

Galactosemia 101

Michelle Fowler, President
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Purpose

Provide information so that you can:

- be and remain positive
- understand the condition
- improve the quality of care
- be informed and be better advocates
- begin interventions earlier
- learn more during the conference

From parents who've "been there" and are now also raising children

- "upbeat, but honest"
- lively and interactive



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Who we are

| | |
|--|---|
| Michelle Fowler | Scott Shepard |
| <ul style="list-style-type: none"> • daughter • galactosemic son • daughter carrier | <ul style="list-style-type: none"> • galactosemic daughter • daughter |



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The fine print



- Not professional medical advice
always consult your healthcare providers(s)
but...
sometimes experts don't know or don't agree
- Solely opinions of the presenters
-  look for this symbol to highlight opinions
- No guarantees
 - Galactosemia Foundation doesn't guarantee accuracy of materials presented at conference



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Topics

- Overview
- Emotional Impacts
- Complications
- Medical Care
- Diet
- Seeking a Cure
- Q&A / Discussion / Wrap-Up



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Topics

- Overview
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- Complications
- Medical Care
- Diet
- Seeking A Cure
- Q&A / Discussion / Wrap-Up

- History
- Genetics
- Metabolism
 - Normal
 - Galactosemic



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Timeline

2011 PGC becomes **Galactosemia Foundation**

1980s & 1990s research indicated ongoing medical and developmental problems

1966 newborn screening began

1985 Support group Parents of Galactosemic Children (PGC) launched

1956 specific enzyme defect identified

1908, 1917 symptoms first noted

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Progress

- our kids are diagnosed earlier through newborn screening
- our kids survive, live better, and have better health!
- it is still *early* in finding a viable treatment

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Genetics of Galactosemia

- genes control our traits
 - hair color, eye color, height, etc.
- we have 2 genes for each trait (inherited)

1 gene from the father

1 gene from the mother

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Genetics of Galactosemia

N = Normal gene
G = Defective gene

4 possibilities for *each* baby

father (carrier) mother (carrier)

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Genetics of Galactosemia

4 possibilities for *each* baby conceived by two galactosemia carriers:

| | | |
|-----------|---|-----|
| NN | Normal GALT production | 25% |
| NG | Carrier (reduced but sufficient GALT production) | 50% |
| GN | | |
| GG | Galactosemia (no GALT production) | 25% |

1 in 4 your next baby has galactosemia
consult a genetic counselor

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Genetics of Galactosemia

- Genes are responsible for disorders, too
- Imagine a gene as a sentence:
 - The sun was hot.
- Defects (mutations) change genes:
 - **by deletion:** The was hot.
 - **by substitution:** The dog was hot.
 - and other changes
- Defects change the meaning of the sentence

Adapted from : Genetic Science Learning Center (1969, December 31) What is a Mutation?.
Learn.Genetics from <http://learn.genetics.utah.edu/archive/mutations/index.html>

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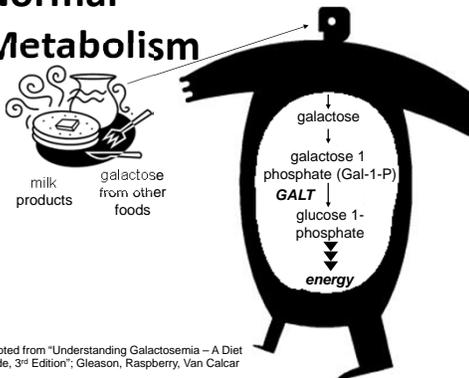
Galactosemia Gene Defects

- Many defects result in GALT deficiency
 - defects named with letters and numbers
e.g. Q188R, L195P, etc (called *genotypes*)
 - represent the type and location of the defect
 - each galactosemic inherited 2 defects (♂♀)
 - same defect = homozygos
 - different defects = heterozygos
 - We don't know relation between any specific defect, or pair of specific defects, and outcome

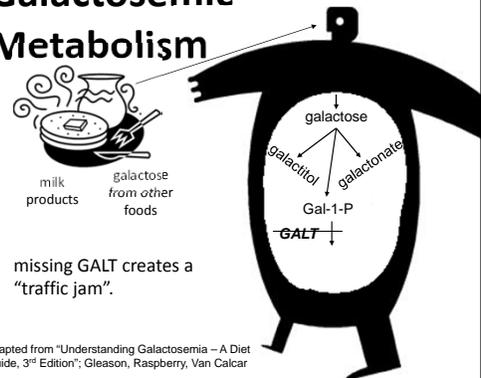
Genetics of Galactosemia

- It's **not** your fault!
- Genes determine traits. They are inherited from **both** parents equally.
- Carriers show no symptoms of galactosemia.
- It's not contagious or 'catchy'.
- Knowledge is power! Gain more and empower yourself!

Normal Metabolism



Galactosemic Metabolism



Metabolism

- GALT enzyme missing or not working
 - called an "inborn error of metabolism"
 - liver, not stomach
 - metabolism not digestion
- "Traffic Jam" in galactose metabolism
 - Gal-1-P, galactose, etc. build up in the body
 - limit galactose and lactose to minimize build-up

Topics

- Overview
 - Emotional Impacts
 - Complications
 - Medical Care
 - Diet
 - Seeking A Cure
 - Q&A / Discussion / Wrap-Up
- Personal
 - Family
 - Siblings
 - Extended Family
 - Marital

Personal Emotions

Denial – Grieving – Acceptance

- Don't skip steps
 - go through them as fast as you can
- Respect your emotions
 - don't be a slave to them
- Don't judge others who don't feel what you feel



Don't let galactosemia define your life

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Emotional Aspects - Family

Siblings

age appropriate explanation



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Emotional Aspects - Family

Extended family

- don't expect them to "get it" - *ignore nonsense*
- try to educate them, especially if you need them
- know when to stop trying
 - *you can't control people's actions*
 - *you can only control your reactions*



Children learn more from your reactions than the actions of others

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Emotional Aspects - Martial

- Do your part
- Help each other
- Accept and respect that your spouse might be handling this differently



Don't let galactosemia define your marriage

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Topics

- Overview
- Emotional Impact
- Complications
 - Newborn
 - Speech & Language
 - Learning Disabilities
 - Fine/Gross motor
 - Other
- Medical Care
- Diet
- Seeking A Cure
- Q&A / Discussion / Wrap-Up

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Beyond Newborn Complications

- You made it through:
 - Not knowing why your new baby was sick
 - feeding difficulties, "failure to thrive", jaundice, infection, etc.

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The Future

- Will our kids grow up to be happy, successful, independent adults?
 - We don't know
 - Wide range of outcomes
- Potential complications
 - Not all galactosemics have every (any?) complication
 - Higher risk than average
- What can you do?
 - Be your child's advocate!
 - Early intervention
 - Get involved / fundraise for research



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Speech & Language

- High risk for speech & language difficulties
 - not typical speech *delay*
 - motor planning disorder
 - childhood apraxia of speech (CAS)
 - verbal dyspraxia
 - *often responsive to speech therapy*
 - *may need more frequent, 1:1 therapy*
 - *'typical' speech therapy may not be effective*
- What can you do?
 - observe your child (see checklists)
 - have your child evaluated
 - Advocate for your child!



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Learning Disabilities

- Higher Risk for difficulties in school
 - May need more repetition
 - May need 1:1 attention
 - Multi-sensory approaches
- What can you do?
 - Advocate for your child!
 - Learn about IDEA
 - Federal Law: Individuals with Disabilities Education Act
 - Galactosemia qualifies as "other health impairment"
 - Early intervention state programs (free) (IDEA Part C)
 - Have your child evaluated (free) at age 3 by your school district (IDEA Part B)



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Fine and Gross Motor

- Higher risk for fine and gross motor difficulties
 - ataxia / motor planning disorders
 - inability to coordinate muscle movements
- What can you do?
 - have your child evaluated
 - occupational therapy for fine motor
 - physical therapy for gross motor



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Other Complications

- Tremors
 - may develop later (as young adults)
 - mostly hand / arm tremors, but also head and body
- Reproductive
 - Premature Ovarian Failure (POF) for women
 - Menopause before age 40
 - Affects most galactosemic women
 - Some pregnancies reported!
 - Reproductive difficulties for men too



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Possible (?) Others

- NOT confirmed by scientific studies
- Feeding Difficulties
- Sensory Integration
 - noises, textures, etc.
- ADHD



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Topics

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- Emotional Impacts
- Complications
- Medical Care
- Diet
- Seeking A Cure
- Q&A / Discussion / Wrap-Up

- Doctors
 - What kinds
 - How often
- Testing
 - What kinds
 - How often

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! Build your child's "team"

- You are the expert on your child
- Advocate for your child
 - Services may not be offered if you don't ask
 - Have child evaluated for potential complications
 - Speech and language
 - IDEA (Individuals with Disabilities Education Act)
 - Part C – Infants and Toddlers (<3 years old)
 - State "early intervention" programs
 - Free
 - Part B – School age (3 – 21 years old)
 - School district
 - Free

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IDEA Resource: From Emotions to Advocacy, Pam and Pete Wright
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Pediatrian

- Your child's medical "home base"
 - select a good one that will work with you
- ! You have to educate them
 - may have heard of galactosemia in med school
 - point them to resources that you know about
 - remind them about galactose in medicines
- Enlist them as an ally
 - referrals to specialists / prescriptions for therapies/ written letters when needed

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Geneticist / Pediatric Metabolic Specialist

- Specialist in genetics & metabolism
 - Will know more about galactosemia
 - You may still have to educate them
 - Monitor galactose levels (Gal-1-P, etc)
 - Clinic team (dieticians, genetic counselors, social workers)
- How often?
 - 3 to 4 times per year first two years
 - Twice per year until age 6
 - Annual until age 18 (?)

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Metabolic Dietician(s)

- Provide guidance on diet issues
 - ! You may have to educate them on galactosemia
 - Not all metabolic clinics agree on galactosemia diet
 - General guidance on formulas and food
 - Help identify safe food substitutes
 - Guidance on calcium, vitamins and minerals
 - Help identify safe supplements
 - Help with social aspects of feeding
- How Often?
 - During metabolic clinic visits
 - Consult as needed

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Developmental Pediatrician

- Specializes in evaluating development
 - Wide range of 'normal' development
 - Has more testing 'tools' than normal pediatrician
 - Help you detect problems and get early intervention
- How often?
 - Two times per year first two years
 - Annually, as needed

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Other Medical Professionals

- Endocrinologist (hormones/growth)
- pediatric gynecologist (girls ~9 years)
- ophthalmologist (sugar cataracts)
- dentist (higher sugars in milk substitutes)
- neuropsychologist
- psychologist
- neurologist

Typical Metabolic Clinic Visit and Testing

- Laboratory tests
 - a) GALT activity levels
 - b) other enzyme analysis as indicated
 - c) Gal-1-P levels at first evaluation
- Nutritional consultation
 - minimize galactose in diet
 - interpret diet prescription
- Social services assessment

Testing: Gal-1-P

What do results mean?

- low Gal-1-P *indicates* good dietary compliance
- elevated Gal-1-P = cause for investigation
 - not necessarily indicative of poor compliance
 - newborns often take several months or more to have their levels stabilize at a low level
 - some children have levels that do not correspond to diet

Testing: DNA

DNA blood test

- ⚠ – once, maybe
- of no therapeutic value (currently)
- ⚠ – maybe of help to research?
- often not covered by insurance

Topics

- Overview
- Emotional Impacts
- Complications
- Medical Care
 - Diet
- Seeking A Cure
 - Newborn
 - Babies
 - Toddlers
 - Preschool and beyond
- Q&A / Discussion / Wrap-Up

Managing Galactosemia Diet In Your Home

- Decide about your household
 - strongly recommend:
 - ⚠ *Integration / Not Isolation*
 - eating is a social function
 - psychological - don't accentuate differences
- Substitutes
 - recipes
 - specialty foods
 - lactose intolerance/ milk allergies / vegans

Newborn Diet

- NOT OK: mother's breast milk or milk based formula
- Convert to a formula with little or no galactose
 - soy formula
 - all OK: **powder, concentrate, ready-to-feed**
 - elemental formulas
 - \$\$\$ but may be appropriate (example: soy allergy)
- Insurance coverage of medically necessary formulas to treat disorders identified through newborn screen:
 - state-by-state coverage (check statutes)
 - never easy, but worth it
 - **save those receipts!**
- Take time to *start* learning - **slowly**

Babies - Beyond Formula

- Prepare before you are ready to start solids
 - metabolic dietitian
 - written guidelines
 - Web resources
- Start **READING LABELS**
 - unacceptable Ingredients lists
 - questionable / restricted / limited foods
- Baby food lists
- Start **TEACHING**
 - yes food / no food

Toddler Diet

- Introduce variety and substitutes
 - toddler formulas, soy milk, rice milk, etc.
 - fruits and vegetables OK!
 - some aged cheeses OK!
- Continue teaching through play
 - **yes** food / **no** food
 - red stickers / green stickers
- Childcare:
 - provide written guidance and supplies
 - communication is key

Pre-school and Beyond

- Communication is key
 - parties, special events, field trips, etc.
 - provide written guidance
- Continue teaching
 - goal: your child makes **safe and good** independent diet decisions
- Accidents happen
 - **don't panic**
 - investigate and take corrective action

Topics

- Overview
- Emotional Impacts
- Complications
- Medical Care
- Diet
- Seeking A Cure
 - Path to Cure
 - Alternatives
 - Get Involved!
- Q&A / Discussion /

What is A Cure?

- ability to have an unrestricted diet?
- elimination of medical and developmental complications?



A Cure - Paths

- First we have to understand
 - What causes the problems?
 - When those problems arise?
 - Are problems reversible or only preventable?
- To understand we have to study galactosemics
 - Need big studies (2000+) to detect small differences
 - Biggest studies so far <500 people

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A Cure – Beyond Galactose Restriction

- Tissue or organ or cell transplants?
 - Very risky; timing?
- Internal enzyme infusion?
 - Not clear that would help
- Gene therapy?
 - Very risky; timing?
- Medicines to activate or inhibit internal processes?
 - Need to know the targets, locations, and timing?
- Additional dietary supplements?
 - Which ones, how much, and timing?

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A Cure - Steps

- Human testing
- Is path to 'cure' worse than the 'disease'?
- \$\$\$\$\$\$
 - better spent elsewhere?

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Alternatives...

- More research into better management?
- Researching treatments for complications?

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Get Involved!

- Nothing feels as good or provides as much benefit as being part of a community of people

Others can help you
You can help others

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- Q&A / Discussion / Wrap-Up

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Q & A / Discussion

What did We miss?

Consider linked in Facebook Twitter galactosemia website discussion site

question is the all sites do you acknowledge connect to your all groups?

Board of Directors.... We are always here to help! So, GF's website discussion forum, is an excellent way to connect with others and share life experiences.

GF President – Michelle Fowler – president@galactosemia.org
GF Vice President – Scott Shepard – vicepresident@galactosemia.org

What you can get each today

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Unacceptable Ingredients

| | |
|------------------------|--|
| Butter | Dry Milk Protein |
| Nonfat Milk | Yogurt |
| Milk | Organ Meats |
| Nonfat Dry Milk | (liver, heart, kidney brains, sweetbreads, pancreas) |
| Buttermilk | Sodium Caseinate |
| Cream | Calcium Caseinate |
| Milk Chocolate | Tragacanth Gum |
| Cheese | Lactostearin |
| Buttermilk Solids | Lactalbumin |
| Nonfat Dry Milk Solids | |
| Milk Derivatives | |
| Dried Cheese | Dough Conditioners * |
| Milk Solids | Hydrolyzed Protein ** |
| Lactose | Margarine *** |
| Casein | MSG (Monosodium Glutamate)**** |
| Sour Cream | Soy Sauce ***** |
| Dry Milk | |
| Whey and Whey Solids | |

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Speech & Language

Checklist for speech delays

- Demonstrates oral inactivity
- Drooling after 15 months
- Minimal interest in talking
 - not attempting to talk at any age >18 months
- Not talking at all by age 2
- Omits many consonants
- Largely unintelligible speech or uses mostly vowel sounds, especially after age 3
- Trouble producing many speech sounds
- Substitutes many sounds for others
 - not age appropriate substitutions
 - major random substitutions
- Has inaccurate speech sound pronunciation
- Consistently drops word endings after age 5
- Exhibits a slow labored rate of word pronunciation
- Exhibits rapid skurred speech
- Minimal interest in communication, verbal or nonverbal
- Not attempting to talk, any age >18 months
- Not using phrases by 2 1/2 - 3
- Not using sentences by age 3
- Does not follow 2-step instructions by age 2
- Embarrassed, disturbed, or uncomfortable about speaking at any age
- Difficulty organizing or expressing an idea or thought

Checklist for language delays

- Excessive imitation / limited spontaneous production
- Often "searching" for words / using "empty" vocabulary
- Often talks "around" a subject
- Difficulty following 2-4 step instructions by 3 or 4
- Difficulty telling/reading story in sequence by 4
- Noticeably faulty sentence structure at 5
- Grammatically correct responses but not appropriate to a situation
- Difficulty learning letters, numbers, sounds, rhymes, and reading

Checklist for Voice

- Excessive nasal, hyponasal, or denasal voice
- Strained, harsh or hoarse voice
- Including evidence of frequent screaming
- Breathily voice
- Voice pitch too high/low for age, size, gender
- Minimal variation in intonation
- Voice Volume
 - Use excessively loud voice
 - Consistently speaks too softly not from shyness

Checklist for Fluency

- Excessive repetition of sounds in words (e.g. "p-p-p-p-p-p-p")
- Frequent prolongation of sounds in words (e.g. "sssssoap")
- Excessive hesitations, pauses, and/or hemming and hawing

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Adapted from Source: Patricia Launer, Ph.D.