Speech and Language Issues in Galactosemia

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Introduction
• Only in recent years have we come to appreciate permanent complications in treated galactosemia.
• Multiple forms and symptoms.
• A common problem, among even those patients treated from birth, in speech disturbance.
• 90% of galactosemic children have articulation and vocabulary difficulty.
• Dyspraxia is the most prominent speech problem, along multiple forms and symptoms.
• Complex metabolic disease and a complex speech disorder which 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
Hackett BE, Horvath PK, Schach M, Sara ML, Polymer B (1979) 1979
• 8 children; 3-6 1/2 yrs old studied.
• Expressive language deficits in 7 of 8, with immediate recall and word retrieval skills notably affected.
• Articulation deficits present in 5 of 8.
• Reception language intact.

Verbal dyspraxia in treated galactosemia
• A specific pattern of speech-language difficulties in galactosemia was described.
• 24 patients studied; 54% had verbal dyspraxia.

Nelson, et. al. (cont.)
• Observed dyspraxia not correlated with age or IQ, severity of symptoms in over 50 patients, 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
- 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
- Evaluation of five biosimilarity risk indicators for verbal dyspraxia in galactosemic patients
- 13 of 24 patients with speech evaluation had verbal dyspraxia
- Concluded that a breath test that 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 patterns for word production
- Taught skills, 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 /v/
- Close larynx for voiced, while continuing airflow from large
- Tongue to mid-level of mouth, elevate soft palate, tongue tip lowered for /w/
- 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

- Gripping to achieve target
- More difficulty with increased length and complexity of words.
- Vowel errors
- Switching sounds and syllables in a word (e.g., "zone", music "mush")

Features of Verbal Dyspraxia (2)

- Poor rapid repetition of 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

- 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)

- Smooths – 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 with oral motor problems, hands-on approach
- Oral sensation, positioning, workbrush, textures
- Multisensory approach—visual, visual auditory, tactile feedback
- Progression from single sound to simple syllables to varied vowels. Ex: ‘tip’ deviation to ‘be’ to ‘face’

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 “boot” with “food” 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 intonations & rhythms
- Instead of asking for“lick” or“kiss”, ask for“lollipop”
- Good review of many treatment techniques can be found at: www.aphasiakids.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

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Recent discovery of new gene that may be responsible for dyspraxia