Introduction

Galactosemia is a rare metabolic disease (1 in 40,000) which is fatal if not detected during newborn screening. People with galactosemia cannot metabolize galactose, a milk sugar found in human and animal milk. Galactose is needed for myelin production. Galactosemia can result in abnormal white matter formation on nerves and in brains. 90% of children with galactosemia have language disorders and 60% have speech disorders, frequently diagnosed as childhood apraxia of speech.

Lack of knowledge about the ramifications of galactosemia frequently results in the denial of services for children with galactosemia.

Questions

Q1. What are the characteristics of the persistent speech, language, and cognitive impairments in children with galactosemia?

Q2. What is the relationship between speech, language, and cognition in children with galactosemia?

Participants

• 14 children with classic galactosemia and speech sound disorders
• Ages 4-14
• Recruited through Parents of Galactosemic Children organization
• Tested in their homes

Methods

Two 60-minute sessions:

1. Standardized tests
   • Goldman-Fristoe Test of Articulation-2 (GFTA)
   • Oral and Written Language Scale (OWLS)
   • Kaufman Brief Intelligence Test (K-BIT)

2. Conversational language sample analyzed with:
   • SALT
   • PEPPER (phonetic transcription
   • Challenging word tasks

Results

Q1. Cognition (full-scale IQ scores)
   • 43% scored below normal range (65-82)
   • 57% scored within normal range (100-111)
   • No children scored in the low normal range

Language

OWLS-36% below 1.5 SD
SALT-64% below 1.5 SD

Analysis revealed significant deficits in:
• More omissions (words and bound morphemes) as utterance length increased
• Fewer words/minute
• Fewer # of root words
• Mean of 4.5 words or less/C-unit across ages (100% of students below the mean)

Speech

GFTA -79% below 1.5 SD
SALT-Mean intelligibility was -3.5 SD

• More speech sound errors as utterance length increased
Consonant errors
• Epenthesis
• Metathesis
• Assimilation
• Voicing/devoicing
• Excessive or weak force
Vowel errors
• Centralized or backed

Q2. Correlations

IQ related only to MLU and performance on OWLS
IQ NOT related to intelligibility, articulation, speech rate, word and morpheme omissions, or vocabulary

When speech rate and turn length increased, more words and bound morphemes were omitted and speech errors increased

Conclusions

Children with galactosemia:

1. Show increasing speech AND language errors as length and complexity of utterance increases.

2. Continue to use slow and short utterances into the adolescent years due to breakdowns in speech and language with increasing utterance length.

3. Need to be tested for speech and language impairments using connected speech in order to evaluate the frequent and unusual errors.

4. Children who are evaluated using standardized tests only may be denied services even when significant impairments exist.

Future Directions

Examine the differences in outcome measures related to metabolic efficiency due to genotype

Examine differences in white matter formation using neuroimaging

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