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Speech Characteristics in Galactosemia

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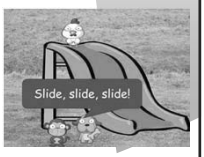
What we will learn

1. **Assessment of CAS & DYS**
2. **Treatment of CAS & DYS**
3. **Ideas for literacy- based therapy materials to encourage repetitive practice**



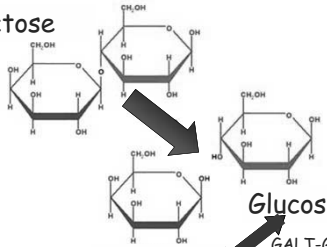
What is classic galactosemia?

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

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

Lactose metabolism

Lactose



Glucose

Galactose

GALT-Galactose-1 phosphate uridyltransferase via the Leloir pathway

http://toresbioclan.pbworks.com/w/page/22377196/Pasteurized%20milk%20Fischman

Prenatal complications of galactosemia

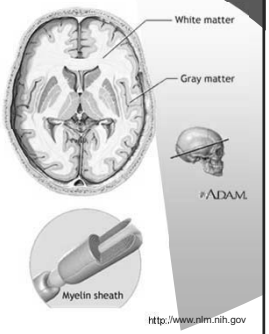
Inability to metabolize galactose
- **Evident at 10 weeks gestation age**
- **Detected in liver and amnio fluid**

Not related to lactose restricted diet by mother during pregnancy

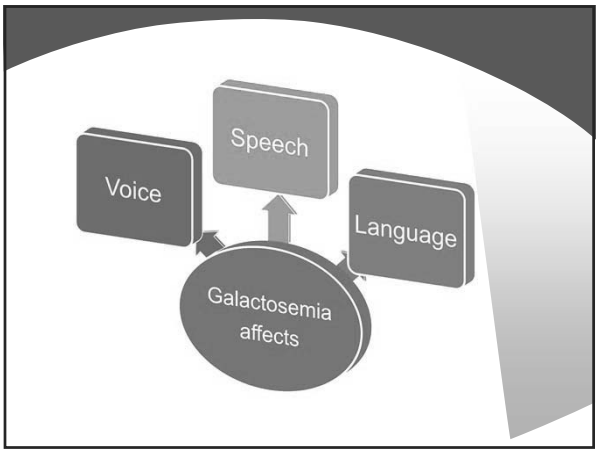
www.sciencemuseum.org.uk/on-line/lifecycle/52.asp

Why does galactose metabolism affect speech?

- **Center of brain is *white matter***
- **White matter = connection fibers (*neurons*) + white fatty covering (*myelin*)**
- **Excess galactose damages myelin → "short-circuits"**



http://www.nlm.nih.gov



What's happening in research?

Childhood Apraxia of Speech Study

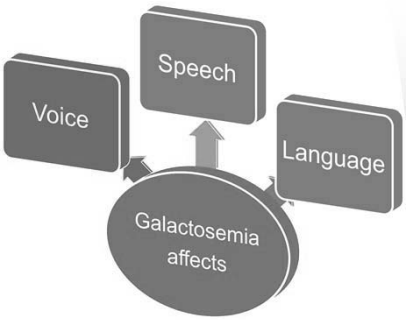
- **Principle Investigator: Lawrence Shriberg**
- **Collaborators: Nancy Potter, Edythe Strand, and other fine folks**
- **Populations include galactosemia, Down Syndrome, Autism, Fragile X, familial genetic disorders**

http://www.waisman.wisc.edu/clinics/staff.html

The Boston Globe

Patients gather in Boston for the largest clinical study ever of a rare disorder

1. **2009**
2. **41 researchers**
   - **From US and Netherlands**
   - **@ Children's Hospital Boston**
3. **Funding**
   - **Parents of Galactosemic Children**
   - **Individual institutions**
   - **Participants paid their own expenses**



Assessing respiration

(As required for adequate voice)

Maximum Exhalation Duration

Maximum Exhalation Duration

1. 5 for 5
   - Blow 5 cm* of water
   - For 5 seconds
   - = adequate breath support for speech (Hixon, 1982)

*5 cm = 2 inches

Assessing voice (phonation)

Maximum Phonation Duration

Assessing respiration-phonation

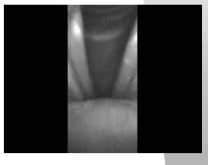
1. Maximum phonation time
   - Sustain "ah"
     - Normal loudness
     - As long as possible
   - Minimum of 8-9 seconds for ages 7-adult
     - For adult length utterances
   - Optimal is 20+ seconds

Test your respiration-phonation

1. Goal = 20 seconds
2. Ready?
3. GO!

Maximum Phonation Duration (Finnegan, 1984 data collapsed across gender)

Age	Mean (seconds)	SD
3	7.1	1.78
4	9.35	2.18
5	10.3	2.81
6	13.9	3.31
7	14.2	2.63
8	17.0	4.57
9	15.7	4.93
10	19.05	5.37



Respiratory-phonatory support for speech in galactosemia

Children ages 4 – 6

- **2/3 of healthy controls (9/30)**

Male age 4 - control

1/8 of children with galactosemia (1/8)

Male age 4 - galactosemia

- **Could sustain "ah" for 8 second minimum**

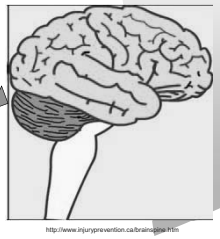
Analysis of children's sustained phonation

1. **Computerized Speech Lab by Kay Elemetrics**
2. **Multi-dimensional voice profile (MDVP)**
3. **Measures 30 parameters**

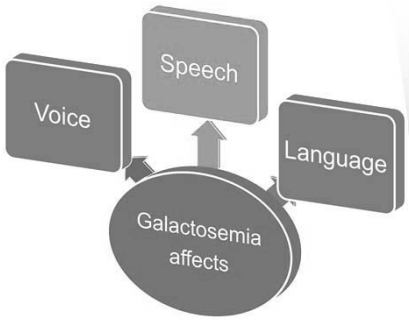
MDVP Results

1/3 of children with galactosemia

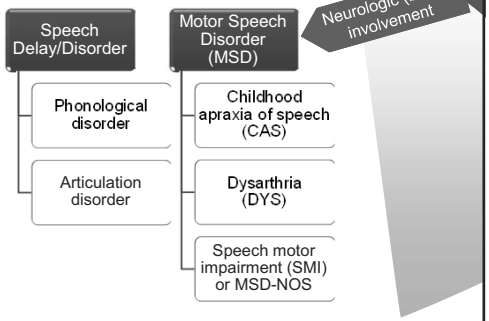
- **Similar to adults and children with cerebellum disorders**



http://www.injuryprevention.ca/brainsignal.htm



Speech sound disorder (SSD) classification system *(Shriberg et al. 2010)*



More or less-agreed upon

3 Features of CAS ASHA (2007)

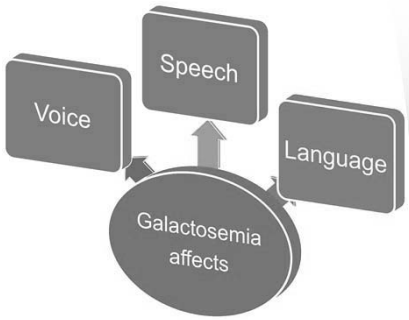
- Inconsistent errors on consonants and vowels in repeated productions of syllables or words
- Lengthened and disrupted coarticulatory transitions between sounds and syllables, and
- Inappropriate prosody, especially in the realization of lexical or phrasal stress.

Features of dysarthria (DYS)

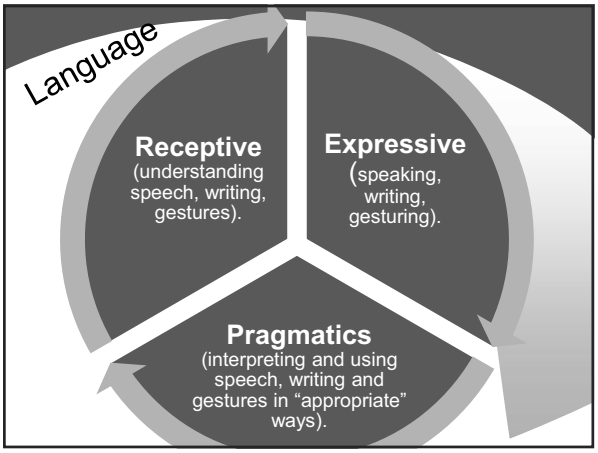
- Decreased strength and coordination
- Low or high tone, poor coordination
- Reduced breath and/or voice support for speech

Well…

IS THERE HOPE?



Describe picture using reading, writing, gesturing & speaking



Language disorder affects 1 or more:

1. Creating words out of sounds
2. Creating gestures or signs out of body movements
3. Modifying words by adding grammatical inflections (jump, jumping, jumped)
4. Combining words grammatically
5. Attaching meaning to words
6. Conversing and telling stories
7. Modifying the language used to suit the situation or the listener

Prevalence of disorders in galactosemia

Disorder	Description	Estimated prevalence
Speech Disorders	180x more prevalent than general population	60%
Motor speech disorders - Pediatrics[1] - (n = 33)	CAS Dysarthria MSD-NOS/Speech motor impairment (SMI)	18% 5% 37%
Motor speech disorders - Adults[2] - (n = 33)	CAS Dysarthria MSD-NOS/Speech motor impairment (SMI)	9% 24% 67%
Respiration/sustained voice[3]	(Say "ah" as long as you can)	58%
Voice quality[3]		33%
Language impairment[4]		90%

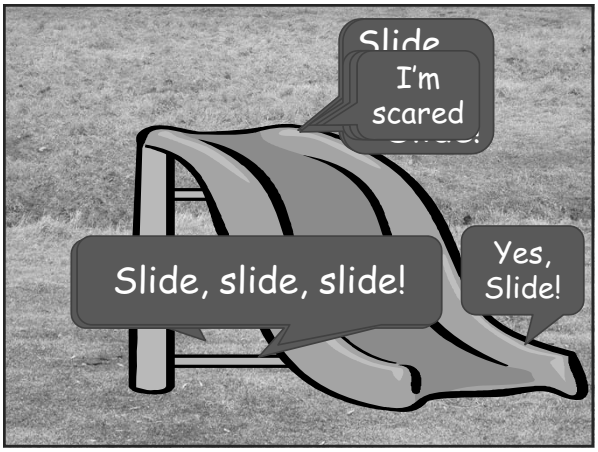
[1]Shriberg et al. (2010); [2]Waisbren et al. (2011); [3]Potter (2010); [4]Waggoner et al. (1990)

Upcoming breakout sessions

Friday @ 1: **Make It, Take It Workshop**

Friday @ 2:15**: Umm, I don't remember: How to teach your child to recall facts**

Saturday @ 10**: Galactosemia and Speech Disorders**



On living with galactosemia

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