Speech and Language Issues in Galactosemia

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Introduction

- Only in recent years have we come to appreciate persistent complications in treated galactosemics.
- Multiple forms and symptoms.
- A common problem, among even those patients treated from birth, is speech disturbance
- 90% of galactosemic children have articulation and vocabulary difficulty.
- Dyspraxia is the most prominent speech problem, also multiple forms and symptoms
- Complex metabolic disease and a complex speech disorder which by itself is poorly understood

Overview

- Review of discovery of coincidence of galactosemia and dyspraxia
- "Psycholinguistics 101"-how we speak
- Discussion of dyspraxia
  - Definition and terminology
  - Controversy re: cause
  - Assessment
  - Treatment

Speech and language deficits in early-treated children with galactosemia

Waisbren SE, Norman TR, Schnell RR, Levy HL. J Pediatr 102:75, 1983

- 8 children, 3.6-11 yr olds studied.
- Expressive language deficits in 7 of 8, with immediate recall and word retrieval skills notably affected.
- Articulation deficits present in 5 of 8.
- Receptive language intact.

Verbal dyspraxia in treated galactosemia

Nelson CD, Waggoner DD, Donnell GN, Tuerck JM, Buist NRM. Pediatrics 88:346, 1991.

- A *specific pattern* of speech/language difficulties in galactosemia was described.
- 24 patients studied. 54% had verbal dyspraxia.

Nelson, et. al. (cont.)

- Occurrence of dyspraxia not correlated with age at Dx, severity of symptoms in newborn period, or biochemical control.
- IQ of patients with dyspraxia was lower than patients without dyspraxia (79 vs 99).
- Not a typical finding in patients with dyspraxia who are not affected by galactosemia

Outcome Analysis of Verbal Dyspraxia in Classic Galactosemia

Robertson, et al., Genetics in Medicine 2:2, 2000

- 43 of 113 patients with classic galactosemia had "speech problems"
- Used a questionnaire completed by speech clinicians
- Conclusion that patients homozygous for Q188R mutation are at significant risk for having dyspraxia

Verbal Dyspraxia and Galactosemia

Webb, A.L., Pediatric Research 53:3, 2003

- Evaluation of five biochemical risk indicators for verbal dyspraxia in galactosemics
- 15 of 24 patients with speech evaluation had verbal dyspraxia.
- Concluded that a breath testthat measures galactose metabolism is a sensitive predictor for verbal dyspraxia.

Summary of Incidence

- 38% to 63% of galactosemic patients have verbal dyspraxia
- 90% have some speech language difficulties

Psycholinguistics 101 how we speak

- Intention or idea
- Word selection
- Phoneme selection
- Motor planning for word production
- Execution of motor plan
- Auditory feedback
- Revised motor plan if incorrect production

Psycholinguistics 101

- Praxis: Learned ability to plan and direct a temporal series of coordinated movements to achieve a result.
  - Motor planning for word production
  - Execution of motor plan

Motor Planning

- The first part of praxis
- Ability to relate a series of movements or motions to each other
- For speech, ability to coordinate movement for sound production into sound pattern for word production
- Learned skill, instantaneous, distinctly human and miraculous!

Say "plan"

- Open the larynx
- Draw diaphragmatic musculature downward to inhale
- Close the lips
- Exert pressure via diaphragm elevation to build up pressure behind the closed mouth
- Explode air through lips without using voice for /p/

- Concurrently, tongue tip lifted for /l/
- Close larynx for voicing, while continuing airflow from lungs
- Tongue to mid-level of mouth, elevate soft palate, tongue tip lowered for /a/
- Finally, tongue tip elevated, soft palate lowered, continued voicing for nasal sound /n/
- Sensory feedback (proprioception)

Verbal dyspraxia

- Impaired motor planning for voluntary production of speech sound sequences
- Sensory motor disturbance, impaired feedback loop?
- Failure to plan, sequence and carry out movements necessary for speech
- An isolated disorder or part of a syndrome

Features of Verbal Dyspraxia

from Nelson, D, Eur J Ped, 1995

- Groping to achieve target
- More difficulty with increased length and complexity of words.
- vowel errors
- Switching sounds and syllables in a word (nose- "zone" , music- "musik")

Features of Verbal Dyspraxia (2)

- Poor rapid repetitionof oral movements
- Poor auditory memory span
- Reordering or substitution of words during repetition of short word list.
- Prosodic disturbances (stress and rate).
- Highly inconsistent errors
- Awareness of error but unable to correct it.

Additional Features

- Significant delay in speech and language development
- Normal or higher ability to understand language
- Increased likelihood of needing other school programs-reading, spelling, writing
- Limited progress with conventional speech therapy

Controversy about Verbal Dyspraxia

- Motor planning and /or linguistic etiology
- Few empirical studies as to the origin and nature of developmental apraxia
- Several subtypes of developmental apraxia not widely accepted.
- Diagnosis critical; often recalcitrant to standard speech treatment, requires specific intervention.

Verbal dyspraxia/Apraxia of speech/ Developmental apraxia of speech

- 3 terms, one definition
- Acquired vs. developmental have etiological differences
- Infrequently, oral apraxia
- Apraxia, more severe. Not uniformly accepted term
- SLPs use apraxia, MDs use dyspraxia

Assessment: Diagnosing Dyspraxia

Early Motor Control Scales (Hayden,et al., 2002)

- 9months – 2 years
- Natural adult-child interactions used to measure motor control to support speech

Assessment (2)

- Kaufman Speech Praxis Test (Kaufman, N., 1995). Ages 2 years to 6 years. Nice progression from motor to simple speech to complex speech tasks.
- Apraxia Profile (Hickman,L., 1997)
- Verbal Motor Production Assessment for Children (Hayden, D., 1999). Ages 3 years to 12+ years. Adds gross motor control to the above.

Assessment (3)

- Assessment of hearing
- Language assessment

Results of assessment

- Oral motor deficits in speech production
- Disorganization of the sound system
- Disorganization of the expressive language system
- Some or all of the above

Treatment of Oral motor deficits

- Goal: help child know where they are in space, gain volitional control of speech movements
- Intensive systematic drill of movements for speech
- Therapist experienced wth oral motor problems, hands-in-the-mouth approach
- Oral sensation, positioning work-brushes, textures
- Multisensory approach –visual, visual auditory, tactile feedback
- Progression from single sound to simple syllables to varied vowels. Ex: tip elevation to /ta/ to /ta,ti/

Treatment of Sound problems

- Development of core vocabulary for repetitive drill of useful words and phrases
- Focus on vowel and diphthong production – can greatly increase intelligibility, "earl" for "oil"
- Sound contrasting-pairing of "bood" with "boot" for auditory discrimination

Treatment of Expressive Language Problems

- Sentence or phrase building
- Target words from sound development for sentence building
- Find the context where sound production helped or hindered
- Move to story telling with core vocabulary
- Use of mastered phrases for drill with varied intonation & rhythms
- Instead of asking to stick out tongue, lick lollipop
- Good review of many treatment techniques well referenced, www.apraxia-kids.org

General Principles of Treatment

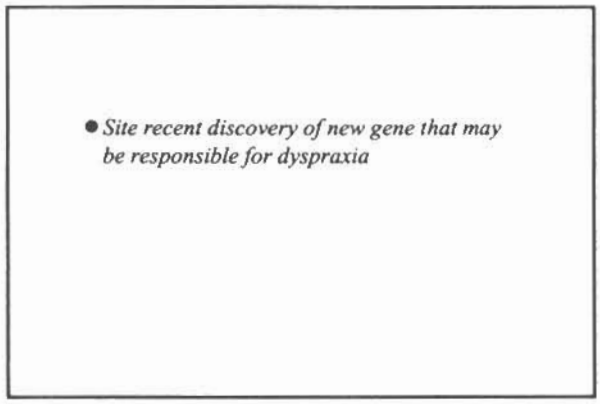
- Establish control of speech movement system from breathing to jaw and tongue movement
- Kinesthetic awareness during speech sequences
- Regulate rate and melodic flow of speech
- Visual feedback helps greatly

Rhythm, rhyme and verbal apraxia

- "sounds like a foreign accent" in early descriptions-prosodic differences
- Disturbance of stress patterns may be diagnostic
- Perception of rhyme may be diagnostic
- Does the stress pattern of spoken language help infants to organize and learn language?

Ideas for parents

- Educate yourselves about dyspraxia
- Advocate for your child –bring this info to your therapists and school staff
- Early intervention
- Read, sing, play games!
- Emphasize sequences, visual & auditory, tactile
- Pair motor rhythms with auditory


• Site recent discovery of new gene that may be responsible for dyspraxia