Identification and Treatment Strategies for Children with Syndrome Disorders

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## General Characteristics

What do you observe in a child when you are performing an evaluation, an assessment or during a treatment session?

Why did I ask you that question??

## **Benefits: Visual Observation**

- Identify a typical patient
- Assess patient's state of health
- Identify patient's risk for genetic problems
- Compile a patient's phenotype
- Reassess appearance
- Long term treatment plan
- Collect research/evidence-based data
  - ► (Khan, 2000)

## Seeing and Observing

#### Seeing

Requires that you have adequate eyesight to view an object

Observing

Requires that you recognize, identify and analyze what you have seen

• (Khan, 2000)

## Physical Characteristics

What are some physical characteristics you observe in a child with a syndrome?



#### Face

#### Symmetry

- R and L sides of face same size and shape
- Bilateral structures located on same plane
- Bilateral structures same size and shape
- Mobility of moveable structures bilaterally similar

## Hair

Protocol for evaluating hair:

Amount of hair
Location of hair on neck, forehead, ears, eyes
Hair texture
Original color of hair
Hair shape
Hair growth pattern

## Hair

Amount: look at entire scalp; observe areas of sparse or missing hair

Location: flared eyebrows; eyebrows that meet at midline; unusually located hair (sideburns); absent lower and/or upper eyelashes

## Hair Texture: coarse; brittle; fragile

Original color: abnormal
 pigmentation (white forelock in dark hair)

Shape: curly; straight

Growth pattern: number and location of hair whorls

## Location of Hairlines

Location is influenced by presence and growth of the vertebrae, cerebral cortex, eyes and ears

Low posterior hairlines in persons with short necks and neck length depends on cervical vertebrae.

 Often seen in Down Syndrome, Fetal Alcohol Syndrome and others that have vertebral involvement

## Hairlines

- Forehead hairline: determined by brain growth and position of eyes
  - Central peak of hair extending downward into the forehead (widow's peak) with closely set or widely set eyes
- Hairline surrounding ears
  - Aberrant patterns associated with ear development
  - Two types: absence where hair should be and presence where hair should not be present.
  - Both conditions diagnostically significant for syndromes and hearing loss (Nager and Treacher Collins)

## Texture of Hair

- Diagnostic significance
  - Very coarse, sparse, brittle, fragile or fine must be evaluated against ethnic background, medical history and social preferences
  - Ask about original hair color: prematurely gray or white forelock in normally dark hair may be a clue in diagnosing hearing loss
    - Very blonde, almost silver hair is a characteristic of ectodermal dysplasia syndromes



An area that spirals downward from a central point

Normally have one hair whorl

Located several centimeters anterior to the posterior fontanel in infants



## Hair Whorl

Presence of more than one whorl, lack of a parietal hair whorl and midline or posteriorly located hair whorls are all diagnostically significant

Imply that brain development was anomalous

 Hair that grows straight upward and backward from the forehead also indicate anomalous brain growth

## Terminology: Hair

- Hypertrichosis: excessive amounts of hair and excessive hair follicles and must be confirmed by microscopic analysis
- Hirsutism: persons who have excessive body or facial hair in a male pattern (especially women)
  - Normal in some ethnic groups, but also characteristic feature in more than 20 syndromes

## Terminology: Hair

- Atrichia: congenital absence of hair and commonly occurs in ectodermal dysplasia syndromes along with an absence of teeth and nails
- Alopecia: had a normal hair pattern and later lost hair; characteristic of some syndromes but may result from other conditions
- Aplasia cutis: midline scalp lesions observed in neonates and associated with several syndromes
  - In adults and older children, lesions may have healed and be visible as bald areas of scar tissue



#### Aberrant hair growth is feature of:

#### Fetal Alcohol Syndrome

Prader-Willi Syndrome

Other syndromes

## Skin

- Protocol for observing skin
  - Examine for areas of hyperpigmentation and vitiligo
  - Presence and number of café' au lait spots
  - Presence, number, and location of vascular nevi
  - Addition of unusual features such as keloids, harmatomas or scars

# Skin Communication problems are components of many harmatoses

Sturge-Weber Syndrome

Neurofibromatosis

#### LEOPARD Syndrome

Ientigines (multiple black or dark brown spots on the skin, electrocardiographic conditions of the heart, ocular hypertelorism, pulmonary stenosis, retarded growth (short stature), deafness or hearing loss



Protocol for observing neck: Observe skin that covers the neck Observe the neck length

Determine if skin has normal attachment to shoulders

(Kahn, 2000)

## Neck

- Neck length is related to the structures and presence of the underlying vertebral column
- Short necks are usually caused by missing or malformed vertebrae
- Common syndromes with webbed necks and low posterior hairlines as a diagnostic feature
  - Noonan
  - Turner

## Ears

Should be approximately identical in appearance and placement

Løcation

--One end of a straight object (tongue depressor) at the outer canthus of the eye and extend it back toward the posterior part of the head

## Hands

- Hypoplastic (small) or aplastic (absent) fingernails are characteristic of more than two (2) dozen syndromes.
  - Can be symptoms of disease or trauma and used only to confirm a diagnosis, not as a diagnosis of the presence of the syndrome
- Cyanosis (blue of dark colored fingernails)
- Simian crease
  - Horizontal crease in the palm
  - Often seen in Down syndrome



## Ears

- Observe size, shape and symmetry of auricles
- Observe size of ear canals
- Observe location of outer ears
- Note auricular and preauricular anomalies
  - Line should intersect with the top third of the ear
  - Not uncommon to find some bilateral ear asymmetry in normal persons as well as in patients with syndromes

## Ears

- Preauricular pits or skin tags may indicate minor anomalies or may accompany microtia of the ear
- Lop ear may indicate congenital absence or hypotonia of one or more auricular muscles that hold ears in place





Observe eye orientation

Imagine a line extending through the palpebral fissures (space between eye lids) connecting the inner and outer canthi of the eye.

Line should be horizontal and parallel to the ground regardless of racial background.



Notice eye position
 Hypertelorism
 Hypotelorism

Observe to see if both eyes are present and are the same size

- Anophthalmia (absence of an eye)
- Microphthalmia (small eyes)
- Exorbitism (bulging eyes)



#### Presence of sclera

Sclera (covering tissue of eyeball)

Often thin and bluish in persons with ectodermal dysplasia syndromes

These syndromes are often associated with a number of communication disorders resulting from deafness, cleft palate and intellectual impairments.

## Eyes

- Persons with blue sclera may also have:
  - digital anomalies
  - microtia of auricles
  - absent eyelashes
  - sparse hair
  - thin or absent tooth enamel
  - hypoplastic fingernails,
  - wiry or brittle light colored hair
  - anodontia (missing teeth)

## Hands

- Observe size and symmetry
- Length of digits relative to palms
- Structure of digits
- Number of digits
- Presence, color and structure of fingernails
- Patterns of palms and fingertips

## Hands

- Hands can provide differential diagnostic clues between disease processes, trauma and syndromes.
- 14 phalanges in each hand approximately the same length
- Syndactyly (fingers are fused to one another)
- Polydactyly (extra digits)
- Triphalangeal thumb (extra phalanx and may give hand a five fingered appearance)

 Associated with syndromes that have sensorineural hearing loss as a component


# Known genesis syndromes

Chromosomal

Tetratogenic

Mechanically induced

# Known Genesis: Genetic

- Autosomal dominant
- Autosomal recessive
- X linked
- Contiguous gene
- Multifactorial inheritance
  - Interaction between genes and environment

Known Genesis: Chromosomal

- 22 pairs of chromosomes and x and/or y
- = deletion
- $\blacksquare$  + = addition
- $\mathbf{P}$  = short arm
- q = long arm
  - ►-5p; +21; 22q.5.137

Known Genesis: Chromosomal

- Monosomy
- Trisomy
- Mosaicism
- Translocation
- Chromosomal analysis
  - Karyotype
  - FISH (fluorescence in situ hybridization and identified in a cytogenetic lab where concern in structure and function of the cell, particularly chromosomes within the cell)

# Known Genesis: Tetratogenic

- Also called environmentally induced
  - Result of a direct or indirect action of an agent external to the genetic material of the fetus

Medicinal drugs, non-medicinal drugs and psychoactive substances (alcohol, cocaine, etc), viruses and some naturally occurring substances in the environment Known Genesis: Mechanically Induced

Presence of a tumor

Overcrowding in the womb

Tears in the amnion

Abnormal uterus

Unknown Genesis Syndromes

Provisionally unique

Recurrent pattern

# Important Definitions

- Association
  - associated factors/characteristics seen in an individual but not enough to call a syndrome or sequence
- Sequence
  - occurrence of multiple anomalies in a single individual where one of the anomalies has led to the development of all the others
- Syndrome
  - presence of multiple anomalies in a single individual with all those anomalies having one primary cause

# Role of SLPs and Audiologists

- The role of the audiologist or speechlanguage pathologist is to observe and document symptoms present.
- It is your role to look for possible symptoms. If you see one symptom, start looking for others. Once you find them, document them carefully.
- It is <u>not</u> the responsibility of the audiologist or speech-language pathologist to diagnose a syndrome. That is the realm of the medical profession.

#### **REMEMBER THIS**

A single sign or symptom does not an association, sequence, or syndrome make! Not all symptoms may be present at the same time. Syndromes are identified by their symptom complex.

#### Selected Syndromes

Stickler Syndrome

Fragile X

CHARGE

Velocardiofacial Syndrome

# Stickler Syndrome

- A connective tissue disorder that is one of the most common but is rarely diagnosed
- Second most common associated with cleft palate
- Caused by collagen gene mutations
- Variance of symptoms and different number of body systems affected
- Characteristics of symptoms vary
- Diagnosed by genetic evaluation

# Stickler SyndromeAutosomal dominant inheritance

Systems affected: vision, orofacial, hearing, bone and joints, speech and language (hearing, early feeding complications, resonance disorders, articulation errors, language development)

## Stickler Syndrome

- High frequency hearing loss
- Hypernasality/hyponasality
- Micrognathia
- Myopia
- Cataracts
- Glaucoma
- Lax joints

#### Two Types of Stickler Syndrome

Type I: Hereditary Progressive

Anthroophthalmopathy caused by gene mutation on Chromosome 12

Has vision joint, craniofacial, hearing disorders

Type II: caused by gene mutation on Chromosome 6

Has same problems as Type I but does not have vision problems



#### Stickler Syndrome—Biology

<u>https://www.youtube.com/wat</u> <u>ch?v=TqvdvKUtJVU#</u> Complications are difficulty breathing and feeding, blindness, dental and heart problems, and ear infections.





#### Case Study: CT

- No prenatal care
- Born at 5 lbs. 14 oz and considered premature but not sure of gestational age
- Born with cataracts and proptosis
- Placed with permanent guardians and was admitted to several hospitals for reflux, seizures, cleft of the soft palate, apena, breathing problems
- Diagnosed with Stickler Syndrome at CHOA

# C.T.

- Followed by Craniofacial Team for feeding, dental, vision, genetics, cleft of soft palate, hearing, nutrition
- Has had sets of PE tubes; hearing in normal range at present
- Cleft repaired at 7 months of age
- Has had 2 surgeries for cataracts and wears contacts
- Diagnosed with glaucoma in 2013
- Feeding difficulties: decreased chewing skills, decreased oral motor strength, pockets foods

## Initial Evaluation/Impressions

--Referred by Craniofacial Clinic of CHOA at 21 months of age

--Reported delays in crawling, walking, standing

--Babbled and used vocal inflections but no attempts at single words

--Testing revealed: severe/profound delays in global language skills

Initial Evaluation/Impressions-continued

- Given PLS-5 and Test of Early Communication and Early Language (TECEL)
- Auditory Comprehension: SS=57; Expressive Communication: SS= 60; Total Language: SS=55
- TECEL: Communicative Ability Index=0; Descriptive Term: Poor; Age Equivalent: 6 months to 6 months, 15 days
- Produces vowels and consonants--/p/,/b/,/m/,/d/; Reduplicated babbling; Inflectional patterns

#### Intervention Plan

- Vocalize for needs and wants
- Imitate vocalizations after clinician and family members
- Receptively identify objects and pictures of familiar items
- Imitate turn-taking games
- Eat foods without pocketing in the mouth.

- After 6 months----
  - Uses a variety of CV, VC, CVCV combinations
  - Verbalizes 20 words to request needs and wants
  - Follows 1 and 2 step directions
  - Understands and express functions of common objects
  - Use a variety of pragmatic functions in expressive language





Once vocalizations were consistent, true words emerged and word combinations were elicited.

Established routines

Used indirect language stimulation and facilitative play

Utilized hybrid approaches that were effective for improving vocabulary and phonological awareness

- Enhanced milieu teaching
  - Shown to be effective for those with at least 10 words, produce some verbal imitation and have a MLU of 1-3.5
  - Parent involvement/parent directed
  - Allows clinician to use imitation, prompting, and cueing during natural activities
    - In Paul, R. (2012). Language disorders from infancy to adolescence. Elsevier. St. Louis, pp. 73-76

 In addition to language goals and activities, began to work on phonological processes
Final consonant deletion

Once the concept of final consonant deletion was learned, speech was 75% intelligible.

- Testing before entering school on CELF-Preschool:
- Core Language: SS=86
- Receptive Language: SS=92
- Expressive Language: SS=88
- Language Content: SS= 87
- Language Structure: SS=90
- On CAAP: SS=87





#### Current Status

- Enrolled in special education preschool in the local school district at age 3
- Now at age 4 is in full time Pre-K
- Monitored at CHOA- No fistula in soft palate repair; Eye drops for diagnosed glaucoma; Allergy meds; Breathing treatments; Feeding issues resolved; Ear health monitored (PE tubes)

# Fragile X

- Most commonly inherited form of intellectual impairment with wide range of cognitive involvement from mild to severe
- May not show early dysmorphic features
- Verbal long term memory is a strength
- Better with gestalt processing rather than sequential processing
- Otitis media is common
- Low muscle tone may affect speech









# Typically developing Fragile X

# Early Markers

- Oral hypotonicity and sensory defensiveness
- Poor sucking and chewing
- Drooling
- Delayed onset and development of expressive language
Markers of Speech/Language

Speech is characterized by:
 Phonological errors
 Reduced intelligibility
 Fast, uneven rate
 Disturbed rhythm
 Hoarse and breathy vocal

quality

Some dyspraxic qualities



Poor planning, sequencing and execution of fluent speech

Trouble with formulating verbal responses on demand and answering direct questions

Word finding problems

Delayed syntax

# Speech/Language Issues

- Pragmatic issues
  - Gaze avoidance
  - Perseveration
  - 'Canned' phrases (Oh my, Oh my)
  - Lack of communication gestures
  - Poor topic maintenance
  - Poor turn taking

#### Intervention Suggestions

- Consider that females and males differ when planning activities/goals---
  - Females: low attention span, hyperactivity, poorer pragmatic abilities, less cognitive deficits (if any) and learning disabilities

Males: intellectually challenged; poor attention; hyperactivity, behaviors similar to those diagnosed with autism

# Suggestions

- Good language and behavioral models such as those found in mainstream classroom environment are particularly important for boys with Fragile X
- This makes use of their tendency to imitate what they see and hear and to prevent their imitating maladaptive behaviors







### CHARGE

#### C=Coloboma

- Cleft/abnormality of eye (iris, retina, choroids, disc)
- Microphthalmia (small eye)
- Cryptophthalmia (absent eyelids)
- May result in severe visual field/acuity deficits
- Cannot be corrected with surgery



#### C=Cranial Nerves

- I Olfactory absence of sense of smell
- VII Facial facial paresis, functional or structural asymmetry of the face
- VIII Auditory Sensorineural or mixed hearing loss
- IX Glossopharyngeal Swallowing difficulties, impaired gag reflex, VPI

#### H=Heart

- Congenital heart defects
- Valvular stenosis

# CHARGE

#### A=Atresia of choanae

- Atresia or stenosis of the choanae (passage from the back of the nose to the throat)
- May require multiple surgeries

#### R=Retarded growth and development

- Average size at birth but because of swallowing, heart and growth hormone deficiency many children will become below average in growth and development
- Delayed in developmental milestones due to sensory and visual impairments
- Mental Retardation



#### G=Genital and urinary abnormalities

- Females may have underdeveloped labia
- Males may have a micropenis or undecended testes
- Both sexes may lack pubertal development and have urinary tract or kidney abnormalities

# CHARGE

#### E=Ear anomalies/hearing loss/deafness

- Anomalies of the stapes and incus with ossicular chain fixation
- Absence of the stapedius muscle, oval window, and semicircular canals
- Short, wide ears with small or absent lob
- Hearing loss range from mild to profound conductive or sensorineural

# Major Characteristics

- Coloboma
- Heart Malformations
- Ear Anomalies
- Mental Retardation
- Deafness

#### Minor Characteristics

Face: square with broad prominent forehead, arched eyebrow, ptosis (drooping eyelids), prominent nasal bridge, flat mid face, small mouth

- Hand –small thumb, short fingers, broad palm
- Hypotonia

### Minor Characteristics

- Tracheoesophageal fistula
- Sloping shoulders
- Oro-facial cleft: palate, submucous cleft and cleft lip
- Esophageal atresia
- Webbed neck
- Behavior problems

# Intervention Considerations

- Articulation/Phonology
  - Secondary to featured deficits and may be a challenge
  - Typical error patterns are devoicing, final consonant deletion, difficulty with plosives and substitutions
  - May be approached through phonological intervention and may need compensatory strategies
    - Richard, G. and Hoge, D. (2000)

# Intervention Considerations

#### Resonance and Voice

- Vowel prolongations for voicing and then to words, phrases, sentences and conversation
- Sustained respiration
- High pressure consonants for vp closure
- If structural deviations have been surgically corrected, teach nasal air flow vs. oral airflow
  - Richard, G. and Hoge, D. (2000)

# Intervention Considerations

- Language and Hearing
  - Delay is common
  - May consider extensive vocabulary intervention (receptively and expressively) along with basic concepts
  - Influenced by cognition, vision, hearing
  - Hearing aids /visual and tactile systems

#### Intervention Comparisons

M.I.
Characteristics
Interventions

Status

- S.A
- Characteristics
- Interventions
- Status

- AKA: DiGeorgeSyndrome-Shprintzen Syndrome
- Fourth most common birth defect
  - 5% of all patients in cleft palate clinics have syndrome
- Occurs in 1 out of every 700 live births
- Currently, 1 in every 2000 persons have the disorder

- 22q11.2 deletion
  - Deletion on band 11 of long arm
- Highly variable with more than 185 associated features
- Most common anomalies are palatal anomalies and vp insufficiency, heart defects and dysmorphic facial features

- Most common characteristic: velopharyngeal insufficiency (about 20% in the absence of a cleft palate)
  - Difference between vp insufficiency, vp incompetence, vp inadequacy, vp mislearning
- Vascular anomalies
  - Medial displacement of carotid arteries

#### Pictures:





VCFS: Speech-Language Characteristics

- Hypotonia
- Oral apraxia
- Problems with feeding: sucking, chewing and swallowing
- Articulation disorders common
- Hypernasal speech
- Poor social interactions

# VCFS: Communication Issues

- Some type of communication disorder is present in nearly all cases, including:
  - Hypernasality
  - Articulation
  - Language disorders
  - Voice disorders
  - Resonance disorders

VCFS: Speech-Language Characteristics

- Abnormal palatal function
- Conductive, sensorineural or mixed hearing loss
- Language-learning disabilities
   Reading comprehension, math concepts, generalized problem solving



Kummer, 2001

#### Developmental disabilities

Intelligence in low to normal range, but mild to moderate intellectual problems are common

Learning disabilities: reading comprehension and extemporaneous speech; become obvious around 7 or 8 years of age

#### Educational goals

- Focus on language and communication skills
- Focus on social/pragmatic abilities

#### Behavioral problems

As age increases, psychiatric problems such as schizophrenia, bipolar illness and depression

#### Intervention



Referred at 20 months by parents with concerns of low frequency of vocalizations, jargon, and a few true words

Decided to pursue private therapy because of distance of driving and was provided with indirect language stimulation techniques to use at home; Referred to pediatric otolaryngologist G.T.

Returned to clinic at age 4 with receptive language at age level, expressive language at 1 S.D. below the mean, moderate phonological disorder and hypernasality and nasal flaring

► Age 4-5:

Phonological therapy: Achieved 40-100% on selected processes (f,l,s) Resonance: 55% in single words with no nasal consonants ► Age 5-6; 6-7; 7-8

G.T.

- Clinicians changed
- Medical referral to ENT and Craniofacial Clinic;VP insufficiency confirmed and VCFS diagnosis; Pharyngoplasty
- Achieved 84-100% accuracy on artic placement and 84% air flow for vowels
- Achieved correct placement by age 7 ½ and goals for artic discontinued

G.T.

- Achieved 75-90% correct airflow for high pressure consonants in conversational speech
- Hypernasality significantly decreased
- Social/emotional behaviors noteddepression, lack of appetite;
   Medical consultation concerning
   ADD and diagnosed; Academic difficulties
- Received services elsewhere--moved

# Treatment Approaches Clinican directed Drill

Drill play

Modeling

# **Treatment Approaches**

#### Child centered or patient centered

- Indirect language stimulation
  - Self/parallel talk
  - Extensions
  - Expansions
  - Imitations
  - Build-ups and Breakdowns
  - Recasting

# **Treatment Approaches**

#### Hybrid

- Milieu teaching
  - Applies operant principles to quasinautralistic settings
  - Make use of prompts, cues and extrinsic reinforcement but do so in interactive activities that have been carefully arranged
  - Incidental teaching, mand, mandmodel, time-delay

# CHALLENGE

#### Add tools to your toolbox

- Observe differently
- Utilize new knowledge
- Network

#### Branch out and try something you've learned today!