Childhood Apraxia of Speech in Children and Adolescents with Galactosemia

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First Hour

Contemporary Research in CAS: Shriberg

- Premises in Three Contemporary Areas of CAS Research
- Premises in a Neurodevelopmental Framework for CAS Research
- Assessment and Analytic Methods in the Present Study

Galactosemia: Potter

- Overview
- Data Collection Methods in the Present Study

Diagnostic Classification of CAS: Strand

- Contemporary Classification Criteria
- Diagnostic Classification Methods in the Present Study

Second Hour

Findings: Shriberg

Discussion: Shriberg, Potter, Strand

Open Discussion

Contemporary Research in CAS: Some Conclusions from the ASHA 2007 CAS Position Paper

There is emerging consensus (but not universal agreement) that CAS[a]:

- is a valid clinical entity
- is neurologically based (congenital or acquired)
- is associated with increased risk for language delay

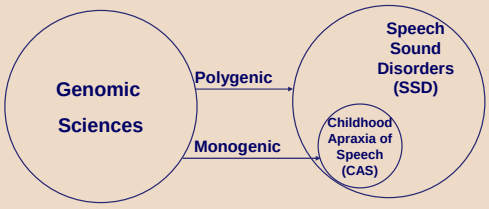
[a]Developmental Verbal Dyspraxia (DVD) continues to be the classificatory term used in medical literatures and in many countries.

Premises In Three Areas of Contemporary CAS Research

- **Premise in Genomic Research**
- **Premise in Motor Speech Research**
- **Premise in Assessment/Treatment Research**

Premises in Contemporary CAS Research

The Distal Substrates of CAS and Other Subtypes of SSD Can Be Identified Using Current and Emerging Methods in the Genomic Sciences



Premises in Contemporary CAS Research

The Distal Substrates of CAS and Other Subtypes of SSD Can Be Identified Using Current and Emerging Methods in the Genomic Sciences

- Molecular genetic and neurocognitive publications on the London 'KE' family (beginning with Hurst et al.,1990) catalyzed worldwide interest in speech- language genes in many scientific disciplines
- Motor speech and verbal trait disorders associated with *FOXP2* disruptions in the KE family confirmed in four other families (MacDermot, 2005; Shriberg et al., 2006; Zeesman et al., 2006)
- First functional genomic analysis of *FOXP2* (Vernes et al., 2006), additional function analyses, and analyses linking *FOXP2* to *CNTNAP2* (Vernes et al., 2008), a gene associated with language disorder and autism

Premises in Contemporary CAS Research

The Distal Substrates of CAS and Other Subtypes of SSD Can Be Identified Using Current and Emerging Methods in the Genomic Sciences

- Continuing functional analyses of *FOXP2*, including knockout, knockdown, and knockin Foxp2 orthologs in vertebrate species (fish, reptiles, birds, mammals)
- Continuing studies of *FOXP2* as the first causal gene implicated in the evolutionary biology of speech-language in humans

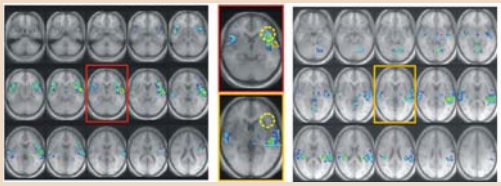
Premises in Contemporary CAS Research

The Proximal Substrates of CAS and Other Subtypes of SSD Can Be Identified Using Current and Emerging Technologies in the Neurosciences

- What are the neural substrates of the speech processing disruptions in CAS — at successive neurodevelopmental periods?
- Which processes in speech production (e.g., encoding, memorial, planning/programing) are disrupted in CAS — at successive neurodevelopmental periods?
- Are the underlying processing disruptions in CAS domain-specific (i.e., limited to speech) or are they domain-general — at successive neurodevelopmental periods?

Premises in Contemporary CAS Research

The Proximal Substrates of CAS and Other Subtypes of SSD Can Be Identified Using Current and Emerging Technologies in the Neurosciences



The activation pattern for an individual SSD participant with a history of CAS (Right; yellow box) during the repetition of a nonword fMRI task. The random effect result of the control group is shown on the left (red box) . Yellow circle indicates Broca's area. (Image Right=Brain Left)

From Lewis et al. (2008)

Premises in Contemporary CAS Research

Neurobiological and Behavioral Research in CAS Will Lead to Effective Assessment and Treatment Methods

- Research Epochs Toward Personalized Medicine
  - Decade of the Brain
  - Postgenomic era
  - Next-generation sequencing
- Comparative Effectiveness Research

  ". . . pharmacogenomic strategies to investigate the efficacy and safety of drugs in genomically-defined subpopulations." (Collins, 2009)
- Evidenced-based Assessment and Treatment Research

Classification of SSD in DSM IV

(Science, October 23, 2003, p. 809)

Today's *DSM* Structure

Axis I: Clinical syndromes and disorders
Classic psychiatric disorders such as depression or complaints such as relationship problems

Axis II: Personality disorders and mental retardation
Disorders characterized primarily by long-standing traits

Axis III: General medical conditions
Any nonmental disorder that might influence mental health

Axis IV: Psychosocial problems
Includes loss of job, homelessness, and other factors that contribute to the other axes

Axis V: Global assessment of functioning
An overall rating of the patient's social, professional, and psychological abilities

Classification of SSD in DSM V?

(Science, October 23, 2003, p. 809)

One View of *DSM*'s Future

Axis I: Genotype
Genes linked to diseases, symptoms, resiliency, and drug response

Axis II: Neurobiological phenotype
Cognitive abilities, emotional regulation, brain-imaging profile, and other qualities

Axis III: Behavioral phenotype
Expression of disease-related behaviors, including their range and frequency

Axis IV: Environmental modifiers or precipitants
Environmental factors that alter the neurobiological or behavioral phenotype

Axis V: Therapeutic targets and response

Constraint Common to All Contemporary Research in CAS: Lack of One or More Validated Diagnostic Markers

The "Circularity" or "Tautological" Problem[a]

"On what bases do you select subjects for study when trying to identify and characterize a new clinical entity? Without established inclusional and exclusional criteria, derived from careful experimentation, usually accumulated over a long period of time, and without models that specify the levels of breakdown and the potential mechanisms responsible for the phenomena . . . it is difficult or impossible to have confidence that the individuals and groups actually represent the subject of interest. In other words, it is difficult or impossible to avoid experimental tautologies. (p. 315)"

McNeil, M. R., Robin, D. A., & Schmidt, R. A. (1997)

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Premises in a Neurodevelopmental Framework for CAS Research

Progress in All CAS Research Requires an Etiology-Based Classification System for Speech Sound Disorders of Currently Unknown Origin

No.	Type	Subtype	Abbreviation	Risk Factors	Processes Affected
1	Speech Delay	Speech Delay–Genetic	SD-GEN	Polygenic/ Environmental	Cognitive-Linguistic
2		Speech Delay– Otitis Media with Effusion	SD-OME	Polygenic/ Environmental	Auditory-Perceptual
3		Speech Delay– Developmental Psychosocial Involvement	SD-DPI	Polygenic/ Environmental	Affective-Temperamental
4	Motor Speech Disorder	Motor Speech Disorder– Apraxia of Speech	MSD-AOS	Monogenic? Oligogenic?	Speech-Motor Control
5		Motor Speech Disorder– Dysarthria	MSD-DYS	Monogenic? Oligogenic?	Speech-Motor Control
6		Motor Speech Disorder- Not Otherwise Specified	MSD-NOS	Monogenic? Polygenic? Oligogenic? Environmental?	Speech-Motor Control
7	Speech Errors	Speech Errors-Sibilants	SE-/s/	Environmental	Phonological Attunement
8		Speech Errors-Rhotics	SE-/r/	Environmental	Phonological Attunement

Six of Eight Proposed Subtypes of Speech Sound Disorders of Currently Unknown Origin

Speech Delay (SD) due to:

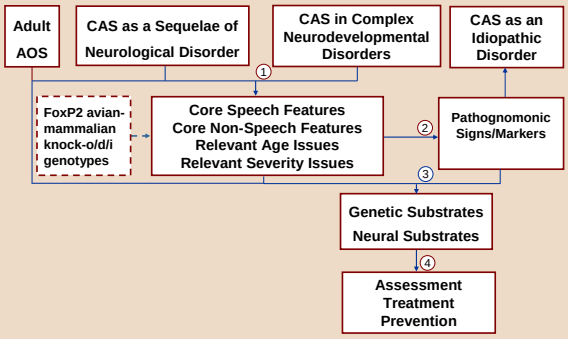
- a genetically-inherited verbal trait disorder
- fluctuant hearing loss associated with early recurrent otitis media with effusion
- developmental psychosocial involvement

Motor Speech Disorder (MSD):

- apraxia
- dysarthria
- not otherwise specified

Premises in a Neurodevelopmental Framework for CAS Research

Diagnostic Markers of CAS in Idiopathic Contexts Can Be Informed by Studying CAS in Neurological and Neurodevelopmental Contexts



Some Complex Neurodevelopmental Disorders Reporting 'Significant' Speech Delay/CAS

- Autism
- Chromosome translocations
- Coffin-Siris syndrome (7q32-34 deletion)
- Down syndrome (Trisomy 21)
- Fragile X syndrome (*FMR1*)
- Galactosemia (9p13)
- Joubert syndrome (*CEP290*; *AHI1*)
- Rett syndrome (*MeCP2*)
- Rolandic Epilepsy
- Russell-Silver syndrome (*FOXP2*)
- Velocardiofacial syndrome (22q11.2 deletion)
- Williams-Buren region microduplication (7q11.23)

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Premises in a Neurodevelopmental Framework for CAS Research

Progress in CAS Research Requires a Standardized Assessment Protocol and a Standardized Suite of Analytics

Standardized Assessment Protocol

The Madison Speech Assessment Protocol (MSAP)[a]

- Scores/subscale scores on all MSAP tasks
- Reference sample of 4-to16-year-old typically developing speakers assessed with the MSAP

Standardized Suite of Analytics

Competence, Precision, and Stability Analytics (CPSA)[a]

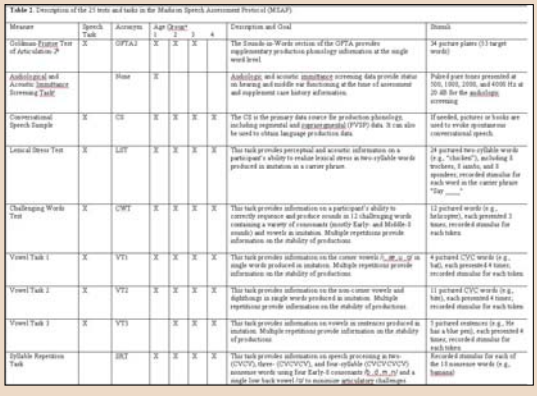
- Currently includes 84 indices/subindices and markers/submarkers
- Indices and markers derived from 1-8 MSAP tasks
- Current criteria for a 'Positive' index/marker: z-score < 1 standard deviation from the mean of typically-developing speakers of same age and sex

[a]Shriberg, Fourakis, et al., 2009

Madison Speech Assessment Protocol (MSAP): Description

- **Four age-based protocols**
  Preschool, school-aged, adolescent, adult
- **Each protocol includes 15 tasks assessing speech, voice, and prosody**
- **About 1 hour to administer**

- Articulation Task
- Challenging Word Tasks (3)
- Challenging Phrase Task
- Consonant Task
- Conversational Sample
- DDK Task
- Phonation Task
- Syllable Repetition Tasks (2)
- Stress Tasks (2)
- Vowel Tasks (3)



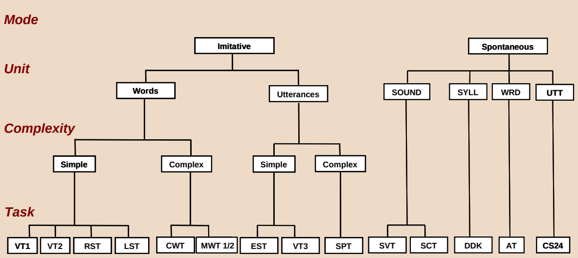
Nonword Repetition Task		NRT	X	X	X	X	This task provides information on speech processing using nonsense words.	Recorded stimulus for each of 16 nonsense words — four each of 1-syllable, 2-syllable, 3-syllable, and 4-syllable words (e.g., [illegible])
Emphatic Stress Task	X	EST	X	X	X	X	This task provides information on a participant's ability to realize emphatic stress within short sentences. In each of the four trials for each sentence, a different word is stressed.	Recorded stimuli for two 4-word sentences (e.g., May I see PETE), repeated 4 times each.
Rhotics and Sibilants Task	X	RST		X	X	X	This task provides information for /r/ and /s/ as obtained in imitated single words produced within the carrier phrase "Say ___ again."	Recorded stimuli for 10 words (soon, bird), each repeated four times
Multisyllabic Words Task 1	X	MWT1	X	X			This task provides information on single words selected to represent difficult articulatory sequences assist in evaluating phonological planning, sound sequencing, and transitions from one sound to another. The MWT1 includes 25 single words for children age 3;0 to 11;11.	Recorded stimulus for each of 25 words (e.g., animal)
Multisyllabic Words Task 2	X	MWT2			X	X	See MWT1 description. The MWT2 includes 20 single words for participants age 12;0 and up.	Recorded stimulus for each of 20 words (e.g., emphasis)
Speech Phrases Task	X	SPT	X	X	X	X	This task provides information on 25 two- and three-word phrases selected to represent difficult articulatory sequences which assist in evaluating phonological planning, sound sequencing, and transitions from one sound to another.	Recorded stimulus for each of 25 phrases (e.g., big farm house)
Diadochokinesis Task	X	DDK	X	X	X	X	This task provides information on a participant's ability to coordinate rapid, accurate, and rhythmic alternating movements of the lips and tongue within a single place of articulation and across 2 and 3 places of articulation (bilabial, alveolar, and velar).	Two 1-consonant syllable strings (e.g., papapa), three alternating 2-consonant syllable strings, one alternating 3-consonant syllable string, and the word "particular"
Sustained Vowel Task	X	SVT	X	X	X	X	This task provides information on a participant's respiratory-laryngeal capacity and laryngeal quality.	The vowel /a/
Sustained Consonant Task	X	SCT	X	X	X	X	This task provides information on a participant's respiratory-laryngeal capacity.	The consonant /s/
Orofacial Examination Task		OET	X	X			This task provides information on the structure and function of the speech mechanism.	None
Oral and Written Language Scales		OWLS	X	X	X	X	This task provides information on language comprehension and production.	Two books of picture plates, one each for the comprehension and production subtests

Woodcock-Johnson III Tests of Achievement		WJ III				X	This task provides information on language skills in adults in the areas of Letter-Word Identification Test 1) and Word Attack (Test 13). [Optional tests include: Test 7 – Spelling, Test 9 - Passage Comprehension, Test 11 – Writing Samples]	Test 1: Single letters and increasingly difficult words (e.g., provincial) are displayed for participant's to pronounce. Test 13: Single letters and increasingly difficult nonwords (e.g., [illegible]) are displayed for participant's to pronounce.
Kaufman Brief Intelligence Test (2nd ed.)		KBIT2	X	X	X	X	This task provides information on cognitive functioning using scores from the KBIT2's three verbal and nonverbal subtests.	Two books of picture plates are used for all of the nonverbal and some of the verbal test items.
Case History Form		CHF	X	X	X	X	This form provides risk factor information on a participant's medical, social, academic, hearing, family aggregation, and speech-language history.	None
Case History Interview		CHI	X	X	X	X	The case history interview supplements and clarifies the information collected on the participant's CHF.	None
Examiner Checklist		EC	X	X	X	X	This form provides information on the examiner's impressions of selected aspects of the participant's behavior and psychosocial development/affect.	None

Preschool=3;0-5;11, School-age=6;0-11;11, Adolescent=12;0-17;11, Adult=18;0+

Goldman, R., & Fristoe, M. (2000). Goldman-Fristoe Test of Articulation (2nd ed). Circle Pines, MN: AGS.
American National Standards Institute (1989). *Specification for audiometers* (ANSI S3.6-1989). New York: Author.
Dollaghan, C., & Campbell, T.F. (1998). Nonword repetition and child language impairment. *Journal of Speech, Language, and Hearing Research*, 41, 1136-1146.
Catts, H. (1986). Speech production/phonological deficits in reading disordered children. Journal of Learning Disabilities, 19, 504–508.
Carrow-Woolfolk, E. (1995). *Oral & Written Language Scales*. Circle Pines, MN: American Guidance Service.
Woodcock, R.W., McGrew, K.S., & Mather, N. (2001). *Woodcock-Johnson III*. Itasca, IL: Riverside Publishing.
Kaufman, A.S., & Kaufman, N.L. (2004). *Kaufman Brief Intelligence Test-Second Edition*. Circle Pines, MN: AGS Publishing.

Madison Speech Assessment Protocol: Speech Sampling Context Hierarchy



Vowel Task 1 (VT1) Corner Vowels

1. beet
2. bat
3. boot
4. beet
5. pot
6. bat
7. boot
8. pot
9. bat
10. beet
11. pot
12. boot
13. bat
14. pot
15. boot
16. beet

Vowel Task 2 (VT2) Other Monophthongs & Diphthongs

1. putt
2. bite
3. bought
4. Bert
5. bit
6. bait
7. pet
8. boat
9. put
10. boy
11. bought
12. bite
13. bit
14. boat
15. pet
16. pout
17. bait
18. putt
19. boy
20. pet
21. Bite
22. pout
23. put
24. bait
25. Bert
26. boat
27. bit
28. boy
29. pet
30. pout
31. bought
32. bite
33. bit
34. putt
35. bought
36. bait
37. put
38. Bert
39. boat
40. boy
41. putt
42. put
43. Bert
44. pout

Vowel Task 3 (VT3)

1. She needs strawberry jam on her toast.
2. He has a blue pen.
3. Did you like the zoo this spring?
4. I am tall.
5. Chuck seems thirsty after the race.
6. He has a blue pen.
7. She needs strawberry jam on her toast.
8. Did you like the zoo this spring?
9. I am tall.
10. She needs strawberry jam on her toast.
11. Did you like the zoo this spring?
12. I am tall.
13. Chuck seems thirsty after the race.
14. Did you like the zoo this spring?
15. He has a blue pen.
16. Chuck seems thirsty after the race.
17. I am tall.
18. She needs strawberry jam on her toast.
19. He has a blue pen.
20. Chuck seems thirsty after the race.

Rhotics and Sibilants Task

1. sin
2. crude
3. soon
4. bird
5. skin
6. burr
7. ride
8. burg
9. kiss
10. spoon
11. skin
12. burg
13. sin
14. crude
15. bird
16. spoon
17. ride
18. kiss
19. soon
20. burr
21. skin
22. crude
23. burg
24. sin
25. spoon
26. burr
27. soon
28. ride
29. bird
30. kiss
31. skin
32. burg
33. spoon
34. sin
35. burr
36. crude
37. bird
38. soon
39. ride
40. kiss

Challenging Word Task (CWT)

1. helicopter
2. kangaroo
3. elephant
4. caterpillar
5. tomato
6. octopus
7. alligator
8. watermelon
9. stars
10. skates
11. scissors
12. zipper

Multisyllabic Words Task 1 (MWT1)

1. animal
2. mobilize
3. catalog
4. calendar
5. syllable
6. governor
7. navigator
8. Colorado
9. permanent
10. hemisphere
11. especially
12. establish
13. consequence
14. associate
15. symphony
16. enthusiasm
17. suspicious
18. skeptical
19. bicyclist
20. orchestra
21. substantial
22. susceptible
23. municipal
24. specific
25. Episcopal church

Multisyllabic Words Task 2 (MWT2)

1. emphasis
2. probably
3. sympathize
4. terminal
5. synthesis
6. especially
7. peculiar
8. skeptical
9. fudgesicle
10. vulnerable
11. consciouness
12. suspicious
13. municipal
14. orchestra
15. specific
16. statistics
17. fire extinguisher
18. Episcopal church
19. Statistician
20. Nicaragua

Speech Phrases Task (SPT)

1. blue brush
2. sea shells
3. blue star
4. just right
5. black broom
6. quite right
7. snow slope
8. weak wrist
9. big farm house
10. dark blue hat
11. small broom
12. Tom wears shoes
13. he makes shirts
14. bright blue beam
15. she sells shirts
16. nine horse flies
17. big black bread
18. wastebaskets
19. blue plaid pants
20. fine fruit flies
21. small wrist band
22. three small crabs
23. Quiet crabs claws
24. Mixed biscuits
25. Swiss wrist watch

Lexical Stress Task (LST)

1. Practice Trials

Item #	Stimulus
1	MOP
2	PUPpet

2. Test Trials

Item #	Stimulus
1	AIRPLANE
2	aWARD
3	baBOON
4	BASEBALL
5	BATHTUB
6	CHICKen
7	COWBOY
8	DISHes
9	FOOTBALL
10	gaRAGE
11	giRAFFE
12	HOTDOG
13	HAMmer
14	guITAR
15	LADder
16	maCHINE
17	PEAnut
18	PUPpy
19	racCOON
20	reMOTE
21	RObot
22	SIDEWALK
23	SNOWMAN
24	WINdow

Emphatic Stress Task

1. May I see PETE?
2. May I SEE Pete?
3. May I see Pete?
4. MAY I see Pete?
5. Bob may go HOME.
6. Bob may GO home.
7. Bob MAY go home.
8. BOB may go home.

Diadochokinesis Task (DDK)

Stimulus	Number of syllables in 5 seconds	Sounds were accurate	Production was rhythmic
pʌ			
kʌ			
pʌtʌ			
pʌkʌ			
tʌkʌ			
pʌtʌkʌ			
pattycake			

Goldman-Fristoe Test of Articulation-2

1. house
2. tree
3. window
4. telephone
5. cup
6. knife
7. spoon
8. girl
9. ball
10. shovel
11. wagon
12. monkey
13. banana
14. zipper
15. scissors
16. duck
17. quack
18. yellow
19. vacuum
20. watch
21. plane
22. swimming
23. watches
24. light/lamp
25. car
26. blue
27. rabbit
28. carrot
29. orange
30. fishing
31. chair
32. feather
33. pencils
34. this
35. bathtub
36. bath
37. ring
38. finger
39. thumb
40. jumping
41. pajamas
42. flowers
43. brush
44. drum
45. frog
46. green
47. green
48. clown
49. balloons
50. crying
51. glasses
52. slide
53. stars
54. five

Nonword Repetition Task (NRT)[a]

1. naɪb
2. voʊp
3. taʊʤ
4. dɔɪf
5. teɪvʌk
6. ʧoʊvæg
7. vætʃaɪp
8. nɔɪtaʊf
9. ʧɪnɔɪtaʊb
10. naɪʧoʊveɪb
11. dɔɪtaʊvæb
12. teɪvɔɪʧaɪg
13. veɪtaʧaɪdɔɪp
14. dævoʊnɔɪʧɪg
15. naɪʧɔɪtaʊvub
16. tævaʧɪnaɪg

[a]Dollaghan & Campbell, 1998

Syllable Repetition Task (SRT)[a]

1. bada
2. dama
3. bama
4. mada
5. naba
6. daba
7. nada
8. maba
9. bamana
10. dabama
11. madaba
12. nabada
13. banada
14. manaba
15. bamadana
16. danabama
17. manabada
18. nadamaba

[a] Shriberg, Lohmeier et al., 2009

Madison Speech Assessment Protocol: Data Reduction

PEPPER Environment

Perceptual

Narrow phonetic transcription

Prosody-Voice Screening Profile coding

Acoustic

TF32-Active X

Automated; high throughput

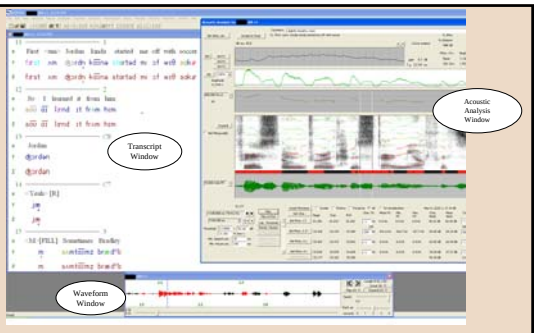


Figure 1 is a display of the three windows viewable during acoustic analysis: the perceptual transcript, the waveform for the conversational sample, and the acoustic analysis window. For acoustic analysis, the transcript window provides information on the coded utterances (displayed to the right of the numeric utterance), any PVSP codes used, the phoneme perceptually transcribed, and the phonemes marked for acoustic analysis (displayed according to a color code). Displayed in this figure is the first coded utterance in a conversational sample. Data for the segmented utterance and all segmented phonemes is viewable in the acoustic analysis window by a scrolling function (to include onset and offset times for the utterance and each individual phoneme, pauses, characteristic F0, Mean F0, minimum and maximum F0, characteristic amplitude, and F1-F3 data). Additionally, for this sample the moment data for a segmented fricative is displayed in the upper right corner of the acoustic analysis window.

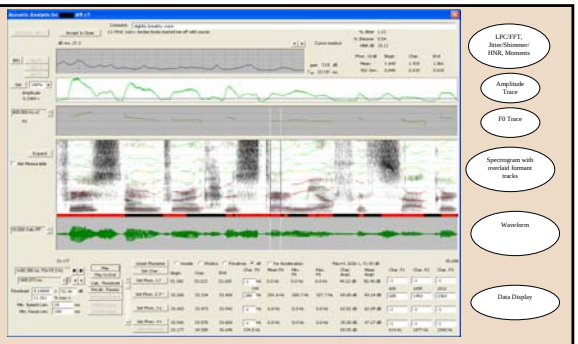


Figure 2 is a display of the analysis window for all acoustic measurements. Moving from the top of the window are: the comment box, the utterance gloss from the transcript, jitter/shimmer/HNR data, the LPC/FFT window, moment data display, the amplitude trace window, the F0 trace window, the spectrogram window, the waveform window, and the data window including data display for all measured data. For this figure the jitter/shimmer/HNR data is displayed for the segmented vowel (phoneme #16 in this utterance: /a/)

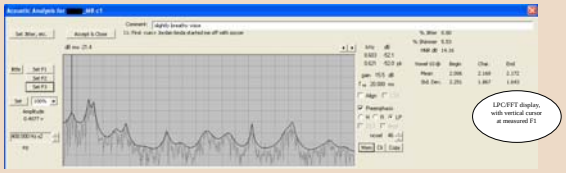


Figure 3 is a display of the portion of the acoustic analysis window that is used to make F1-F3 measurements. An LPC trace is overlaid on the FFT trace. Each formant value is determined based on the LPC peak, with visual confirmation from the formant band in the spectrogram. In this figure, data for F1 for the vowel /a/ is displayed.

Premises in a Neurodevelopmental Framework for CAS Research

Progress in CAS Research Requires a Standardized Assessment Protocol and a Standardized Suite of Analytics

Standardized Assessment Protocol

The Madison Speech Assessment Protocol (MSAP)[a]

- Scores/subscale scores on all MSAP tasks
- Reference sample of 4-to 16-year-old typically developing speakers assessed with the MSAP

Standardized Suite of Analytics

Competence, Precision, and Stability Analytics (CPSA)[a]

- Currently includes 84 indices/subindices and markers/submarkers
- Each index/subindex/marker is derived from 1-8 MSAP tasks
- Current criteria for a 'Positive' marker: z-score < 1 standard deviation from the mean of typically-developing speakers of same age and sex

[a]Shriberg, Fourakis, et al., 2009

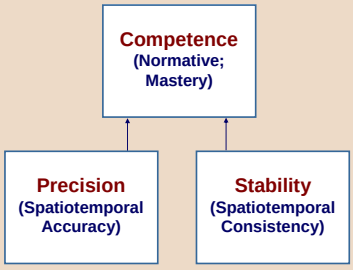
Example of Current Diagnostic Markers and Classification Criteria for CAS[a]

For a judgment of the presence of CAS, the child had to exhibit at least 4 of the following 10 characteristics in at least 3 of the MSAP tasks:

- Difficulty achieving initial articulatory configurations and transitions into vowels
- Lexical stress errors or equal stress
- Vowel or consonant distortions including distorted substitutions
- Syllable segregation
- Groping
- Intrusive schwa
- Voicing errors
- Slow rate
- Slow diadochokinetic rates
- Increased difficulty with longer or more phonetically complex words

[a]Shriberg, Potter, & Strand (2009)

Competence, Precision, and Stability Analytics (CPSA)



30 Competence Indices[a]

Tier	Domain	Metric
Segmental		
	1. Vowels	Percentage of Non-rhotic Vowels/Diphthongs Correct Percentage of Rhotic Vowels/Diphthongs Correct Percentage of Phonemic Diphthongs Correct Percentage of Vowels/Diphthongs Correct: CS Percentage of Vowels/Diphthongs Correct: AT Percentage of Non-rhotic Vowels/Diphthongs Correct Revised Percentage of Rhotic Vowels/Diphthongs Correct Revised Percentage of Phonemic Diphthongs Correct Revised Percentage of Vowels/Diphthongs Correct Revised: CS Percentage of Vowels/Diphthongs Correct Revised: AT Percentage of Relative Non-rhotic Vowel/Diphthong Distortions
	2. Consonants	Percentage of Consonants in Inventory Percentage of Consonants Correct: CS Percentage of Consonants Correct: AT Percentage of Consonants Correct- Revised: CS Percentage of Consonants Correct- Revised: AT Percentage of Consonants Correct in Complex Words: MWT Relative Omission Index Relative Substitution Index Relative Distortion Index
	3. Vowels & Consonants	Speech Disorders Classification System Intelligibility Index Percentage of Structurally Correct Words
Suprasegmental		
	4. Phrasing	Percentage Appropriate Phrasing
	5. Rate	Percentage Appropriate Rate
	6. Stress	Percentage Appropriate Stress
	7. Loudness	Percentage Appropriate Loudness
	8. Pitch	Percentage Appropriate Pitch
	9. Laryngeal Quality	Percentage Appropriate Laryngeal Quality
	10. Resonance Quality	Percentage Appropriate Resonance Quality

[a] **All competence metrics obtained by perceptual methods (phonetic transcription; prosody-voice coding)**

54 Precision and Stability Indices

Segmental	Precision	Stability
1. Vowels	Reduced Vowel Space Lengthened Vowels Distorted Rhotics Reduced Pairwise Vowel Duration Variability	Less Stable Vowel Space Less Stable F1 Less Stable F2 Less Stable Vowel Duration Less Stable Rhotic Distortions: F3-F2 Less Stable Vowel Errors
2. Consonants	Nasal Emissions Reduced % Glides Correct Lowered Sibilant Centroids Lengthened Cluster Durations	Less Stable Consonant Errors Less Stable Sibilant Centroids
3.Vowels and Consonants	Increased Percentage of Phoneme Distortions Syllable/Word Segregation: Increased % Between/Within-Word Pauses	Less Stable Whole Word Errors Less Stable % Phonemes Correct in Complex Words
Suprasegmental	**Precision**	**Stability**
4. Phrasing	Increased Repetitions and Revisions	Reduced Speech-Pause Duration Variability Ratio
5. Rate	Slower Speaking Rate Slower Articulation Rate	Less Stable Speaking Rate Less Stable Articulation Rate
6. Stress	Reduced Lexical Stress Reduced Emphatic Stress Reduced Sentential Stress	Less Stable Lexical Stress Less Stable Emphatic Stress Less Stable Sentential Stress
7. Loudness	Reduced Vowels-Consonants Intensity Ratios Increased Vowels-Consonants Intensity Ratios	Less Stable Vowels-Consonants Intensity Ratios
8. Pitch	Lowered Fundamental Frequency Mean Raised Fundamental Frequency Mean Lowered Fundamental Frequency Range Increased Fundamental Frequency Range	Less Stable Mean Fundamental Frequency
9. Laryngeal Quality	Increased Jitter Increased Shimmer Reduced Harmonics-to-Noise Ratio Increased % Breathy Utterances Increased % Rough Utterances Increased % Strained Utterances Increased % Break/Shift/Tremorous Utterances	Less Stable Jitter Less Stable Shimmer Less Stable Harmonics-to-Noise Ratio
10. Resonance Quality	Increased % Nasal Utterances Nasal: Lowered F1:/i/ Nasopharyngeal: Lowered F2: High Vowels	Nasopharyngeal: Less Stable F2: High Vowels

25 Putative Diagnostic Markers of MSD-AOS

Segmental	Precision	Stability
1. Vowels/Diphthongs		Less Stable Vowel Space Less Stable F1 Less Stable F2 Less Stable Vowel Duration Less Stable Rhotic Distortions: F3-F2 Less Stable Vowel Errors
2. Consonants	Reduced % Glides Correct	Less Stable Consonant Errors Less Stable Sibilant Centroids
3.Vowels/Diph & Consonants		Less Stable Whole Word Errors Less Stable % Phonemes Correct in Complex Words
Suprasegmental	**Precision**	**Stability**
4. Phrasing	Increased Repetitions and Revisions	Reduced Speech-Pause Duration Variability Ratio
5. Rate		Less Stable Speaking Rate Less Stable Articulation Rate
6. Stress		Less Stable Lexical Stress Less Stable Emphatic Stress Less Stable Sentential Stress
7. Loudness		Less Stable Vowels-Consonants Intensity Ratios
8. Pitch		Less Stable Mean Fundamental Frequency
9. Laryngeal Quality		Less Stable Jitter Less Stable Shimmer Less Stable Harmonics-to-Noise Ratio
10. Resonance Quality	Nasopharyngeal: Lowered F2: High Vowels	Nasopharyneal: Less Stable F2: High Vowels

12 Putative Diagnostic Markers of MSD-DYS

Segmental	Precision	Stability
1. Vowels/Diphthongs		
2. Consonants	Nasal Emissions	
3. Vowels/Diph & Consonants		
Suprasegmental	**Precision**	**Stability**
4. Phrasing		
5. Rate		
6. Stress		
7. Loudness		
8. Pitch	Lowered Fundamental Frequency Mean Lowered Fundamental Frequency Range	
9. Laryngeal Quality	Increased Jitter Increased Shimmer Reduced Harmonics-to-Noise Ratio Increased % Breathy Utterances Increased % Rough Utterances Increased % Strained Utterances Increased % Break/Shift/Tremorous Utterances	
10. Resonance Quality	Increased % Nasal Utterances Nasal: Lowered F1:/i/	

17 Indices Currently Assigned to MSD-NOS

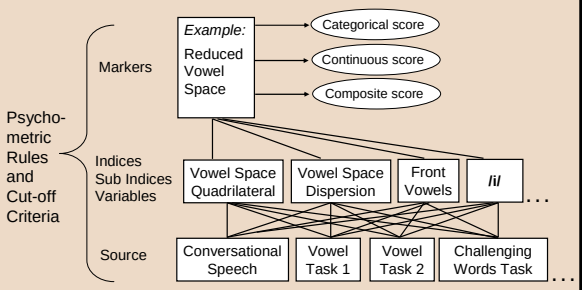
Segmental	Precision	Stability
1. Vowels/Diphthongs	Reduced Vowel Space Lengthened Vowels Distorted Rhotics Reduced Pairwise Vowel Duration Variability	
2. Consonants	Lowered Sibilant Centroids Lengthened Cluster Durations	
3. Vowels/Diph & Consonants	Increased Percentage of Phoneme Distortions Syllable/Word Segregation: Increased % Between/Within Word Pauses	
Suprasegmental	**Precision**	**Stability**
4. Phrasing		
5. Rate	Slower Speaking Rate Slower Articulation Rate	
6. Stress	Reduced Lexical Stress Reduced Emphatic Stress Reduced Sentential Stress	
7. Loudness	Reduced Vowels-Consonants Intensity Ratios Increased Vowels-Consonants Intensity Ratios	
8. Pitch	Raised Fundamental Frequency Mean Increased Fundamental Frequency Range	
9. Laryngeal Quality		
10. Resonance Quality		

Number of Precision and Stability Markers/Indices for the Three Classifications of Motor Speech Disorders

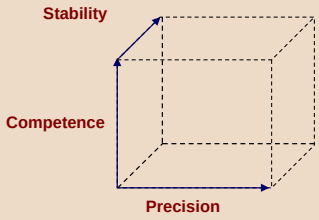
Subtype	Precision			Stability			Total		
	P[a]	A[a]	T[a]	P	A	T	P	A	T
MSD-AOS	2	1	3	4	18	22	6	19	25
MSD-DYS	2	10	12	0	0	0	2	10	12
MSD-NOS	1	16	17	0	0	0	1	16	17
Total MSD	5	27	32	4	18	22	9	45	54

[a] **P= Perceptual (Transcription; Prosody-Voice Coding); A=Acoustic; T=Total**

Competence, Precision, and Stability Analytics (CPSA): Psychometric Structures



Competence, Precision, and Stability Protocol (CPSA): Current and Future Analyses



Current analyses: Box score tallies (i.e, % Positive Markers for each participant and averaged over participant groups and subgroups.)

Future analyses: Multivariate modeling (e.g., cluster analyses, structural equation analyses)

Three Questions About CAS in a Complex Neurodevelopmental Disorder: Galactosemia

- What is the estimated prevalence of CAS in children with galactosemia and prior or persistent speech sound disorder?
- What sociodemographic, cognitive-linguistic, orofacial, or dietary management variables in youth with galactosemia are significant correlates of or risk factors for CAS?
- What speech, prosody, and/or voice indices best discriminate participants with galactosemia and CAS from participants with galactosemia and other speech sound disorders?

First Hour

Contemporary Research in CAS: Shriberg

- Premises in Three Contemporary Areas of CAS Research
- Premises in a Neurodevelopmental Framework for CAS Research
- Assessment and Analytic Methods in the Present Study

Galactosemia: Potter

- **Overview**
- **Data Collection Methods in the Present Study**

Diagnostic Classification of CAS: Strand

- Contemporary Classification Criteria
- Diagnostic Classification Methods in the Present Study

Second Hour

Findings: Shriberg

Discussion: Shriberg, Potter, Strand

Open Discussion

Overview of Galactosemia

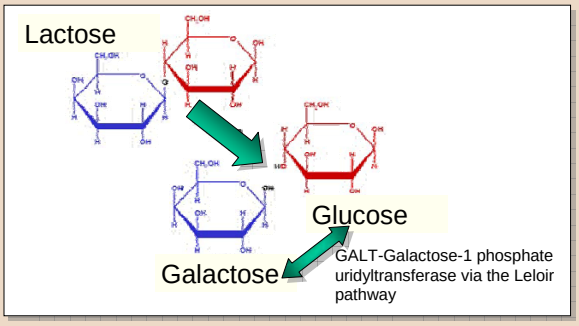
- Rare genetic inborn error of metabolism
  - Gene location-short arm of chromosome 9
- Detected during newborn screening
- Inability to fully breakdown the sugar in milk (lactose)
- Incidence-1:1,000,000 in Japanese, 1:40,000 in European descent, 1:480 in the Irish Traveller (Murphy et al., 1999)

http://www.milton-keynes.gov.uk/pandemic/images/TES_Road_side_

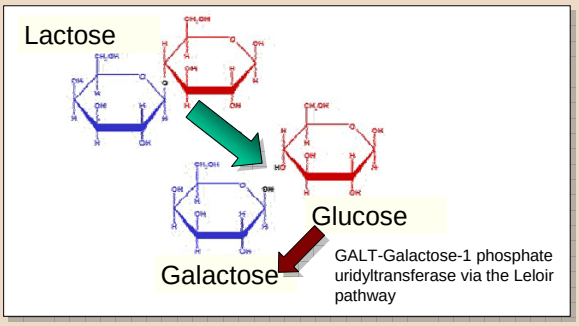
http://www.swisslab.de/com

http://www.nature.com/nature/supplements/collections/humangenome/chromosomes/images/chrom_9.gif

Lactose metabolism-typical



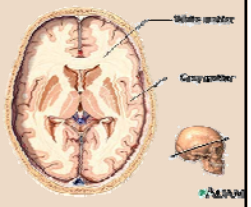
Lactose metabolism-galactosemia



GALT-Galactose-1 phosphate uridyltransferase via the Leloir pathway

Why do we need galactose?

- Excess galactose
  - Toxic to myelin
- Galactose needed
  - For normal myelin formation
- Leading to white matter abnormalities
  - Brain
  - Myelin sheath on nerves



http://www.nlm.nih.gov

Prenatal complications of galactosemia

- Inability to metabolize galactose
  - Evident at 10 weeks gestation age
  - Detected in liver and amnio fluid
- Not related to lactose restricted diet by mother during pregnancy

www.sciencemuseum.org.uk/on-line/lifecycle/52.asp

Postnatal complications of galactosemia

If undetected leads to

- Toxicity due to increase galactose-1-phosphate in red blood cells
- Jaundice with enlargement of liver and spleen
- Kidney dysfunction
- Muscle hypotonia
- Cataract formation
- Sepsis
- Death within ~2-10 weeks after birth

If detected early and lactose-restricted

Children may exhibit

- Cognitive impairments~50% of children (Waggoner et al., 1990)
- Speech sound disorders in ~50-60% of children (Nelson et al., 1991; Schweitzer et al., 1993; Webb et al., 2003)
- Language impairments in ~90% of children (Waggoner et al., 1990)
- Motor impairments~20% of children (Waggoner, et al. 1990)
- Ovarian failure in ~99.9% of females (Fridovich-Keil, 2006)
- Diminished bone density (Panis et al., 2005)

Data Collection Methods in the Present Study

Participants

- 33 children with classic galactosemia and a history of speech sound disorders
- Ages 4-16
- Recruited through
  - Parents of Galactosemic Children organization
  - Metabolic clinics
- Tested in their homes in 17 states

Data Collection Methods in the Present Study

Procedure

- **Case history**
- **Oral exam, hearing screening**
- **Standardized tests**
  - Goldman-Fristoe Test of Articulation-2 (GFTA-2)
  - Oral and Written Language Scale (OWLS)
  - Kaufman Brief Intelligence Test (KBIT-2)
  - Movement Assessment Battery for Children (MABC)
- **Conversational speech**
- **Sustained phonation of /a/**
- **Computerized speech tasks**
  - Madison Speech Assessment Protocol (MSAP)

MSAP: Vowel Task 1 (VT-1)

Video Sample 1

MSAP: Nonword Repetition Task (NRT)

Video Sample 2

First Hour

Contemporary Research in CAS: Shriberg

- Premises in Three Contemporary Areas of CAS Research
- Premises in a Neurodevelopmental Framework for CAS Research
- Assessment and Analytic Methods in the Present Study

Galactosemia: Potter

- Overview
- Data Collection Methods in the Present Study

Diagnostic Classification of CAS: Strand

- **Contemporary Classification Criteria**
- **Diagnostic Classification Methods in the Present Study**

Second Hour

Findings: Shriberg

Discussion: Shriberg, Potter, Strand

Open Discussion

Contemporary Clinical Diagnosis of CAS & CAS Classification Method for the Current Study

Edythe Strand, Ph.D.
Mayo Clinic
ASHA, New Orleans, 2009

Contemporary Clinical Diagnosis of CAS

- My charge this morning:
  - Is to provide a clinical view of those speech behaviors which help us in our differential diagnosis of CAS
  - Review the methods for classification of CAS in this study
- Nosology is often difficult
  - Especially true for CAS
  - Historically there has been a lack of consensus on what which speech and non-speech characteristics are pathognomonic for CAS

Why?

- Clinicians and researchers who study speech sound disorders come from different backgrounds
  - Linguistics
  - Speech pathology – language and phonology backgrounds
  - Speech pathology – motor speech backgrounds
- Clinicians and Researchers also work in different environments, influencing their perspective – medical setting vs. clinic or University

ALSO

- No physiologic marker
- Have to rely on behavioral markers, observed in habitual and spontaneous utterances, but also in carefully elicited utterances – such as in a motor speech exam – but
- No current validated method for diagnosis with good specificity and sensitivity

So, how did the commonly accepted speech characteristics of CAS come to be?

- Many of them are similar to those noted in adults with acquired apraxia of speech (e.g.)
  - Difficulty achieving initial articulatory configurations
  - Vowel distortions
  - Inconsistency across repeated trials
- There has been some disagreement among clinician/researchers about whether making these comparisons between adults and children is appropriate

Rationale for characteristics considered as providing evidence for CAS

- There are a number of reasons why characteristics of CAS are similar to those with acquired AOS
  - Observations of children with acquired strokes show similar characteristics to those children with idiopathic forms
  - The concept of "praxis" leads to logical inferences about what might be seen if a person has difficulty with "praxis"

Clinical diagnosis - terminology

Praxis

- The conception and planning of a motor act in response to an environmental demand (Stedman's Medical Dictionary)
- The ability to perform purposeful and learned voluntary movement (Benarroch, 2008)

Apraxia

- Frequently used in neurology to refer to deficits in planning and programming movement for voluntary purposeful movement
  - Limb apraxia
  - Oral apraxia
  - Verbal apraxia
- Typically from acquired cortical neurologic impairment

Acquired Apraxia of Speech

- Fred Darley was the person who proposed and developed the modern concept of apraxia of speech
- In 1968,he posited the use of this term to discriminate the language impairment associated with aphasia, and the movement disorders represented by the dysarthrias – through the use of this third category, which he believed resulted from impairment in planning, and programming movement gestures for speech

Apraxia of Speech

- Debate raged for the next 20 years as to whether there is indeed a category of impairment separate from either aphasia or dysarthria
- A large body of perceptual, acoustic and physiologic research has demonstrated that there is validity for such a category

Apraxia of Speech

- Because the original work in AOS focused on acquired AOS, it may be overlooked that apraxia in childhood was posited in the 1950s and 1960s (Morley, Court, Miller & Garside, 1955; Morley, 1965)
  - Because of observations that a subgroup of children with speech sound disorders seemed to exhibit characteristics different than children with typical speech errors.
  - These characteristics were also different from dysarthria.

Then, in 1974, Yoss and Darley:

- Empirically examined what before had been descriptions of similar speech symptomology.
- They studied 30 children with articulation deficits and were able to identify behaviors that could differentiate a subset of children with articulation disorders who exhibited difficulty with praxis (Yoss & Darley, 1974a, 1974b).

Differentiating children who likely have difficulty with "praxis"

- The speech observations currently accepted as being characteristic of CAS represent behaviors long thought in neurology to be pathognomonic of difficulty with praxis
- These characteristics have been shown to differentiate a subgroup of speech disordered children (Yoss and Darley,1974; Strand, McCauley, et al., 2006)

What are these characteristics?

- Difficulty with achieving initial articulatory configurations making the transition from one articulatory configuration to another
- Groping and/or trial and error behavior
- Presence of vowel distortions
- Limited consonant and vowel repertoire
- Increased errors with increased word length and phonetic complexity

What are these characteristics?

- Difficulty completing a movement gesture for a phoneme easily produced in a simple context, but not in a longer one
- Altered suprasegmentals
  - lexical and sentential stress
  - overall prosodic contours
- Inconsistent error patterns
- Segmented syllables
- Slow DDK

Keep in mind that the diagnosis of CAS is dynamic

- Diagnosis is made at one point in time during development.
- Children vary in the type and severity of the characteristics of CAS
- Characteristics within a child change over time with neural maturation + appropriate therapy

CAS in Children with Galactosemia

Methods for identifying participants as exhibiting characteristics of a motor speech disorder–CAS and/or dysarthria

- Now I'll shift to describing the methodology for determining the speech status of each participant with galactosemia relative to contemporary classification criteria for CAS and dysarthria
- One clinician/researcher experienced in differential diagnosis, and using the Mayo Clinic system of perceptual classification of motor speech disorders, made the judgments.

Procedure

- Each video or audio tape of the MSAP for each child with galactosemia was judged with respect to:
  - The presence or absence of 10 speech behaviors identified as being characteristic of CAS
  - The presence or absence of 10 speech behaviors identified as being characteristic of dysarthria

Criteria for Identification as CAS

- Observation of at least 4 of 10 speech characteristics frequently associated with CAS
- One or more of the characteristics must be observed in at least 3 of the MSAP tasks

10 Speech characteristics - CAS

- Difficulty achieving initial articulatory configurations and transitions into vowels (DAAC+T)
- Lexical stress errors – equal stress (LSE)
- Vowel or Consonant Distortions including distorted substitutions (VorCD)
- Syllable segregation (SS)
- Groping (GR)

10 Speech characteristics - CAS

- Intrusive schwa (IS)
- Voicing errors (VE)
- Slow rate (SR)
- Slow diadochokinetic rates (slow DDK)
- Increased difficulty with longer or more phonetically complex words (IDLPCW)

Criteria for Identification as Dysarthric

- Observation of 3/10 selected speech characteristics
- One or more of the characteristics must be observed in at least 3 of the MSAP tasks

Characteristics related to Dysarthria

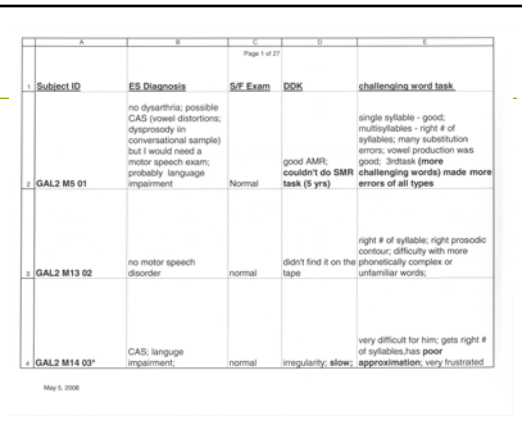
- Scanning speech (SS)
- Equal stress (ES)
- Sound distortions (SD)
- Irregular diadochokinetic rate (ataxia) (DDK)
- Slow rate (SR)
- Reduced range of motion (RRM)

Characteristics related to Dysarthria

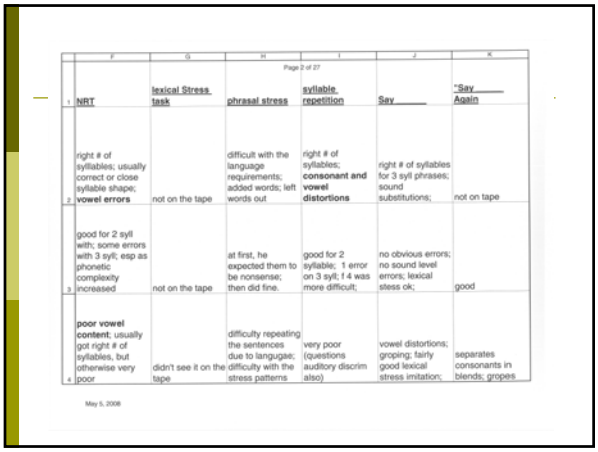
- Reduced strength of articulatory contacts (RS)
- Reduced respiratory support or respiratory incoordination (RRS-I)
- Strained or breathy phonatory quality (PQ)
- Adventitious movement (AD)

Illustration of the working excel sheet

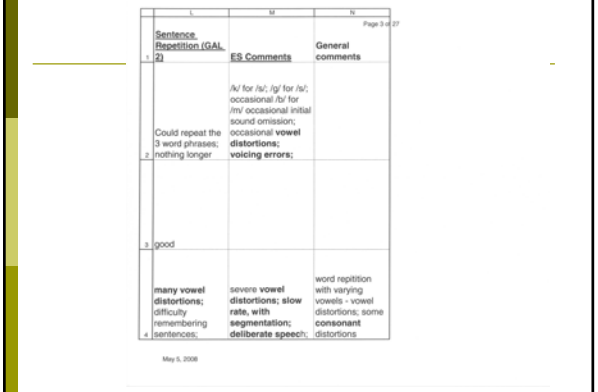
- Note the kinds of comments I made regarding observations of speech and non-speech characteristics
- Bolded observations were those characteristics I would later tally to make decisions about clinical diagnosis



	A	B	C	D	E
1	Subject ID	ES Diagnosis	S/F Exam	DDK	challenging word task
2	GAL2 M5 01	no dysarthria; possible CAS (vowel distortions; dysprosody iin conversational sample) but I would need a motor speech exam; probably language impairment	Normal	good AMR; **couldn't do SMR task (5 yrs)**	single syllable - good; multisyllables - right # of syllables; many substitution errors; vowel production was good; 3rdtask **(more challenging words) made more errors of all types**
3	GAL2 M13 02	no motor speech disorder	normal	didn't find it on the tape	right # of syllable; right prosodic contour; difficulty with more phonetically complex or unfamiliar words;
4	GAL2 M14 03*	CAS; languge impairment;	normal	irregularity; **slow;**	very difficult for him; gets right # of syllables,has **poor approximation**; very frustrated



NRT	lexical Stress task	phrasal stress	syllable repetition	Say ____	"Say ____ Again
right # of syllables; usually correct or close syllable shape; **vowel errors**	not on the tape	difficult with the language requirements; added words; left words out	right # of syllables; **consonant and vowel distortions**	right # of syllables for 3 syll phrases; sound substitutions;	not on tape
good for 2 syll with; some errors with 3 syll; esp as phonetic complexity increased	not on the tape	at first, he expected them to be nonsense; then did fine.	good for 2 syllable; 1 error on 3 syll; f 4 was more difficult;	no obvious errors; no sound level errors; lexical stess ok;	good
poor vowel content; usually got right # of syllables, but otherwise very poor	didn't see it on the tape	difficulty repeating the sentences due to language; difficulty with the stress patterns	very poor (questions auditory discrim also)	vowel distortions; groping; fairly good lexical stress imitation;	separates consonants in blends; gropes



Sentence Repetition (GAL 2)	ES Comments	General comments
Could repeat the 3 word phrases; nothing longer	/k/ for /s/; /g/ for /s/; occasional /b/ for /m/ occasional initial sound omission; occasional **vowel distortions; voicing errors;**	
good		
many vowel distortions; difficulty remembering sentences;	severe **vowel distortions; slow rate, with segmentation; deliberate speech**;	word repitition with varying vowels - vowel distortions; some **consonant** distortions

Example of Worksheet Summary

Participant ID **GAL 2 M14 03**

S/F Exam — Normal

DDK — Slow; awkward;

NRT ✓ — Poor vowel content; vowel distortions

EST ✓ — Stress errors; slow rate

VT1 ✓ — Vowel distortions; groping

VT2 ✓ — Separates consonants in blends; intrusive schwa; gropes

VT3 ✓ ✓ — Many vowel distortions; difficulty remembering sentences

ES Comments — severe vowel distortions; slow rate, with segmentation; deliberate speech; stress errors; some consonant distortions

ES Diagnosis — CAS

First Hour

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- Assessment and Analytic Methods in the Present Study

Galactosemia: Potter

- Overview
- Data Collection Methods in the Present Study

Diagnostic Classification of CAS: Strand

- Contemporary Classification Criteria
- Diagnostic Classification Methods in the Present Study

Second Hour

Findings: Shriberg

Discussion: Shriberg, Potter, Strand

Open Discussion

Tallies of Behavioral Markers per CAS Participant

	F8-02	M10-04	M5-01	M14-03
DAAC+T	X			
LSE	X	X	X	X
VorCD	X	X	X	X
SS	X			
GRP				X
IS	X			X
VE	X	X		
SR	X	X		X
Slow DDK		X	X	X
IDLPCW			X	

Three Galactosemia Subgroups and a Comparison Group of 3- to 6-year-old Children with Significant Speech Delay[a]

	Group			
	GALACTOSEMIA			
	Group 1 (n=8) GALT CAS	Group 2 (n=9) GALT SD	Group 3 (n=15) GALT SE	Group 4 (n=25) SD
Galactosemia (GALT)	✓	✓	✓	
Speech Delay (SD)	✓	✓		✓
Childhood Apraxia of Speech (CAS)	✓			

[a]Shriberg, Potter, & Strand (2009)

Q1. What is the estimated prevalence of CAS in children with galactosemia and prior or persistent speech sound disorder?

- Conditional prevalence estimate = 24%
  8 of 33 participants with galactosemia and prior or persistent SSD met EAS's classification criteria for CAS
- Unconditional prevalence estimate = 18%
  8 of 44 participants (adding 11 candidate participants without prior or persistent SSD) met EAS's classification criteria for CAS

Some Prevalence Estimate Comparisons

- Estimated prevalence of CAS in youth with galactosemia
  - Prior studies: 48%
  - Present study: 18-24%
- Estimated prevalence of idiopathic CAS
  - .1% (Shriberg, Aram, & Kwiatkowski, 1997)
- Risk for CAS in galactosemia compared to risk for idiopathic CAS in the general population
  - 180-fold increased risk (i.e., 0.18/0.001)

Q2. What socio-demographic, cognitive-linguistic, orofacial, or dietary variables in youth with galactosemia are significant correlates of, or risk factors for, CAS?

Variable	Group 1 GALT CAS	Group 2 GALT SD	Group 3 GALT SE	Comparisons
Socio-demographic				
Gender (% Male)	75%	33%	48%	—
Age (years)	9	7	10	Grp 3 > Grp 2*
Mother Educ (years)	15	15	16	—
Father Educ (years)	13	15	16	—

* Significant effect size

Q2. What sociodemographic, cognitive-linguistic, orofacial, or dietary variables in youth with galactosemia are significant correlates of, or risk factors for, CAS?

Variable	Group 1 GALT CAS	Group 2 GALT SD	Group 3 GALT SE	Significant Effect Size Comparisons
Cognitive				
KBIT-2 <85%	75%	33%	47%	—
Language				
OWLS Comp. (SS)	75.1	86.0	80.9	—
OWLS Express. (SS)	68.8	86.2	83.6	Grp 1 < Grp 2 Grp 1 < Grp 3
Nonword Repetition				
NRT (z scores)	- 6.4	- 2.5	- 1.8	Grp 1 < Grp 2 Grp 1 < Grp 3
SRT (z scores)	- 4.9	- 1.9	- 1.7	Grp 1 < Grp 3

Q2. What sociodemographic, cognitive-linguistic, orofacial, or dietary variables in youth with galactosemia are significant correlates of or risk factors for CAS?

Variable	Group 1 GALT CAS	Group 2 GALT SD	Group 3 GALT SE	Significant Effect Size Comparisons
Orofacial				
Atypical Structure	25%	44%	33%	——
Atypical Function	100%	100%	87%	——
Galactosemia				
Days Until Diagnosis	8.3	8.3	8.6	——
Days on Milk	7.1	6.9	7.9	——

Q3. What speech, prosody, and/or voice domains best discriminate participants with galactosemia and CAS from participants with galactosemia and other speech sound disorders?

Competence Indices (30) Findings

Tier	Domain	Percentage of Significant Effect Size Comparisons: Group 1 < Group 2 GALT CAS	GALT SD	Group 1 < Group 4 GALT CAS	SD
Segmental					
Vowels	1	73%*		44%*	
Consonants	2	33%		0%	
Vowels & Cons.	3	33%		0%	

*Significant

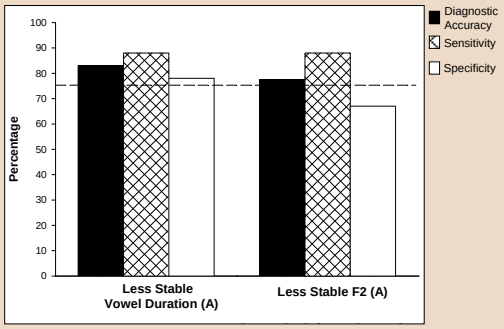
Q3. What speech, prosody, and/or voice domains best discriminate participants with galactosemia and CAS from participants with galactosemia and other speech sound disorders?

Competence Indices (30) Findings

Tier	Domain	Significant Effect Size Comparisons: Group 1 < Group 2 GALT CAS	GALT SD	Group 1 < Group 4 GALT CAS	SD
Suprasegmental					
Prosody					
Phrasing	4	—		—	
Rate	5	—		Slow	
Stress	6	—		Inappropriate	
Voice					
Loudness	7	—		—	
Pitch	8	—		—	
Laryngeal	9	—		—	
Resonance	10	—		Nasopharyngeal	

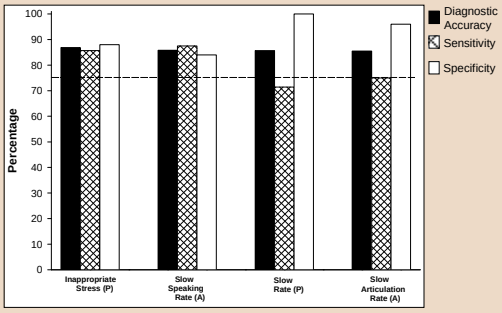
Promising Diagnostic Markers for CAS

Markers Discriminating GALT CAS from GALT SD



Promising Diagnostic Markers for CAS

Markers Discriminating GALT CAS from SD



Summary of Significant Findings: Prevalence, Correlates, Risk Factors of/for CAS

In a sample of youth with galactosemia and speech delay or speech errors, participants meeting criteria for CAS had:

- Higher prevalence of CAS than estimated for children with idiopathic CAS
- High prevalence of motor speech disorder than estimated for children with idiopathic SSD
- High percentage of participants with < 85 cognitive status than found for other participants with galactosemia
- Lower expressive language scores than other participants with galactosemia
- No differences in dietary histories from other participants with galactosemia

Summary of Significant Findings: Speech, Prosody, and Voice Markers of CAS

- Lowered percentage of correct vowels
- Less stable F2
- Less stable vowel duration
- Slower Rate
- Inappropriate stress

First Hour

Contemporary Research in CAS: Shriberg

- Premises in Three Contemporary Areas of CAS Research
- Premises in a Neurodevelopmental Framework for CAS Research
- Assessment and Analytic Methods in the Present Study

Galactosemia: Potter

- Overview
- Data Collection Methods in the Present Study

Diagnostic Classification of CAS: Strand

- Contemporary Classification Criteria
- Diagnostic Classification Methods in the Present Study

Second Hour

Findings: Shriberg

Discussion: Shriberg, Potter, Strand

Open Discussion

Discussion

- Present findings are our first using a neurodevelopmental research framework for CAS research. They are viewed as support for:
  - Etiologic-based classification, including MSD-NOS
  - Standardized assessment protocol
  - Perceptual and acoustic methods
  - Standardized suite of analytics
- Studies in progress
  - CAS in neurological and other neurodevelopmental contexts
  - Comparative studies of adult AOS and idiopathic CAS
  - Genomic and collaborative genomic/neuroimaging projects
- Significant research needs
  - Diagnostic procedures to assess very young children
  - Diagnostic procedures for children with minimal or no speech

Participant with galactosemia and CAS

Multisyllabic Word Repetition Task-2 (MWT-2)

Video Sample 3

Participant with galactosemia and CAS

Challenging Word Task (CWT)

Video Sample 4

Participant with galactosemia and DYS

Conversational Speech Sample

Video Sample 5

Participant with galactosemia and DYS

Follow-up Conversational Speech Sample

Video Sample 6

Motor Disorders in Galactosemia

Movement Assessment Battery for Children (MABC)

- **MABC performance <5th%**
  - Associated with diagnosis of developmental coordination disorder (DCD)
  - 100% of participants with galactosemia and DYS
  - 85% of participants with galactosemia and CAS
  - 44% of participants with galactosemia not diagnosed with DYS or CAS
- **Tremors**
  - 3 participants diagnosed with postural/extension tremor
    - None diagnosed with CAS or DYS
    - 100% MABC performance <5%

Lessons learned…

August 2009 Adult galactosemia study
Harvard University/Boston Children's Hospital
Video Sample 7

After testing the adults with galactosemia, the examiner would have used stricter criteria on structure function exam

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Thanks . . .

http://www.waisman.wisc.edu/phonology/