10-35%)
- Submucous cleft (33%)
- Submucous cleft and velar paresis (33%)
- Tonsils small or aplastic (50%)
- Adenoids small or aplastic (85%)
- Malocclusion
- Hypernasal speech
Airway Obstruction is common
- 50% of neonates with VCFS have OSA
- Adenotonsillectomy should be avoided if not indicated
- Oral airway needed in urgent setting
- Cleft palate repair required
Facial Findings:

- Maxillary excess
- Malar flatness
- Facial asymmetry
- Long philtrum
- Thin upper lip

Nasal Findings:
- Prominent nasal root
- Large tip
- Pinched, hypoplastic alar base

• **Ear findings**
  - Small auricles (48%)
  - CHL secondary to serous effusions and ETD (75%)
  • PE tubes effective
  - SNHL (8%)
  • Amplification devices
Cardiovascular Findings

- 75-80% with cardiac anomalies
- 10% of patients with VCFS die in early infancy due to these anomalies
- VSD (65%)
- Right sided aortic arch (35%)
- Tetralogy of Fallot (20%)
- Aberrant subclavian artery (20%)
VCFS

MRA:
Tortuous and medially deviated internal carotid artery

• Growth and mental retardation
• Flat affect and poor social interaction with impulsive behavior
• Renal anomalies in 35%
• T cell dysfuction in 10% with hypocalcemia

VCFS
Branchio-Otorenal Syndrome
BORS

- First termed by Melnick et al in 1975
- 1 in every 40,000 births
- Autosomal dominant inheritance
  - Isolated to 8q13.3 locus
- Characteristics:
  - Branchial cleft cysts or fistulas
  - Preauricular pits
  - Malformed auricles
  - Hearing loss
  - Renal anomalies
• Branchial cleft cysts and fistulas
  – Present in 50-60% of cases
  – Usually bilateral
  – Found in lower third of neck
  – Fistulas may connect to tonsillar fossa
• Facial nerve paralysis (10%)
• Aplasia or stenosis of lacrimal duct (25%)
- **External ear anomalies**
  - Auricular malformation (30-60%) or abnormal position
    - Minor aberration of anatomy to severe microtia
  - Helical or preauricular pits (70-80%)
- **Middle ear anomalies**
  - Malformation and/or fixation of ossicles
  - Abnormal size/structure of the tympanic cavity
• Inner ear anomalies (rare)
  – Dilated vestibule and/or endolymphatic duct/sac
  – Bulbous internal auditory canal
  – Small semicircular canals
  – Hypoplastic cochlea
• Mondini

• Hearing loss (75-95%)
  – CHL (30%)
  – SNHL (20%)
  – MHL (50%)
Renal anomalies (12-20\%)

- Likely underreported when a disease process not involved
- Renal agenesis or hypoplasia
- Structural anomalies of renal pelvis or ureters
• **Diagnosis and Treatment**
  – History and Physical Examination
  – Audiogram, CT temporal bones
  – CT neck
  – Renal Ultrasound, IVP
• **Diagnosis and Treatment**
  - Surgical excision of branchial cleft cyst, sinus, or fistula
  - Otoplasty
  - Excision of pits
  - Possible ossicular chain reconstruction
  - Hearing aids
  - Urology consultation for renal anomalies
Treacher Collins Syndrome
• First described by Thomson and Toynbee in 1846-7
  – Later, essential components described by Treacher Collins in 1960
• Autosomal dominant inheritance
  – TCOF1, mapped to 5q32-33.1
• 60% are from new mutation
  – Associated with increased paternal age
• Prevalence of 1 in 50,000
• a.k.a. Mandibulofacial dysostosis
Characteristics

- Likely due to abnormal migration of neural crest cells into first and second branchial arch structures
- Usually bilateral and symmetric
- Malar and supraorbital hypoplasia
- Non-fused zygomatic arches
- Cleft palate in 35%
- Hypoplastic paranasal sinuses
- Downward slanting palpebral fissures
- Mandibular hypoplasia with increased angulation
- Coloboma of lower eyelid with absent cilia
- Malformed pinna
- Normal intelligence
OP/Airway concerns

- Cleft palate
- Choanal atresia may be present
  - Respiratory distress in newborn
  - Oral airway, McGovern nipple
- Obstructive sleep apnea is the most common airway dysfunction
  - Mandibular hypoplasia results in retrodisplacement of tongue into oropharynx
  - Oral airway, tracheotomy
  - Distraction osteogenesis vs. free fibular transfer
• **Otologic concerns**
  – Malpositioned auricles
  – Malformed pinna
  – EAC atresia
  – Ossicular abnormalities
  – Conductive hearing loss is common
  • Hearing aids are effective
  – Normal intelligence
Apert and Crouzon Syndromes
• Belong to family of Craniosynostoses
• Apert Syndrome (Acrocephalosyndactyly)
  – First described by Wheaton in 1894
  – Apert further expanded in 1906
• Crouzon Syndrome (Craniofacial Dysostosis)
  – Described by Crouzon in 1912
• Autosomal dominant inheritance
  – Most are sporadic in Apert Syndrome
  – 1/3 are sporadic in Crouzon Syndrome
• Prevalence: 15 - 16 per 1,000,000
**Apert and Crouzon**

- **Typical characteristics**
  - **Craniosynostosis**
    - Coronal sutures fused at birth
    - Larger than average head circumference at birth
  - Midfacial malformation and hypoplasia
  - Shallow orbits with exophthalmos
  - Apert Syndrome: symmetric syndactyly of hands and feet
Apert and Crouzon

Crouzon and Apert Syndromes facial features

- Shallow orbits with exophthalmos
- Retruded midface with relative prognathism
- Beaked nose
- Hypertelorism
- Downward slanting palpebral fissures
• **Airway concerns**
  – Reduced nasopharyngeal dimensions and choanal stenosis
  – OSA
  – Cor pulmonale
• **Polysomnography**
• **Treatment**
  – Adenoidectomy
  – Endotracheal intubation
  – Tracheotomy
Apert and Crouzon

- **Otologic concerns**
  - CHL resulting from ETD
  - Congenital fixation of stapes footplate in Apert syndrome
- **Treatment**
  - Ventilation tubes
  - Stapedectomy or OCR
- **Fronto-Orbital advancement**
  - Brain growth and expansion of cranial vault, orbital depth
- **Orthodontics**
  - Maxillary teeth abnormalities
  - Crossbite
Apert and Crouzon

Fronto-Orbital Advancement Surgery

Syndactyly reconstruction in Apert Syndrome

Pierre Robin Sequence
• Triad of micrognathia, glossoptosis and cleft palate
  – First described by St. Hilaire in 1822
  – Pierre Robin first recognized the association of micrognathia and glossoptosis in 1923

• Prevalence: 1 of every 8,500 newborns
  – Syndromic 80%
    • Treacher Collins Syndrome
    • Velocardiofacial Syndrome
    • Fetal Alcohol Syndrome
  – Nonsyndromic 20%
Mandibular Deficiency

- Hypoplastic and Retrused Mandible (Micrognathia)

Tongue Remains Retrused and High in Oropharynx (Glossoptosis)

Cleft Palate

- Failure of Fusion of Lateral Palatal Shelves
• Airway Obstruction
  – Anatomic and Neuromuscular Components
    • Micrognathia, Retruded Mandible
    • Glossoptosis
    • Impaired Genioglossus and Parapharyngeal Muscles
Airway Management

- Temporizing Modalities
  - Prone Positioning
  - Nasopharyngeal Airway
    - NG tube and gavage feeds
  - Mandibular Traction Devices
  - Tongue Lip Adhesion
- Tracheotomy
- Distraction Osteogenesis
• Otologic Concerns
  – 80% have bilateral CHL
  – Eustachian Tube Dysfunction
  – Serous Otitis Media
  – Placement of Ventilation Tubes is Effective
CHARGE Association
- Colobomas
- Heart Abnormalities
- Atresia Choanae
- Growth/Mental Retardation
- Genitourinary Anomalies
- Ear Abnormalities
Proposed by Pagon et al in 1981
Incidence unknown
Associated with paternal age > 34
Head and Neck anomalies:

- Coloboma
- Choanal Atresia
- External Ear Abnormalities
- Facial Nerve Palsy
- Laryngomalacia
- OSA
- GERD
- Mondini Malformation
- Semicircular Canal Hypoplasia
- Vocal Cord Paresis
Coloboma

- Failure of fusion of embryonic (choroidal) fissure
  - Optic nerve, inferior nasal fundus, or inferior iris may be involved
- Redundant tissue of upper or lower eyelid lacking skin appendages

Choanal Atresia

- Prevalence: 1/5000 to 1/8000
- Females/Males: 2/1
- Unilateral 65-75%
- 75% with Bilateral have CHARGE, or other syndromes

Choanal Atresia

- Neonates are obligate nasal breathers
- Mouth breathing is a learned response, developed at 4-6 weeks
- Bilateral CA presents at birth with respiratory distress and cyanosis, relieved with crying
- Unilateral CA usually presents later in life with chronic nasal discharge
Choanal Atresia

- **Diagnosis:**
  - 6 French catheter
  - Nasal endoscopy
  - Bell of Stethoscope
  - Mirror

- **Radiology**
  - CT (preferred method)
Choanal Atresia

- **Treatment:**
  - Unilateral CA does not require immediate correction
    - May be delayed until starting school
  - Bilateral CA requires immediate interventions:
    - Oral Airway
    - McGovern Nipple
    - Intubation
    - Tracheostomy
Choanal Atresia

- Surgical Correction:
  - Transnasal
  - Transpalatal
  - Laser
  - +/- Stenting
  - +/- Mitomycin-C Topical (0.3 mg/cc)


