Syndrome Identification for the SLP

Presented by
Melinda Chalfonte-Evans, Ph.D. CCC-SLP
Diane Geiger, Ph.D. CCC-SLP
Christina Yeager, M.A. CCC-SLP

SYNDROME VS. SEQUENCE

- **Syndrome:** A group or recognizable pattern of symptoms, characteristics or abnormalities that indicate a particular trait or disease

- **Sequence**: Significant, initial defect results in a chain of defects

What's In a Name?

- Medical conditions and diseases named after a person are called an **eponyms**
- Possessive and non-possessive names were in use until 1974
- US National Institute of Health standardized rules regarding the naming of diseases and conditions.
  - "The possessive form of an eponym should be discontinued, since the author neither had nor owned the disorder."

Down Syndrome

- Genetic Condition
- Incidence is 1/800 to 1000 live births
- Chromosomes
  - Trisomy 21 (95%)
  - Translocation (21/21 and 21/14)
  - Mosaic

Associated Conditions

- Congenital heart defects in 40-50%
- Gastrointestinal malformations in 12-25%
- Congenital cataracts
- Congenital hypothyroidism in 1%

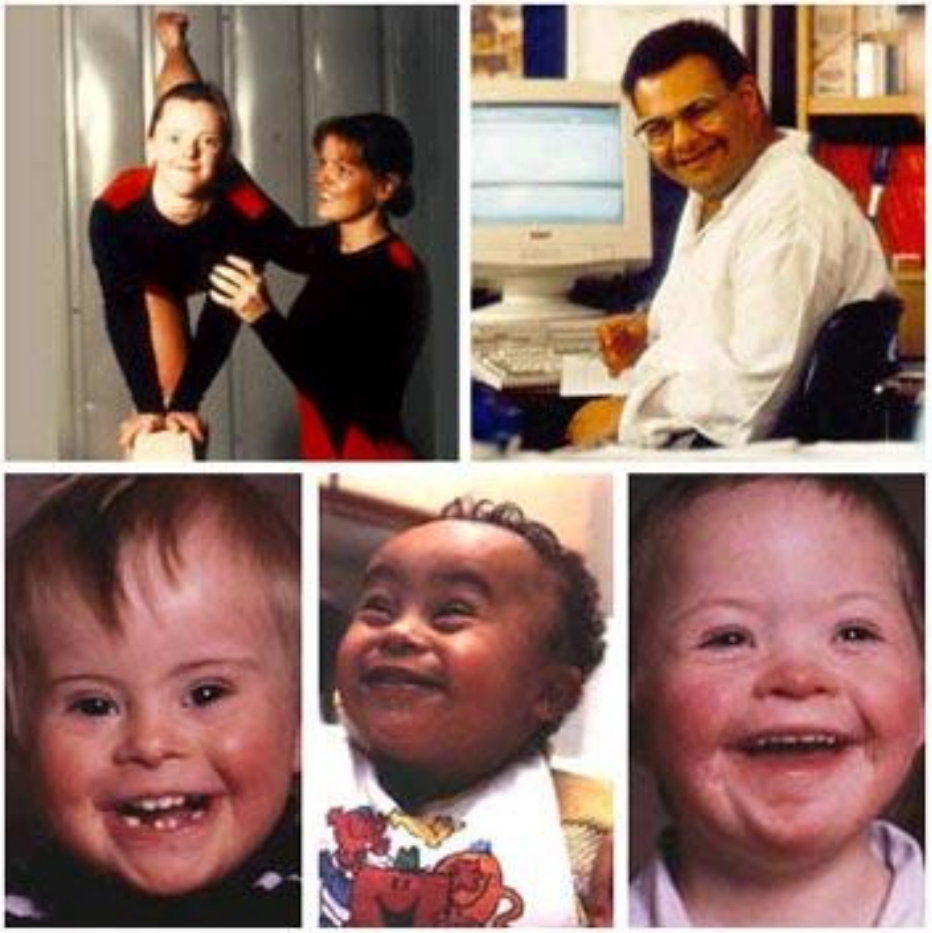
Common Medical Issues

- Vision problems including strabismus, nystagmus
- ENT/Audiologic disorders
- Sleep apnea
- Celiac disease
- Hypothyroidism
- Skeletal (atlanto-axial instability)

Principle Physical Features

- Short stature
- Small, simplified ears
- Transverse palmar crease
- Fifth finger clinodactyly
- Wide space between first and second toe
- Hypotonia

Beautiful Faces



Craniofacial Characteristics

- Brachycephaly
- Microcephaly
- Maxillary hypoplasia
- Macroglossia
- Geographic tongue
- Small ears
- Over-folded hellices
- Epicanthal folds
- Up-slanting palpebral fissures

Cognitive Abilities

- Cognitive delay, usually in the mild to moderate range
- Broad variability across individuals
- Having Down syndrome doesn’t rule out other disorders (autism, ADHD, depression, etc.)

Speech Disorders

- More affected by an overriding neurological component characterized by:
  - Dysarthria
  - Dyspraxia
- Slurred speech pattern
- Increased rate of speech
- Increased incidence of stuttering

Communication Challenges

- Health
- Hearing
- Auditory Memory
- Articulation
- Language production
- Cognition

Communication Strengths

- Visual processing and memory
- Receptive language
  - Better than expressive language
  - Vocabulary stronger than syntax comprehension
- Gesture use
- Music and rhythm

Prader-Willi Syndrome

- Spontaneous genetic error on Chromosome 15
- Affects both sexes, all races

Prader-Willi Syndrome: Clinical Features

- Low muscle tone
- Short stature
- Small hands and feet
- Cognitive disability
- Problem behaviors (OC)
- Chronic feeling of hunger
- Cognitive impairment in the mild to moderate range

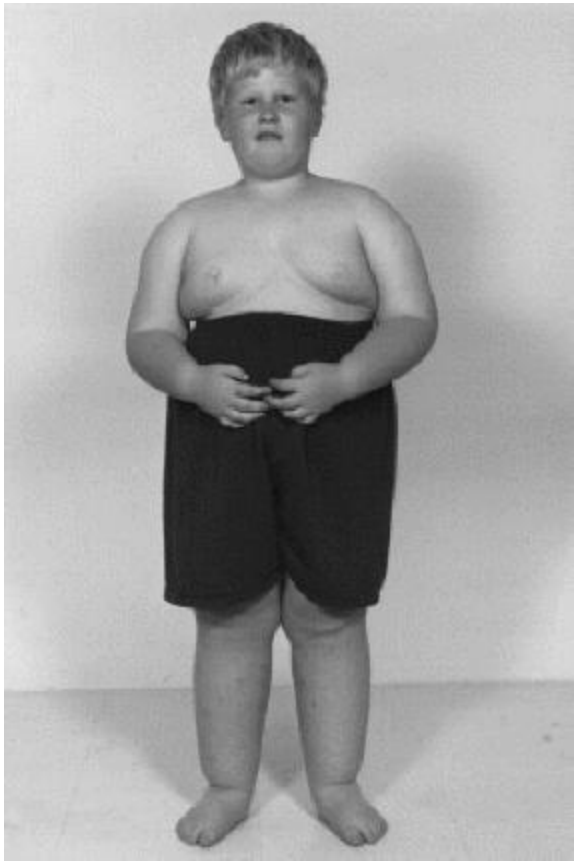
Craniofacial Features

- Bitemporal narrowing
- Thin upper lip
- Up-slanting almond shaped eyes

Feeding Disorders

- Neonatal period
  - Severe hypotonia
  - Poor suck
  - Poor feeding
  - Failure to thrive

Prader-Willi Syndrome



Rett Syndrome

Special thanks to:

Patricia Manning-Courtney, M.D.

Director, The Kelly O'Leary Center for Autism Spectrum Disorders

Cincinnati Children's Hospital Medical Center

Rett Syndrome – Physical Characteristics

- “Normal” development until 6 – 18 months old
- Loss of speech, hand skills, slowing of head growth, stereotypic hand movements
- Still a clinical diagnosis

Rett Syndrome – Necessary Diagnostic Criteria

- Apparently normal prenatal and perinatal history
- Development varies - normal through first 6 months or may be delayed from birth
- Normal head circumference at birth
- Post natal deceleration of head growth (in majority)
- Loss of achieved purposeful hand skill between 6 – 30 months
- Stereotypic hand movements
- Emerging social withdrawal, communication dysfunction
- Impaired or failing locomotion

Rett Syndrome – Physical/Behavioral Features

- Agitation
- Sleep disturbance
- Constipation
- Reflux
- Seizures
- Self injury

Fragile X Syndrome

Again, special thanks to:

Patricia Manning-Courtney, M.D.

Director, The Kelly O'Leary Center for Autism Spectrum Disorders

Cincinnati Children's Hospital Medical Center

Fragile X Syndrome – Physical Features

- Large head, prominent forehead
- Prominent ears
- High palate
- Soft skin
- Flexible joints, flat feet
- Macroorchidism (puberty)
- Long face

Language Characteristics

- Language impairment more severe than degree of cognitive impairment
- Frequent echolalia
- Off-topic/nonsensical utterances
- Telegraphic speech
- Lack of intonation
- Language patterns similar to individuals with ASD

Behavioral Features

- Cognitive delay (up to 80% of males)
- LD (girls)
- Attentional difficulties, hyperactivity
- Anxiety, obsessive/compulsive
- Sensory integration
- Autism

Waardenburg Syndrome

- Genetic disorder
- Two differently colored eyes
- Or brilliant blue eyes
- Distinctive hair coloring
  - Patch of white hair
  - Premature gray hair

Principle Features

- May include a side space between the inner corners of the eyes
- Low hairline
- Connected eyebrows
- Hearing loss ranging from moderate to profound

Four Types of WS

- Type I: widely spaced eyes; hearing impairment in 20%
- Type 2: other physical characteristics without widely spaced eyes; 50% have hearing impairment or are deaf

Smith-Magenis Syndrome

- Microdeletion on chromosome 17
- 1 in 25,000 live births
- Characteristics
  - Moderate to severe cognitive delays
  - Low muscle tone
  - Feeding difficulties
  - Failure to thrive
  - Frequent ear infections

Behavioral Characteristics

- Self-injurious behaviors
- Aggression
- Temper tantrums
- Hyperactivity
- Restlessness and distractibility
- Severe sleep disturbances
- Autistic like behaviors

Physical Features

- Flat, broad head
- Prominent forehead
- Heavy brows
- Up-slanting eyes
- Depressed nasal bridge
- Wide mouth with fleshy upper lip

Velo-cardio-facial syndrome

- **Cause:**
  - Genetic syndrome
  - 90 percent of patients with the features of this syndrome are missing a small part of their chromosome 22 at the q11 region

Features of Velo-cardio-facial

- palatal abnormalities (such as cleft lip and/or palate)
- feeding difficulties
- heart defects
- hearing loss or abnormal ear exams
- Renal anomalies (absent or malformed kidney)

VCF features (cont.)

- hypocalcemia (low blood calcium levels)
- microcephaly (small head)
- mental retardation (usually borderline to mild)
- IQs are generally in the 70 to 90 range
- psychiatric disorders in adults (e.g., schizophrenia, bipolar disorder)

Facial features of children with VCF syndrome

- Syndrome may include the following:
- small ears with squared upper ear
- hooded eyelids
- cleft lip and/or palate
- asymmetric facies
- small mouth, chin, and side areas of the nose tip

Turner Syndrome

Special thanks to:

Nancy Lanphear M.D.

Carol Forssell M.D.

Cincinnati Children's Hospital Medical Center

Turner Syndrome

- Chromosomal disorder in girls in which all or part of one X chromosome is missing
- Approximate incidence of 1 of 2000-5000 live female births

Turner Syndrome

- Sexual characteristics present but underdeveloped
- 90% exhibit normal intelligence
- Relatively normal Verbal IQ
- Lower Performance IQ
- Increased risk of Nonverbal Learning Disability
  Verbal/visual memory issues
- Attention difficulties
- Visual-spatial and visual-motor difficulties
- Hyperactivity early, hypoactivity later
- Problems with social relations, language

Original case described by Ullrich: Normal torso, head; unusually short arms and legs; webbed neck

Ears

- Simplified and/or prominent
- Increased incidence of Otitis media
  - due to a combination of small, dysfunctional Eustachian tubes and palatal dysfunction
- May require PE tubes
- Adenoids should not be removed in most cases, as worsens palatal dysfunction

Hearing

- Progressive sensorineural hearing loss
- 5% of children and 17% of adults have hearing loss enough to wear hearing aids

Feeding

Feeding issues are common initially: due to hypotonia of cheeks and lips

Effects of Hypotonia

- Difficulty latching on
- Difficulty sucking
- Poor chewing skills, aspiration
- Gastroesophageal reflux
- Failure to thrive

Treacher Collins Syndrome

Wide variability in features:

- downslanting palpebral fissures
- malar and mandibular hypoplasia, large mouth
- malformation of auricles
- cleft palate
- partial to total absence of lower eyelashes

Treacher Collins

- Autosomal, dominant
- Conductive hearing loss from ear anomalies
- Vision problems (coloboma, need for glasses, strabismus, ptosis, amblyopia)
- Can have learning problems, but MR in only 5%.
- Congenital heart disease
- Cleft palate and VP involvement

Neurofibromatosis

- Condition with skin findings (café-au-lait spots or neurofibromas)
- Neurofibromas can occur on the auditory nerve (acoustic neuroma) causing hearing loss
- Other tumors can occur in the brain in spinal cord

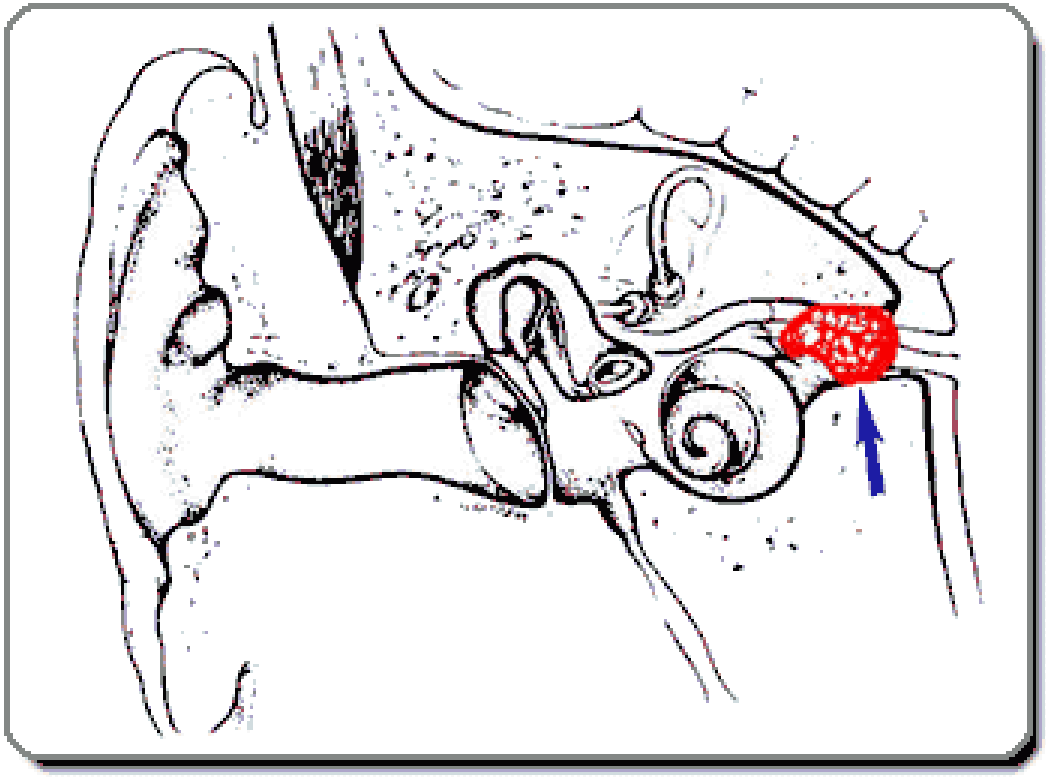


Figure B. A small acoustic neuroma (less than 1 centimeter in diameter) is typically confined the internal auditory canal.

Williams Syndrome

- Special thanks to:

Nancy Lanphear M.D.

Cincinnati Children's Hospital Medical Center

Clinical Features

- Facial features
  - broad smile
  - peri-orbital fullness
  - short nose with a broad tip
  - long philtrum
  - full lips and small chin
  - blue eyes
  - stellate pattern of the iris

Features continued:

- Short stature (compared to family)
- Congenital heart, blood vessel problems
  - Most common = Supravalvular aortic stenosis
- Friendly (distinctive) personality
  - Pragmatic Language Difficulty
- Developmental delay

Other Features

- Elfin-like facial features
- Hoarse voice
- Hernias
- Joint laxity
- Hypertension
- Hyperacusis
- Difficulty with fine motor, Hoarse voice
- Hypotonia
- Toe Walking
- Increased reflexes

CHARGE Syndrome

- A constellation of congenital malformations (genetic)
- Occurs in about one in every 9-10,000 births worldwide
- Very complex disorder involving extensive medical and physical difficulties
- Developmental skills vary widely from child to child
- Acronym no longer used to diagnose

CHARGE:

- The name of the condition is an acronym of some of the most frequent features:
- **C** = **C**oloboma (cleft) of the eye (80% of cases) and **C**ranial nerve abnormalities (CN I (smell), VII (facial palsy), IX, X (swallowing difficulties/aspiration)),
- **H** = **H**eart malformation (75% of cases),
- **A** = Choanal **A**tresia (blockage of the nasal passageways) (50-60% of cases)—unilateral or bilateral; bony or membranous,
- **R** = **R**etardation of growth after birth (87% of cases) and **R**etardation of development (94% of cases),
- **G** = **G**enital hypoplasia (underdevelopment) in males and females (75% of cases) and urinary tract malformations, and
- **E** = **E**ar malformations and/or deafness (88% of cases)—outer, middle, and inner ear

Other Features of CHARGE: consistent with CHARGE, possibly medically significant, but less helpful in making a diagnosis

- Chronic ear problems—OM, excess fluid, PE tubes (85%)
- CNS abnormalities (hydrocephaly, seizures, other abnormalities on scans)
- Hypotonia (90% of cases)
- Scholiosis—usually due to hypotonia

Cause

- Genetic condition, caused by a change (mutation) in a single gene, most often CHD7
- The gene test is very expensive and is not perfect--only about 2/3 of people with CHARGE have a positive gene test
- Therefore, a clinical diagnosis is still common (assessing major and minor features)

Communication

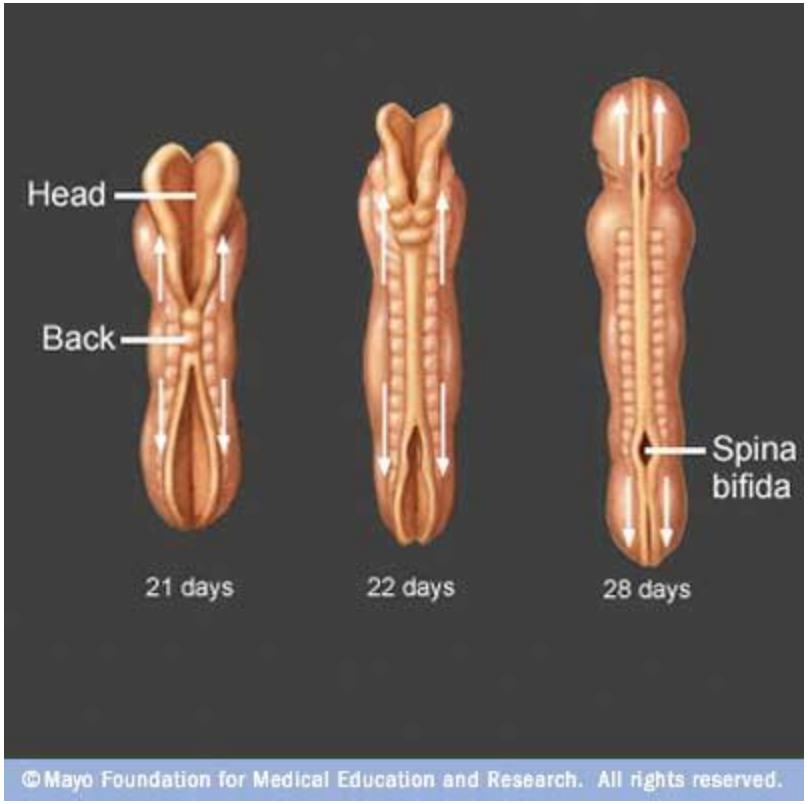
- Hearing loss
- Language delays
- Cognitive deficits
- Feeding deficits

CHARGE

Spina Bifida

- Most common neural tube defect
- Neural tube does not close (between 22 and 28 days after conception); part of spinal cord develops outside of vertebral column; large cyst may develop
- 1/1,000 children affected each year in US
- Affects more female fetuses than males
- Detected by early second trimester

What happens?



Types of Spina Bifida

- Anencephaly—most of brain above the brain stem does not develop; lethal
- Spina Bifida Occulta--mild and most common; hidden under skin; typically does not require treatment
- Spina Bifida Manifesta—rare; may be associated with nerve damage
  - Meningocele—fluid leaks from spinal canal, causing a swollen area over the baby's spine
  - Myelomeningocele—segment of spinal nerves push out of spinal canal

Prevention

- B-complex vitamin folic acid
- Diet—folic acid fortification added to enriched grains (flour, cornmeal, bread, pasta, rice, cereal)

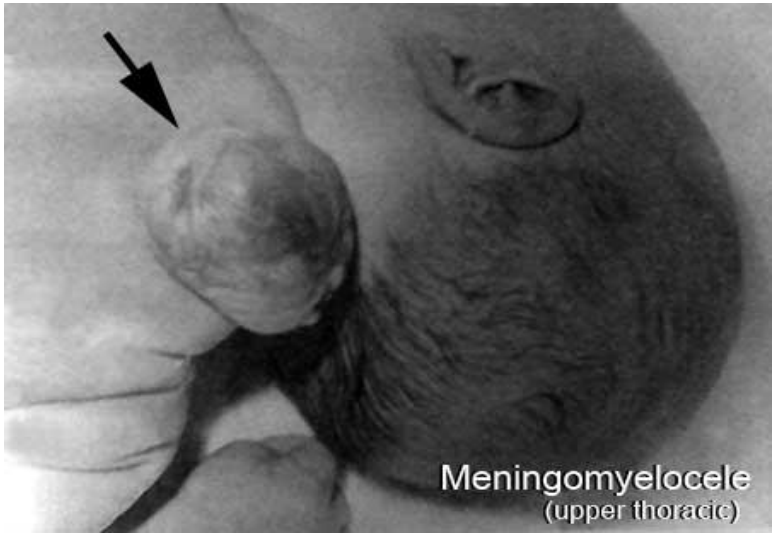
Detection

- Alpha-fetoprotein (AFP) in blood or amniotic fluid
- Ultrasound
  - 93% of NTDs are detected on an ultrasound

Communication

- Speech and language development may not be affected
- Feeding difficulties

Infants with Spina Bifida



Rubinstein-Taybi Syndrome

- Prevalence: One in 125,000; 1,051 cases reported internationally (CCDDD, 2005)

Characteristics and Prevalence of RTS

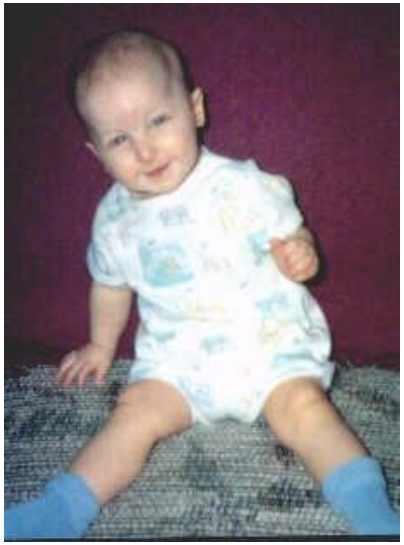
- Mental retardation
- Short stature
- Microcephaly
- Board thumbs and first toes
- Highly arched palate
- Hearing impairment
- Eye infections
- Feeding difficulties
- Intelligence: below average; range: 34-79
- Personality: “loving, friendly, and happy”

Cause of RTS

- Chromosomal deletion at 16p13.3
- Link between loss of aceytl transferase activity
- Mutation in CREB- binding protein gene (CBP) and EP300 gene (Roelfsema, et al., 2005)
- Sporadic mutation
- Within families; autosomal dominant (Marion, Garcia, & Karaski, 1993)

Communication of Individuals with RTS

- Delays and difficulties in speech and language
  - Speech, articulation, language, and fluency (Stevens, et al., 1990)
  - Speech, AAC, combination (Moore, Grether, & Creaghead, 2004)
- Limited number of communication partners in circles two (good friends), three (acquaintances), four (paid partners), and five (unfamiliar partners) (Moore, et al., 2004)



Pierre Robin Sequence

- It is not a syndrome; it is typically referred to as a sequence
- AKA: “Pierre Robin Malformation Sequence", "Robin Anomalad", and "Cleft Palate, Micrognathia and Glossoptosis”

Characteristics

- Small lower jaw (micrognathia)
- Tongue tends to “ball up” at the back of the mouth and fall back towards the throat (glossoptosis)
- Tongue may appear to be large in relation to jaw
- Breathing problems
- High-arched palate
- Horsehoe-shaped cleft soft palate may or may not be present
- Natal teeth (teeth that are present at birth)

Additional Symptoms

- Lack of facial expressions
- Impaired ability to suck
- Excessive drooling
- Crossed eyes
- Facial and limb deformities (club foot; missing or webbed fingers)
- Low muscle tone

Cause

- Remains unknown
- If a child is only affected by Pierre Robin, it is thought that it is the result of the fetus's positioning in the womb in the first weeks of development
- 15 to 17% of individuals with Pierre Robin have an isolated case

Children with Pierre Robin are at increased risk for the following:

- Feeding problems in infancy
- Ear infections
- Reduced hearing
- Approximately 40% of infants with Pierre Robin have Stickler Syndrome and about 15% have Velocardiofacial Syndrome

Possible treatments

- Surgery to repair the cleft palate
- Infant placed on stomach to keep tongue from blocking airway
- Special devices to protect the airway and aid in feeding
- Surgery to improve breathing
- Small jaw associated with Pierre Robin usually grows out on its own during the first two years, and usually no surgery is necessary on the jaw.

Communication

- Feeding difficulties
- Language deficits
- Articulation difficulties


Arthur Nouel

Fetal Alcohol Spectrum Disorders

- Prenatal exposure to alcohol can cause a range of disorders
- FASD—umbrella term describing the range of effects that can occur in an individual whose mother drank alcohol during pregnancy.
  - Includes fetal alcohol effects (FAE), alcohol-related neurodevelopmental disorder (ARND), and alcohol-related birth defects (ARBD)
- FAS—One of the most severe effects of drinking during pregnancy
- FAS is one of the leading known preventable causes of mental retardation and birth defects.
- All FASDs are 100% preventable
- FAS is a life-long condition

Characteristics of FASDs

- Mental retardation
- Birth defects
- Abnormal facial features
- Growth problems
- Problems with the central nervous system
- Trouble remembering and/or learning
- Vision or hearing problems
- Behavior problems (i.e. hyperactivity)
- Poor reasoning and judgment skills

Characteristics of FAS

- Abnormal facial features, growth deficiencies, and central nervous system (CNS) problems.
- Difficulties with learning, memory, attention span, communication, vision, hearing, or a combination of these.

Statistics

- Vary widely depending on populations studied and surveillance methods
- May range from 0.2 to 1.5 per 1,000 live births
- FASDs are believed to occur approximately three times as often as FAS.

Communication

- Language deficits
- Memory difficulties
- Cognitive deficits
- Hearing loss


YIELD

Resources

Syndrome Identification for Speech-Language Pathology

Robert J. Shprintzen

Smith's Recognizable Patterns of Human Malformation

Kenneth Jones

Questions and Discussion

Thank you for your attention!