Navigating Galactosemia Life Stages

A Handbook for the Galactosemia Community

2022 Edition

Galactosemia Foundation
Linked for Life

JAGUAR GENE THERAPY

This galactosemia handbook presents current information for each life stage, including various symptoms that may appear. Cited recommendations are based on the international clinical guideline, peer-reviewed data, expert insights, and first-hand experience from the galactosemia community.
As you review this handbook, please keep in mind that every person develops differently. Consult with your or your child's care team regarding any specific concerns you have or any questions regarding potential therapeutic options or approaches. The information presented here is not meant to replace advice from a healthcare provider and does not constitute a medical diagnosis or advice.

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This document was developed by the Galactosemia Foundation and Jaguar Gene Therapy, with support from experts in galactosemia and members of the galactosemia community.

Version 1.0.

Acknowledgments

The development of a comprehensive guidebook such as this is no small task. The Galactosemia Foundation and Jaguar Gene Therapy would like to thank all those whose expertise supported the development of this important tool, including:

- Members of the galactosemia community:
  - Parents of children living with galactosemia
  - Individuals living with galactosemia

All **PARENT TIPS** come from these parents as well as the Galactosemia Foundation.

- Experts in galactosemia, who are referenced throughout this document:
  - Gerard T. Berry, MD
  - Can Ficicioglu, MD, PhD
  - Judith L. Fridovich-Keil, PhD
  - Lauren E. Hitchins, DNP, CPNP, AGN-BC
  - Quinton Katler, MD, MSc
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  - Jessica B. Spencer, MD, MSc
  - Sandy van Calcar, PhD, RD, LD
  - Susan E. Waisbren, PhD

All **TIPS FROM THE EXPERTS** are from these experts.

The photos you will see throughout are of actual members of the galactosemia community. We are grateful to the families who shared these images and authorized their use in this document.

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Introduction

Introduction

This handbook provides information about galactosemia, an inborn error of metabolism. It includes insights from families, medical professionals, galactosemia researchers and experts, and published research. This resource was developed for the galactosemia community, and also for teachers, psychologists, social workers, or others who may encounter someone with this rare metabolic disorder.

This handbook is intended to provide you with knowledge and information and does not constitute a medical diagnosis or advice. The guideline recommendations and literature references mentioned are intended to serve as a starting point in understanding your or your child's diagnosis. It is important to remember every guideline recommendation may or may not be applicable to your or your child's experience, as one person with galactosemia may experience the disease differently than others.

Because everyone's healthcare coverage will vary, it is possible that not all recommendations will be covered by all insurance plans. Please check with your providers to understand your coverage.

This galactosemia handbook presents current information for each life stage, including various symptoms that may appear. Cited recommendations are based on the international clinical guideline, peer-reviewed data, expert insights, and first-hand experience from the galactosemia community.
As you review this handbook, please keep in mind that every person develops differently. Consult with your or your child's care team regarding any specific concerns you have or any questions regarding potential therapeutic options or approaches. The information presented here is not meant to replace advice from a healthcare provider and does not constitute a medical diagnosis or advice.

Galactosemia Foundation
Linked for Life

A Message from the Galactosemia Foundation

This guide does a remarkable job providing you with education on galactosemia and tools to help ensure the best care moving forward, along with practical ways to advocate for the needs of someone living with galactosemia. Being educated and able to advocate are vital, but one piece that a guide cannot provide is connection to community. We know that receiving this diagnosis can be daunting at first. Connecting with those in the community—those who have walked in your shoes—can make it a little less overwhelming. You can easily ask advice of others, find families in your area with whom to build relationships, and you've always got a listening ear of someone who truly understands. Connection with others can be the peace you need from someone who shares your same diagnosis and in return you may be part of bringing that peace to someone in the future. While this may not be what you had planned, we live this journey together, and welcome you to be a part of it. For more information, please e-mail us at outreach@galactosemia.org.

Galactosemia Foundation Conference

Every 2 years, the Galactosemia Foundation organizes a conference for families, researchers, and clinicians, solely focused on galactosemia. It is a great place to learn more, meet other families, get involved, and hear about the latest research from experts in galactosemia. They serve safe foods, provide childcare for ages 0-8 years, and create age-specific content for ages 9 and older, as well as for parents. Some of the conferences have both live and virtual components and they typically fall on even-numbered years.

Look for this icon in each section for corresponding age-specific information on activities at the conference.

Nicole Casale, *President*

Brittany Cudzilo, *Vice President*

Note: This is reflective of the Galactosemia Foundation's leadership as of 2022.

How to Use This Handbook

This handbook was developed to help support you in your understanding of galactosemia and what to expect at different life stages. We have broken the information down into stages beginning at birth, but it is important to remember each person is different. You or your child may or may not experience symptoms, and if symptoms do develop, they may develop at different times. The hope is this knowledge will help prepare you for navigating your or your child's care.

This handbook was developed using current research and clinical literature available as of April 2022 and draws from multiple sources of published data, including clinical guidelines, registries, and expert-led medical journals, as well as insights from families navigating life with galactosemia. Specifically:

- **International Clinical Guideline for the Management of Classical Galactosemia: Diagnosis, Treatment, and Follow-Up:** This guideline was published in 2016 by members of the Galactosemia Network (GalNet) and was developed by a panel of 21 experts from around the world to provide a consistent set of recommendations for the diagnosis, treatment, and follow-up for people with galactosemia.[1] Recommendations from this guideline are discussed within relevant life-stage chapters.

- **GalNet Registry:** Registries are a valuable source for understanding how diseases affect the lives of people living with a disease. The data gathered from registries can provide information about when and how often people experience various symptoms and complications. The Galactosemia Network developed a registry in 2014 (GalNet Registry).[2] The published registry data from 2019 includes information about the outcomes of 509 (48.1% male and 51.9% female) participants with galactosemia.[2] Of the 255 participants with a documented GALT activity test, approximately 83% had classic galactosemia (GALT enzyme activity ≤1%) and 17% had clinical variant galactosemia (GALT enzyme activity between 1% and 10% based on laboratory data collected from historical medical records).[2] At time of publication, this is the largest published study describing cross-sectional outcomes of people with galactosemia.

- **Expert-Led Medical Journals:** Publications written by leading researchers and experts in galactosemia.

Please refer to the ***References*** section at the back of this handbook for a comprehensive list of sources used in development of the content.

We recommend reading this ***Introduction*** section in its entirety, as well as ***The Galactose-Restricted Diet (Standard of Care)*** chapter, and then navigating to the age-range chapter relevant to your needs. There are helpful resources and a Glossary at the end of the guide.

Throughout this handbook, callout styles will be utilized to categorize certain types of information for ease of use and quick reference.

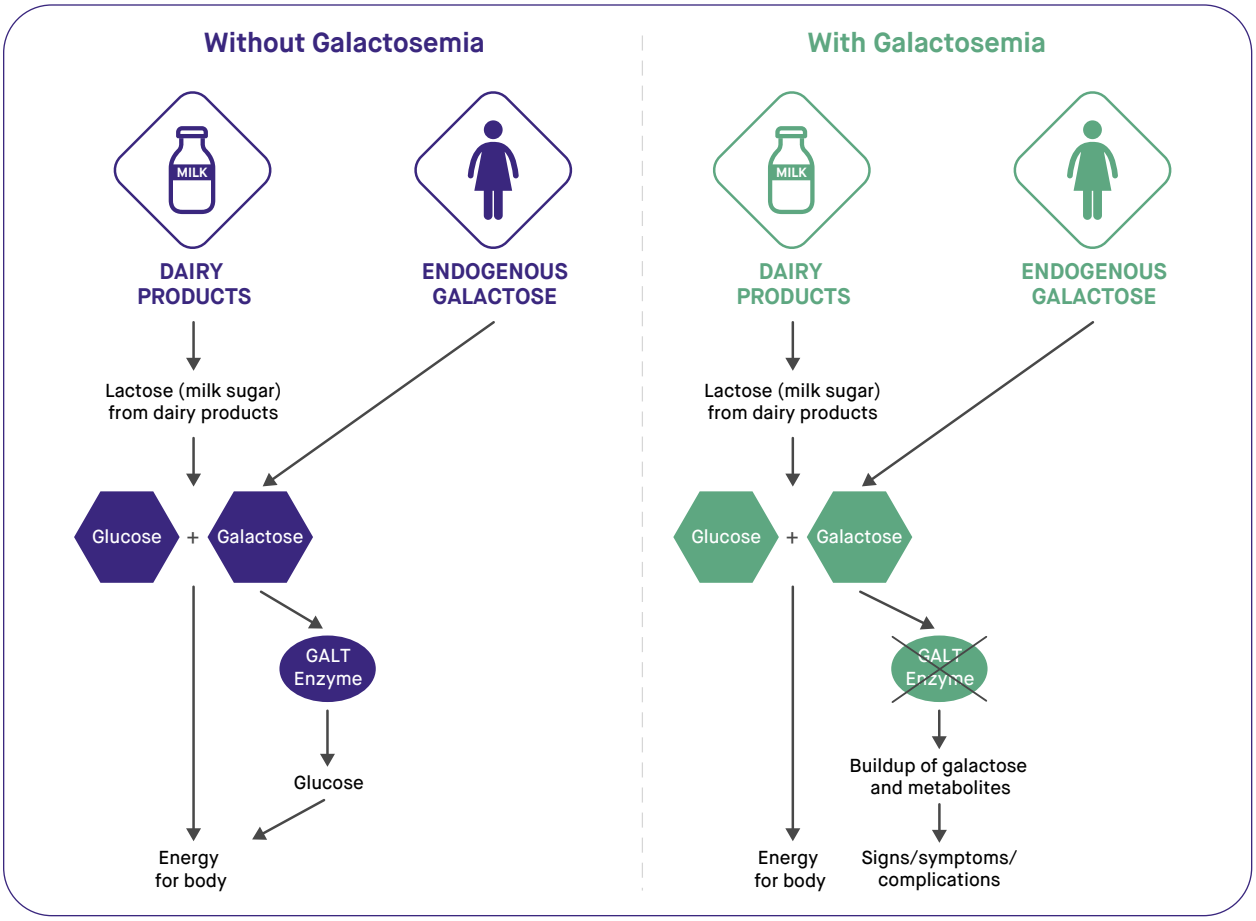
What Is Galactosemia?

Galactosemia is a genetic disorder that affects the body's ability to break down galactose. The inability to break down galactose in milk or formula at birth can be life-threatening for newborns. In the immediate newborn period, substituting non-dairy formula for breast milk or milk-containing infant formulas is critical.[3]

Type 1 galactosemia is caused by mutations in the *GALT* gene. The *GALT* gene tells the body's cells how to make the GALT enzyme, which helps the body break down or process galactose.[3] Galactose is a sugar that is produced in the body naturally, or endogenously, and is also found in dairy, breast milk, and other foods. This sugar is used in the human body as a means of energy production in your cells and as a building block for other cellular components, making it essential for human life.[4] Genetic mutations in the *GALT* gene impact whether the body can produce functional GALT enzyme. Without sufficient functional GALT enzyme, a buildup of galactose occurs, which results in the accumulation of additional substances or metabolites, including galactose-1 phosphate (Gal-1P), and galactitol. Too much galactose, Gal-1P, and galactitol in the body is considered toxic and may contribute to lifelong complications.[3,5]

Type 1 galactosemia includes classic, clinical variant, and Duarte galactosemia, because they are all caused by mutations in the *GALT* gene. However, symptoms of galactosemia typically only occur in classic and clinical variant galactosemia.[6,7] Because experts agree following a galactose-restricted diet is not necessary for Duarte galactosemia,[1] this handbook is intended for use by families managing classic and clinical variant galactosemia. Any future mention of Type 1 galactosemia or galactosemia assumes a GALT deficiency that causes clinical symptoms and excludes Duarte galactosemia.

Figure 1: Galactose is an essential sugar that is either created within our own bodies or acquired through the foods we eat (such as dairy products). Galactose in our bodies is broken down into glucose which provides energy for the body. One of the enzymes responsible for galactose breakdown is GALT. When the GALT enzyme is missing or deficient, the buildup of galactose and its metabolites is thought to contribute to the symptoms associated with Type 1 galactosemia.[3]



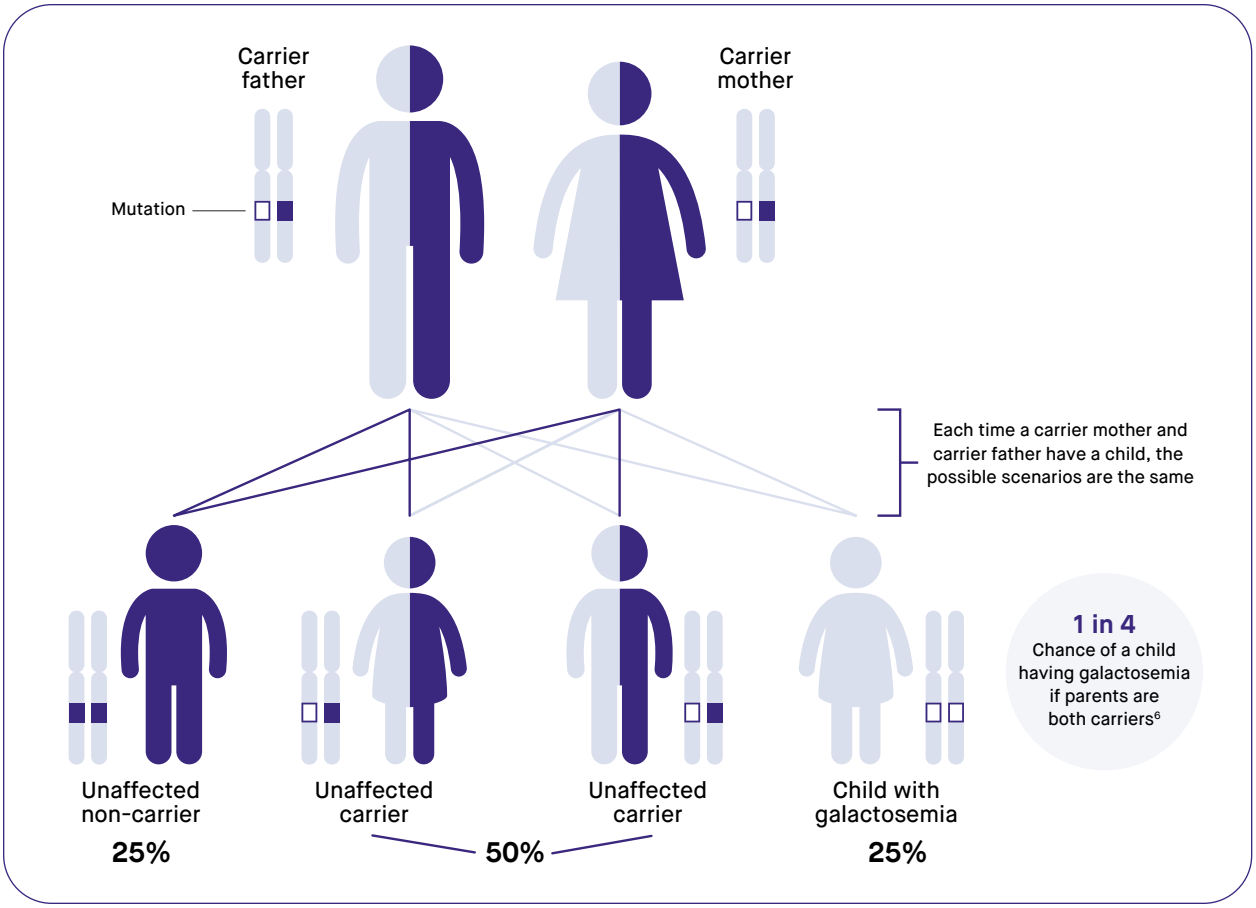
Classic galactosemia, the disorder caused by the absence or near-absence of GALT activity, is associated with cognitive, motor, and other neurological challenges.[2,6] Most people with galactosemia experience speech and language delays, disorders, and/or impairments.[2] Girls and women usually experience premature ovarian insufficiency (POI) and possible infertility.[2,6] For people with residual enzyme activity, as in clinical variant, the effects may be less severe. The risk and severity of galactosemia may depend on the types of mutations in the *GALT* gene and their resulting level of residual GALT enzyme activity, but scientists and doctors do not yet understand all of the factors that contribute to symptom presence and outcome severity.[1,6] In both classic and clinical variant galactosemia, insufficient levels of functional GALT enzyme are produced. The typical GALT enzyme activity levels for classic and clinical variant galactosemia are listed in **Table 1**.[6]

Table 1: Residual GALT enzyme activity and common genetic mutations in people with Type 1 galactosemia[6]

Type 1 galactosemia variant	Residual GALT enzyme activity	Common/known genetic mutations
Classic	Absent or barely detectable (0% to <1%)	Q188R, K285N, L195P, Δ5.2kb del
Clinical variant	1% to 10%	S135L, T350A,[8] Y323D,[8] V151A,[8] R67C,[8] L139P,[8] M219K,[9] c.1-96T>G,[9] V128I,[9] R201H[9]

Galactosemia is an autosomal recessive disease.[6] This means the parents of a child with galactosemia are almost always carriers of the genetic mutation. Everyone has two copies of each gene (alleles) which we inherit from our biological parents. Carriers for galactosemia have one allele that produces functional GALT enzyme and one allele that produces non-functional GALT enzyme due to a mutation. Carriers do not experience symptoms of the disease. Children are born with galactosemia when they inherit two copies of the affected gene, one from each parent, each with a mutation that produces non-functional or less-functional GALT enzyme (see **Figure 2**).[6]

Figure 2: How a child is born with galactosemia



Current Standard of Care

The standard of care for galactosemia is a galactose-restricted diet. By starting a galactose-restricted diet, severe symptoms that present in the newborn period can resolve or be avoided.[1] These include liver failure, severe infection (sepsis), blood clotting issues, and, if not diagnosed, death. A galactose-restricted diet is typically initiated immediately after newborn screening results indicate a possible diagnosis of galactosemia.[1]

Once the critical newborn period is over, a long-term treatment plan begins.[1] This involves avoidance of dairy products and other foods containing even small amounts of milk products.[1] However, because our bodies produce galactose naturally (endogenous galactose) and people with galactosemia are unable to properly break down galactose, lifelong symptoms can still occur even when the diet is followed.[1] Metabolic specialists hypothesize this endogenous galactose may contribute to cognitive, neurological, and other health issues that can develop with galactosemia.[6,10] More details on the diet itself can be found in the ***Galactose-Restricted Diet (Current Standard of Care)*** chapter.

> "I thought if we followed the diet,
> he would have zero issues.
> I didn't realize until months later
> that wasn't the case.
> We didn't know he was still naturally
> producing galactose—it was like
> brand-new shocking information to me
> that I wish I knew sooner."
>
> *– Parent of a 2-year-old with galactosemia*

Treatments in Development

Because a galactose-restricted diet does not fully address the underlying biological issues with galactosemia and does not prevent lifelong symptoms or complications, researchers are developing potential new treatments. Learn more by visiting the Galactosemia Foundation website at www.galactosemia.org or entering "galactosemia" as a search term at www.ClinicalTrials.gov and discuss with your care team.

Care Team

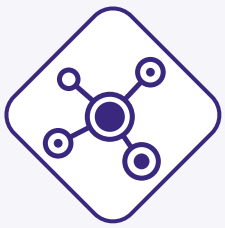
Each person with galactosemia may experience different symptoms. This difference in presentation and severity of symptoms means galactosemia is a heterogeneous disease. One person with galactosemia may experience the disease differently than others.[1] A person with galactosemia may see all or some of the following specialists, depending on their needs. Studies are ongoing to collect more data about the natural course of the disease across individuals.

PEDIATRICIAN

A pediatrician is trained to diagnose, treat, and provide ongoing care for a variety of childhood illnesses, from mild to severe. For a baby with galactosemia, this doctor plays an integral role in their care. The pediatrician may refer a family to specialists for many of the symptoms a child may experience. In addition, the pediatrician will keep track of growth rates, measuring height and weight at all milestone visits.

NEONATOLOGIST

A neonatologist specializes in the care of newborns with serious illnesses. Some babies with galactosemia will experience complications such as liver failure and sepsis following birth and be cared for in a neonatal intensive care unit (NICU).

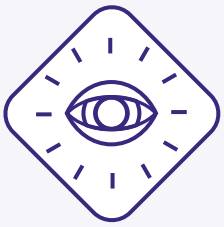


METABOLIC GENETICIST

A metabolic geneticist specializes in the diagnosis and management of metabolic disorders. They help confirm the genetic diagnosis of galactosemia when a newborn's screening test is abnormal and may remain a part of a long-term care team.

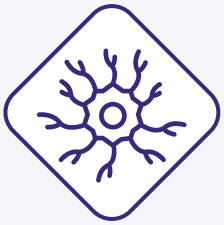
DIETICIAN

A dietician can provide guidance on the galactose-restricted diet. For infants, they can identify appropriate substitutes for dairy-based formula or breast milk, and for children and adults, what kinds of foods are safe to eat. A dietician may also help monitor levels of certain vitamins, like vitamin D, to ensure adequate nutrition.



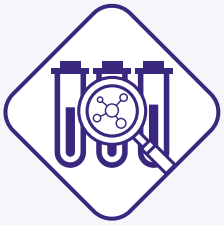
OPHTHALMOLOGIST

An ophthalmologist is a medical and surgical specialist who focuses specifically with the structure, function, diseases, and treatment of the eyes. It is recommended an ophthalmologist check for cataracts, a known complication of galactosemia. This includes screening shortly after birth and monitoring people with galactosemia over time.[1]



NEUROLOGIST

A neurologist is a medical doctor with specialized training in the diagnosis, treatment, and management of disorders of the brain and nervous system. They may help treat symptoms such as motor dysfunction, tremors, and seizures, which are experienced by some people with galactosemia.[2]



ENDOCRINOLOGIST

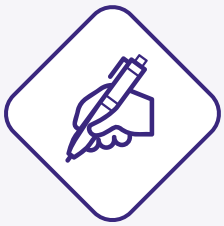
An endocrinologist is a doctor who specializes in the diagnosis and treatment of hormone-related diseases and conditions. They can help to manage primary ovarian insufficiency (POI), which is common for girls and women with galactosemia.[1] Depending on the medical center and geographic location, you may also coordinate care with a pediatric or adolescent gynecologist or obstetrician/gynecologist (OB-GYN). Many girls see an endocrinologist initially and transition to an OB-GYN when they become a teenager. Adults may consult with a reproductive endocrinologist for ongoing hormone replacement therapy or fertility issues. Endocrinologists can also support management of bone health in girls (women) and boys (men).[11]

SOCIAL WORKER

A social worker can support people with galactosemia and their families through difficult times post diagnosis, with the goal of improving outcomes. They act as guides and advocates for families. A social worker can also support families with insurance issues and help to access services such as early intervention, special education, adaptive housing, and paying bills related to care.

SPEECH-LANGUAGE PATHOLOGIST

A speech-language pathologist conducts evaluations to assess feeding, swallowing, speech, and language. They also work with families on various therapeutic interventions related to speech and language to help prevent or address any issues identified. Speech and language complications are common for people with galactosemia.[2]



OCCUPATIONAL THERAPIST

An occupational therapist can help assess any issues interfering with conducting activities needed for daily living. They also work with the child and family on development of techniques to overcome barriers related to feeding problems, sensory issues, and fine motor challenges (e.g., holding a pencil, buttoning clothing).

PHYSICAL THERAPIST

A physical therapist can help assess any movement and coordination challenges and provide therapeutic support if necessary. People with galactosemia can experience coordination and movement challenges, as well issues with gross motor skills[2] (e.g., standing, walking, running).

PSYCHOLOGIST

A psychologist is specially trained to evaluate development, educational achievement, and functioning, and to provide counseling. Children with galactosemia often experience some developmental delays and some may have behaviors on the autism spectrum (although a diagnosis of autism is rare).[2]

PSYCHIATRIST

A psychiatrist is a medical doctor with a specialty in mental health. Psychiatrists are consulted when medication is needed. Some psychiatrists also provide counseling. People with galactosemia may experience behavioral and psychosocial challenges such as anxiety, depression, and attention-deficit hyperactivity disorder (ADHD).[2]

TIPS FROM THE EXPERTS

UNFORTUNATELY, ACCESS TO PSYCHOLOGICAL SERVICES CAN BE DIFFICULT TO SECURE. WHILE ADVOCATING FOR YOURSELF OR YOUR CHILD WITH INSURANCE CARRIERS, ALWAYS PROVIDE THE MEDICAL RATIONALE FOR THESE SERVICES. IT IS IMPORTANT TO DOCUMENT THE NEED FOR ACCURATE ASSESSMENTS TO TAILOR TREATMENT AND SUPPLEMENTAL THERAPIES IN ACCORDANCE WITH THE MEDICAL GUIDELINES.

The Galactose-Restricted Diet (Current Standard of Care)

The Galactose-Restricted Diet (Current Standard of Care)

The major sugar found in milk and milk products is lactose. Lactose is comprised of glucose and galactose.[3] Once a person's body has utilized the necessary amount of galactose, excess galactose must be broken down to prevent it from accumulating. Excess galactose is ultimately broken down and used to generate glucose which is a major energy source for the body.[3] People with galactosemia lack the essential GALT enzyme required to break down galactose.[3] Therefore, the standard of care for people with galactosemia is a galactose-restricted diet,[1] which is outlined in this chapter.

The international clinical guideline provides guidance for this diet.[1] Because the galactose-restricted diet is so critical for your or your child's well-being, it is important to stay up-to-date on the latest dietary guidelines for galactosemia.

As researchers learn more about approaches to dietary restriction of galactose, future guidelines about the galactose-restricted diet might look different. Your dietician is the best resource for the latest information. Because researchers have learned more about the diet over the past few decades, you might see conflicting information online about safe foods. The Galactosemia Foundation website (www.galactosemia.org) has the most up-to-date dietary guidelines for the United States.

GUIDELINE RECOMMENDATION

Individuals with <10% GALT activity should follow a life-long galactose-restricted diet that eliminates concentrated sources of lactose and galactose including most dairy products.[1]

TIPS FROM THE EXPERTS

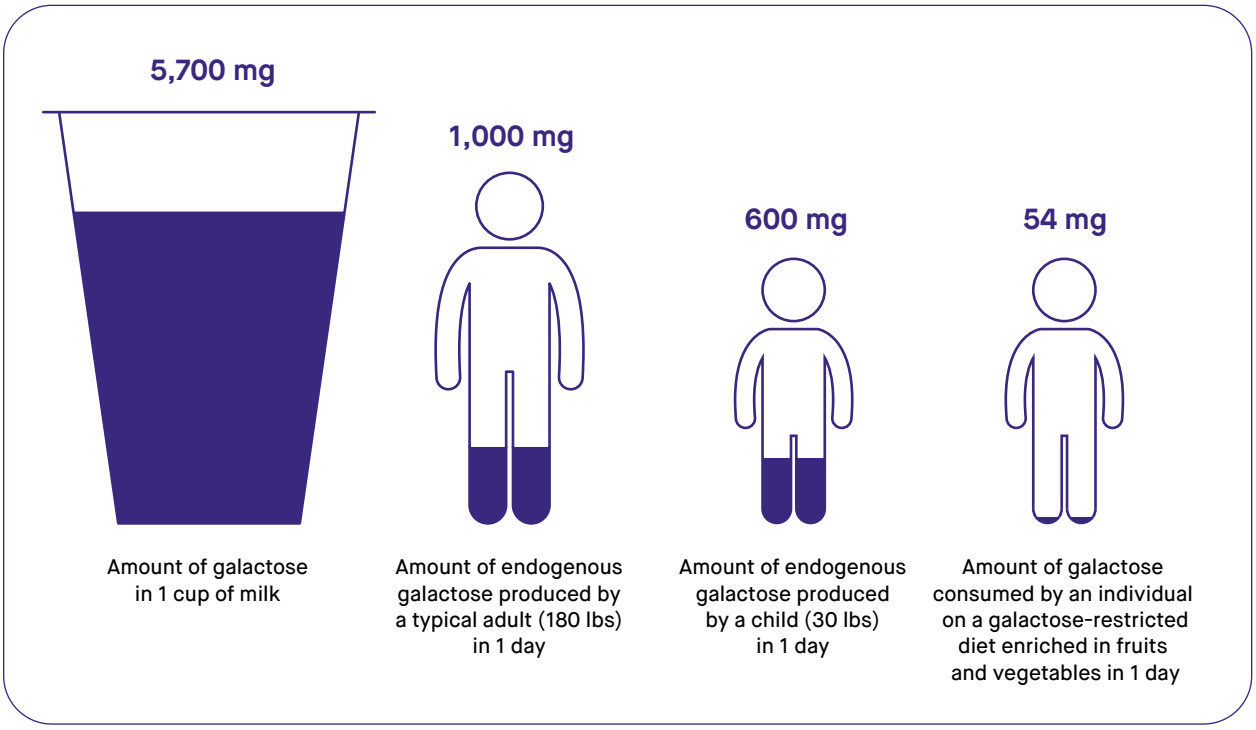
MILK SUBSTITUTES (E.G., OAT, COCONUT, AND ALMOND MILK) ARE NOW OFTEN FORTIFIED WITH CALCIUM AND VITAMIN D, BUT YOU WILL STILL WANT TO ENSURE THAT YOUR OR YOUR CHILD'S DOCTOR IS REGULARLY ASSESSING DIETARY INTAKE OF CALCIUM AND VITAMIN D.

Diet Is Not Enough

The galactose-restricted diet is typically initiated immediately after newborn screening results indicate that a baby has galactosemia. One cup of milk may contain 5,700 mg of galactose. A galactose-restricted diet may reduce the daily consumption of galactose by as much as 99%.[12,13]

Our bodies produce galactose naturally (endogenous galactose).[1] The amount of endogenous galactose production does decrease with age (for example, newborns produce more than 24.8 mg/kg per day while adults produce 8.4 mg/kg per day).[1] The typical adult still produces approximately 1,000 mg of endogenous galactose per day.[12] **Figure 3** provides a visual reference for this. While this amount is much lower than the amount contained within a single glass of milk, it is still much higher than the dietary galactose consumed by a person on a galactose-restricted diet. It is hypothesized that because the body endogenously produces galactose, lifelong symptoms can still occur even when the galactose-restricted diet is followed. It is important for all people with galactosemia to be monitored for associated complications so that if a problem presents, it can be addressed early, when an intervention may provide greater opportunity for benefit.

Figure 3: Relative amounts of galactose from various sources[12-14,a]



[a]Fruits and vegetables contain bound (very little digestible) galactose and are safe to eat.

Fruits, Vegetables, and Legumes

Galactose found in plants can exist in the free form or the bound form.[15] Free galactose is easily absorbed in the digestive tract and adds to the galactose levels in the body.[15] Bound galactose is tightly connected to food components that are not digested and does not increase galactose.[15] Most of the galactose in fruits, vegetables, and legumes is bound and the amount of free galactose is very small and far less than other foods, as shown in **Figure 4**. Therefore, all fruits, vegetables, and legumes can be consumed by people with galactosemia.[15] For many years, it was thought that some of these foods were not safe, but recent data shows that endogenous production of galactose is greater than the intake of galactose from fruits, vegetables, and legumes, and the benefits from these foods are important for good health.[13]

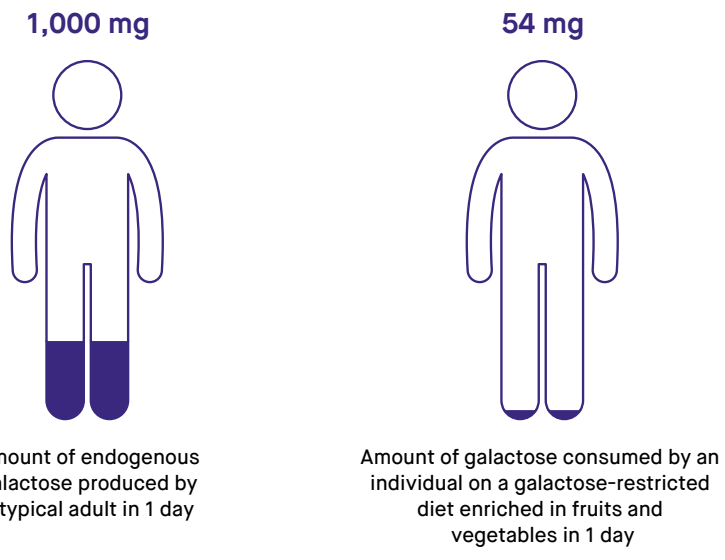
TIPS FROM THE EXPERTS

*ENZYME SUPPLEMENTS CONTAINING AN ENZYME α-GALACTOSIDASE SHOULD BE AVOIDED. α-GALACTOSIDASE BREAKS DOWN BOUND GALACTOSE IN LEGUMES AND INCREASES FREE GALACTOSE.[16,17] FOR THIS REASON, PEOPLE WITH GALACTOSEMIA SHOULD AVOID ANY SUPPLEMENT OR DIGESTIVE ENZYMES WITH α-GALACTOSIDASE SUCH AS BEANO®.[18] THIS IS ONE EXAMPLE OF SOMETHING UNSAFE; PLEASE SEE THE **PHARMACY: LACTOSE IN MEDICATIONS** SECTION OF THIS CHAPTER FOR OTHERS.*

GUIDELINE RECOMMENDATION

Individuals may eat any amount and types of fruits, vegetables, and legumes.[1]

Figure 4: Amount of endogenous galactose produced per day versus daily amount from fruits and vegetables per day[1,a]



[a]Fruits and vegetables contain bound (very little digestible) galactose and are safe to eat.

Mature Cheeses

In 2014, researchers reported aged cheeses produced in the United States that contain less than 25 mg galactose/100 g are safe for people with galactosemia to eat.[13] These include[13]:

- Swiss, Emmentaler, Gruyere
- Sharp Cheddar (aged over 1 year)
- Parmesan (aged over 10 months)
- Jarlsberg
- Tilsiter[19]

The cheese-making process causes milk to separate into curds (solids) and whey (liquid).[15] The whey contains most of the lactose and is drained away from the curds prior to pressing into cheese. The bacteria used to age cheese further breaks down lactose and galactose over time.[15] The type of bacteria, processing temperature, and length of aging all impact the safety of cheeses in the galactose-restricted diet.[20]

GUIDELINE RECOMMENDATION

Individuals may eat any amount and type of mature cheeses (with galactose content <25 mg/100 g).[1]

TIPS FROM THE EXPERTS

BEWARE OF ANY PROCESSED CHEESE PRODUCTS THAT ARE CHEDDAR- OR SWISS-"FLAVORED." ALSO AVOID SOFT OR SEMI-SOFT CHEESES, SUCH AS CREAM CHEESE OR CHEESE-BASED SPREADS. THESE TYPES OF CHEESES ARE NOT AGED LONG ENOUGH TO BE GALACTOSE-FREE.[1]

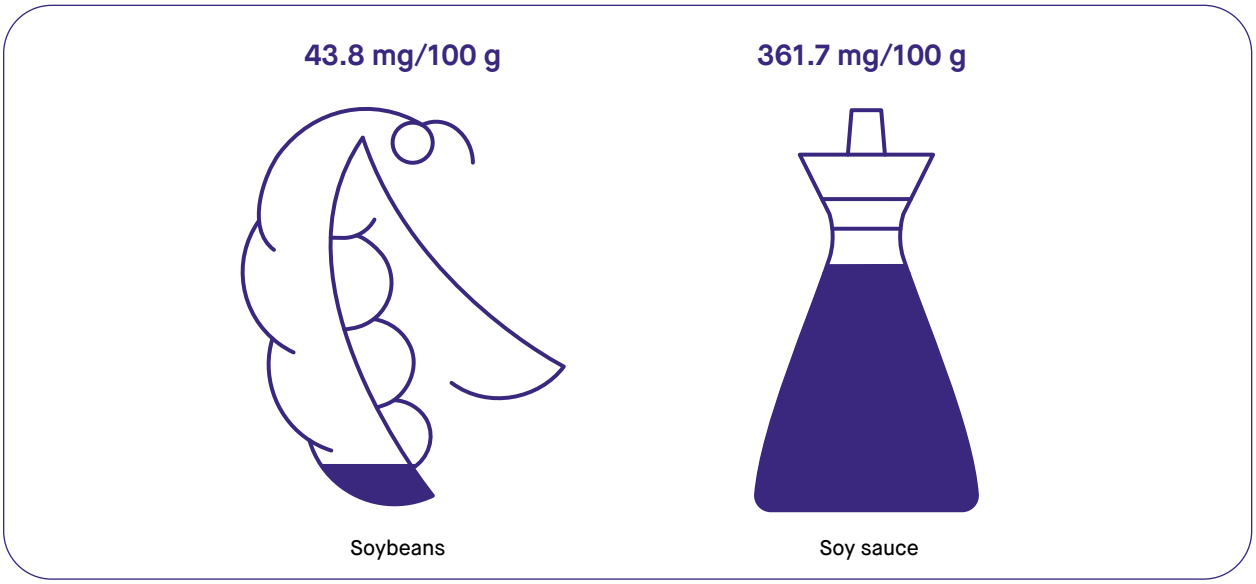
Soy-Based Products

Unfermented soy-based products are considered safe for people with galactosemia to eat.[15] The process of fermentation releases bound galactose.[21] Therefore, fermented soy-based products have higher free galactose content and products such as soy sauce, miso, and tempeh should be consumed only in moderation (consumed infrequently and only in small amounts).[22]

PARENT TIP

ONE FAMILY RECOMMENDED COCONUT AMINOS AS A SUBSTITUTE FOR SOY SAUCE, BECAUSE IT HAS A SIMILAR FLAVOR AND DOES NOT CONTAIN GALACTOSE. ONE FAMILY ALSO RECOMMENDED NON-DAIRY MARGARINES, AVOCADO OIL, AND OLIVE OIL AS SUBSTITUTES FOR BUTTER, AS THEY DO NOT CONTAIN GALACTOSE.

Figure 5: Relative amounts of free galactose in soy-based products[13]



GUIDELINE RECOMMENDATION

Individuals may eat any amount and type of unfermented soy-based products but should consume fermented soy-based products only in moderation.[1]

Organ Meats and Meat By-Products

Organ meats (e.g., liver, tripe, brain, and heart) and meat by-products found in some processed meats contain free galactose. Therefore, these foods should be eaten only in moderation.[22] Even though not all meat by-products contain organ meat, you can easily look for "meat by-products" on the packaging.

PARENT TIP

WE LOOK FOR KOSHER PAREVE (OR PARVE) FOODS BECAUSE THEY ARE DAIRY-FREE. WE ALSO LOOK FOR VEGAN OPTIONS, BECAUSE THEY DO NOT CONTAIN ANIMAL PRODUCTS. BUT, WE ALWAYS CHECK LABELS, EVEN FOR KOSHER PAREVE OR VEGAN PRODUCTS.

Table 2: Current diet restriction for galactosemia[1,22,23,a]

Safe foods and ingredients	Foods to be used in moderation	Unsafe foods and ingredients
Soy-based infant formulas containing soy protein isolate, amino acid-based elemental infant formulas All fruits, vegetables, and their juices, pickled fruit and vegetables All legumes (e.g., navy beans, kidney beans, garbanzo beans/ chick peas, soybeans) Soy-based products that are not fermented (soy milk, tofu, textured soy protein, hydrolyzed vegetable protein, soy protein concentrate, meat substitutes) Aged cheeses[b]: Jarlsberg, Emmentaler, Swiss, Gruyere, Tilsiter, mature Parmesan, mature Cheddar cheese Sodium and calcium caseinate All cacao products (like dark chocolate) but NOT milk chocolate Eggs Additional ingredients: natural and artificial flavorings, all gums, including carrageenan	Soy sauce, soy products that are fermented (e.g., miso, natto, tempeh, sufu) Meat by-products Organ meats Ghee	Breast milk, all animal milk–based infant formulas Processed meats containing lactose All milk-based foods and beverages, including low- or no-lactose milk, except for caseinates and aged cheeses, listed in the "safe" column All milk-based ingredients including buttermilk solids, casein, dry milk protein, dry milk solids, hydrolyzed whey protein, hydrolyzed casein protein, lactose, lactalbumin, whey All cheese and cheese-based products except those listed in the "safe" column Butter

[a]All manufactured foods need to be checked for the presence of milk by reading food ingredient labels.
[b]Galactose content and consequently allowed types of cheese may vary in different countries.
Note: some recommendations may vary based on source.

Reading Food Labels

The United States Food and Drug Administration (FDA) has very strict requirements for labeling the eight major food allergens, including milk, on commercial food products sold in the U.S.[24]

> "Read the ingredients on the label and look for the restricted foods or ingredients. Even if a label says, "may contain milk," if you are not seeing a restricted ingredient on the label, then it is ok to use."
>
> *– Sandy van Calcar, PhD, RD, LD*

The law requires the name of the food allergen must either[24]:

- Appear in parentheses following the name of the ingredient
  Examples: "lecithin (soy)", "whey (milk)", or
- Immediately after or next to the list of ingredients in a "contains" statement
  Example: "Contains wheat, milk, and soy"

Because galactosemia is not an allergy to milk, products with statements indicating a potential cross-contamination with milk products are considered safe (e.g., "This product was made in a facility that has milk products" or "May contain milk"). This is because any possible contamination with milk will be only a trace amount.

Some labels may contain ingredients that contain dairy (and by extension, lactose and/or galactose) but may not contain the word "milk," like sour cream or cream cheese (see **Figures 6 and 7**).

Figure 6: Common ingredients in labels[15]

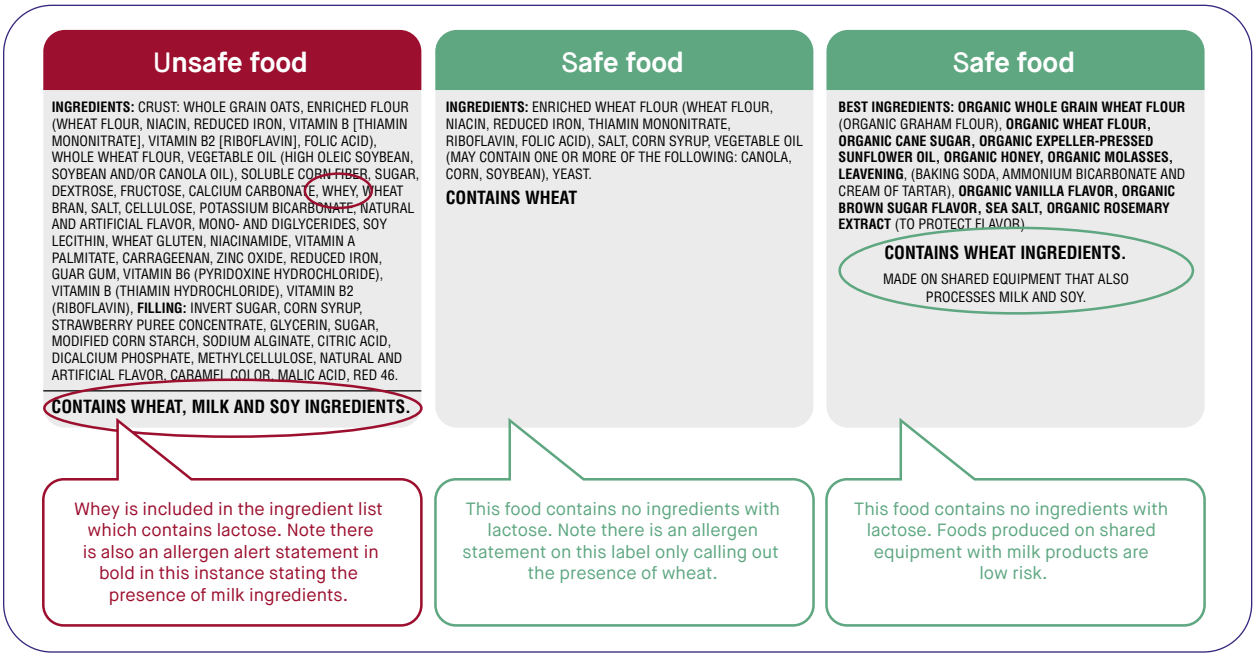
Common unsafe lactose-containing ingredients	Common safe ingredients that do not contain lactose
Casein[a]	Calcium carbonate
Curds	Calcium caseinate[a]
Lactalbumin	Cocoa butter
Lactose	Coconut cream
Whey	Lecithin oleoresin
	Sodium caseinate

[a]Approximately 80% of the protein in cow's milk originates from casein. Caseins are solubilized into caseinates. During this process, much of the whey is removed and washed leaving very low levels of lactose, and consequently galactose, in the caseinate (average of only 35.5 mg/100 g). Therefore, ingredients listed as casein are unsafe, but caseinate ingredients are considered safe.

GUIDELINE RECOMMENDATION

Individuals may consume the food additives sodium caseinate or calcium caseinate.[1]

Figure 7: Food label examples



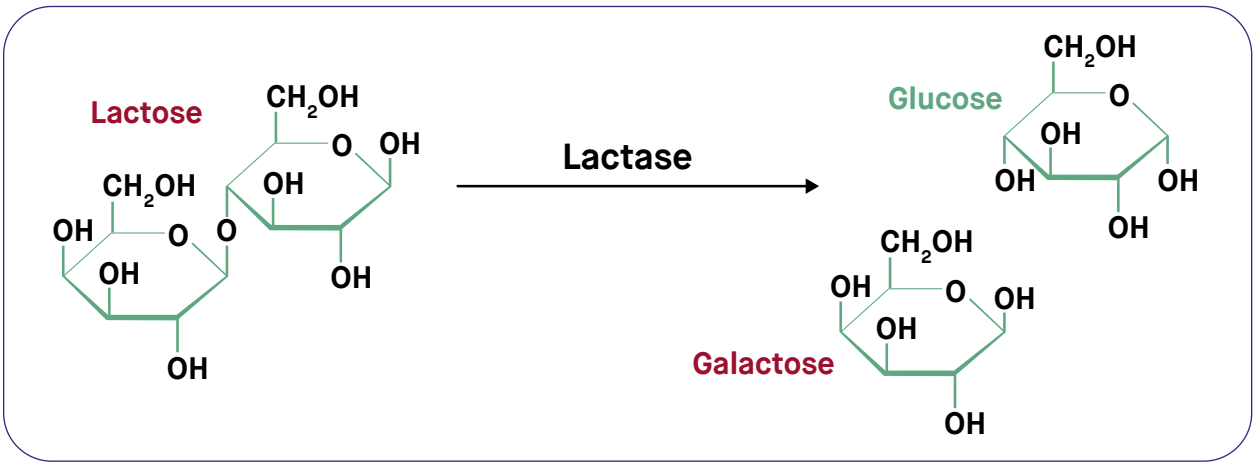
PARENT TIP

ALWAYS RE-READ LABELS WHEN SHOPPING AT THE GROCERY STORE, EVEN FOR ITEMS YOU HAVE PURCHASED BEFORE. INGREDIENTS CAN CHANGE FREQUENTLY.

Lactose-Free Is Not Galactose-Free

The main source of dietary galactose is lactose. After consumption, lactose is broken down by the lactase enzyme into glucose and galactose (as shown in **Figure 8**).[17]

Figure 8: Lactose[a] breakdown into glucose and galactose by lactase[17]



[a]Foods labeled "lactose-free" often still contain galactose and are unsafe for people with galactosemia. Always check the labels.

In many lactose-free or low-lactose products created for lactose-intolerant people, **lactase is added** and results in the breakdown of lactose into glucose and galactose.[1] This makes these products easier to digest for people who are lactose-intolerant, but it does not remove the galactose. For this reason, lactose-free or low-lactose products may not be safe for people with galactosemia.[1]

Pharmacy: Lactose in Medications

Because lactose is a sugar and can add sweetness, many drug manufacturers add it to their medications to counteract bitterness.[23] The National Institutes of Health (NIH) DailyMed website (https://dailymed.nlm.nih.gov) allows you to search for your or your child's medication and provides a full ingredient list. The actual level of lactose in medications is usually very low. Ask your or your child's doctor or pharmacist if the medication is safe, or if there are alternative dairy-free brands or formulations.

Advocating for Yourself or Your Child with Family, Daycare, School, and Restaurants

One of the most important ways to ensure your own or your child's well-being is to educate family and care providers about galactosemia. While restriction of certain foods has similarities to management of food allergies, it is important to distinguish the difference. It can be helpful to call a restaurant ahead of time before eating out. Because the restaurant representative may not realize that some ingredients fall into the restricted category (e.g., butter is a dairy product), you may want to ask for an ingredient list and review that list with the manager.

> "I usually call the restaurant when they aren't busy; so the waitstaff isn't put on the spot and my sons aren't embarrassed. I try to talk to people when they aren't in a hurry."
>
> *– Parent of two teens with galactosemia*

Diet and Growth/Height

Some studies suggest that growth (both height and weight) is delayed in children and adolescents with galactosemia.[25,26] However, by the time they reach their final adult height, they have caught up to peers. While the reason for this is not known, researchers hypothesize people with galactosemia might experience these delays due to either disease-related factors or diet-related factors.[25]

Advice on Managing a Galactose-Restricted Diet from Families in the Galactosemia Community

- ✔ Educate friends, family, teachers, and school staff about galactosemia.
- ✔ Communicate about what your child can eat safely.
- ✔ Contact the restaurant ahead of time.
- ✔ Check online allergy menus for chain restaurants.
- ✔ Keep a list of unsafe ingredients handy to share with others.
- ✔ Share your child's story with local restaurants you frequently visit to build relationships.
- ✔ Try new restaurants ahead of time without your child to fully understand what they have to offer.
- ✔ Keep a snack box with safe foods for your child in the classroom to proactively prepare for impromptu celebrations and events involving food.
- ✔ Ask the teacher to post a picture of your child with their dietary restrictions so any substitute or classroom helpers will be aware.
- ✔ Request a 1:1 meeting with your child's teacher before the start of the school year to explain restrictions and answer any questions.
- ✔ Provide daytime contact information for yourself where the teacher can contact you with questions about food labels and ingredients in real time.
- ✔ Check school menus posted ahead of time for allergens (many counties provide alternative food options for people with allergies).
- ✔ Apply green (for safe) and red (for unsafe) stickers on food in the refrigerator and pantry to educate your child and give them some independence as they mature.

Newborn

Newborn

Introduction

> "I know it's a scary time, but my advice is to remember that your baby is perfect and beautiful! So, enjoy your time with them when they're little."
>
> *– Parent of a 7-year-old with galactosemia*

The arrival of a new baby is an exciting time filled with hopes and expectations. It can be a very emotional, overwhelming time. When your child has been diagnosed with galactosemia, an additional flood of emotions may pour in, including worry and fear for your child's health. It can be difficult to accept that the future course for your child may be different than what you had imagined. Remember you are not alone. Through the Galactosemia Foundation, there is a community of support and resources that can help you navigate the road ahead with your child. The Galactosemia Foundation maintains an official Facebook account for families navigating life with galactosemia. The Galactosemia Foundation also sponsors a conference every 2 years which focuses on community connection, education on the latest research, and provides families with strategies for managing symptoms of galactosemia throughout the different stages of life. Connecting with other families who share similar experiences is a great way to feel less isolated.

Screening and Diagnosis

Galactosemia is included in newborn screening programs in all 50 states of the U.S. and in many other countries.[5] Newborn screening is usually done within 48 hours of birth; however, the results may not be available for several days or more. Because most parents are unaware that they are carriers, a child may be fed breast milk or infant formula containing lactose and galactose before the positive screening result is received.

The method used in newborn screening for galactosemia varies by state and either detects decreased functional GALT enzyme activity or elevated galactose metabolites in a blood sample.[5] The risk and severity of galactosemia depends on many factors, including the types of mutations in the *GALT* gene and the level of GALT enzyme activity detected in a fresh blood sample.[5] There are guideline recommendations assembled by experts regarding which people might have the most severe symptoms, but all people with <10% GALT activity should adhere to a galactose-restricted diet.[1]

GUIDELINE RECOMMENDATION

Make an appointment to see a metabolic geneticist to help confirm your child's galactosemia diagnosis. They will measure the amount of GALT enzyme activity that is present in your child's red blood cells and confirm it is missing or significantly decreased. They may also determine the *GALT* gene mutations your child has. If your child has *GALT* gene mutations known to cause disease, this may be enough information to confirm your child's diagnosis.[1]

Diagnosis requires quantitative testing of GALT enzyme activity and/or genetic testing.[1] GALT enzyme activity in people with galactosemia may sometimes be expressed as a relative percentage of GALT enzyme activity observed in individuals with functioning *GALT* genes. These people are sometimes called controls.[1] Absent or barely detectable GALT enzyme activity is usually diagnostic for classic galactosemia (CG).[1] Blood concentrations of the metabolite galactose-1-phosphate (Gal-1P) may also be elevated in newborns, especially following exposure to milk.[1] Your child's doctor may also conduct additional tests to measure levels of galactose in the blood and the level of the metabolite galactitol in the urine.

GALNET REGISTRY DATA

Most Common Abnormalities Reported in Newborns with Galactosemia[2]

Note: these symptoms typically resolve after switching to a non-milk-based formula.

- Elevated liver enzymes (70%)
- Bruising easily (43%)
- Encephalopathy (damage or disease that affects the brain) (29%)
- Signs of infection (27%)
- Cataracts (26%)
- Hypoglycemia (25%)

Galactose-Restricted Diet in Newborns

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information.

Newborns with galactosemia who consume breast milk or milk-based formula may experience life-threatening symptoms.[1] The symptoms typically resolve once a galactose-restricted diet is started.[1] Following diagnosis, you will need to stop feeding your baby with breast milk or whey-based formulas. In most cases, your baby can safely consume soy-based or elemental formulas.[1] Elemental formulas contain proteins that are broken down into their simplest form.[1] These formulas are often used for infants with soy and milk protein allergies, and are lactose-free.[1] While most babies can be successfully switched to soy-based formulas, each family is different. If your baby has a soy allergy, an alternative formula will be necessary. See **Table 3** for some examples.

Table 3: Examples of formulas indicated for infants with galactosemia[27,28]

Class	Brand names
Soy	Enfamil™ Prosobee®, Gerber® Good Start® Soy, Similac® Soy Isomil®, and others
Elemental	Elecare®, Neocate, Nutramigen® AA, PurAmino™, Alfamino®, and others

Note: Generic formulas of these brands may also be safe, provided ingredient list is comparable.

Enfamil Prosobee, Nutramigen, and PurAmino are trademarks or registered trademarks of Mead Johnson & Company, LLC. Gerber Good Start Soy and Alphamino are registered trademarks of Societe des Produits Nestle S.A. Elecare and Similac Isomil are registered trademarks of Abbott Laboratories.

Babies with galactosemia can have feeding difficulties such as poor sucking reflex.[6,29] If you notice these issues in your baby, ask for an evaluation with a specialist (this may include a feeding therapist, speech-language pathologist, or occupational therapist). These difficulties can often be supported with feeding therapy either at the hospital or once you return home. Feeding and swallowing centers can provide valuable support. In the U.S., early intervention services for infants and toddlers can help with these and other developmental issues.

PARENT TIP

IN SOME STATES, SOY AND ELEMENTAL FORMULAS ARE COVERED BY HEALTH INSURANCE. CHECK WITH YOUR INSURANCE COMPANY AND ADVOCATE FOR COVERAGE. DO NOT BE SURPRISED IF YOU ARE DENIED INITIALLY. YOU MAY HAVE TO APPEAL THE DECISION TO GET COVERAGE. YOUR PEDIATRICIAN CAN WRITE A PRESCRIPTION CLEARLY STATING GALACTOSEMIA DIAGNOSIS FOR THE INSURANCE COMPANY'S REFERENCE AND MAY ALSO HAVE SAMPLES OF SOME FORMULAS.

Cataracts

Cataracts are a condition in which the lens of the eye becomes cloudy and may result in vision problems, such as blurriness and defective night vision.[30] In many cases cataracts that develop in infants can resolve or markedly improve with adherence to the galactose-restricted diet.[1] Cataracts that do not resolve may be surgically removed and the lens replaced with an artificial version.[30] Because cataracts commonly occur in newborns with classic galactosemia, your baby's eyes can be evaluated by an ophthalmologist upon diagnosis. Experts tend to agree that if no issues are detected in the eyes shortly after birth and adherence to a galactose-restricted diet is good, eye exams can be conducted on an as-needed basis.

GALNET REGISTRY DATA

26% of neonatal participants experienced cataracts.[2,a]

[a]Participants may or may not have been adherent to a galactose-restricted diet at time of cataract diagnosis.

GUIDELINE RECOMMENDATION

Upon diagnosis, your child may be referred to an ophthalmologist to assess for cataracts.[1]

Newborn: Monitoring and Referrals[1]

Ophthalmology referral at diagnosis to assess for cataracts

Monitor for infection

Monitor liver function

Monitor feeding and weight

First Year of Life

First Year of Life

Introduction

Your baby's first year will be filled with excitement as they start interacting with the world. You may feel overwhelmed as you learn to navigate caring for your child. Connecting with other parents may help guide you through this time. You may even seek out a mentor in a parent with an older child who can provide insights and practical advice.

"The first few months can be isolating and overwhelming. It's important to make connections, reach out for support, and remember to soak in the precious moments with your baby as you adjust to a new normal."

– Parent of a 7-year-old with galactosemia

"Just like in the airplane, it is important to put on your own oxygen mask first before tending to your child. For parents of a newly diagnosed baby with galactosemia, this means taking care of yourself. Use your supports. And, moms, if you experience postpartum depression or anxiety, check in with a healthcare provider or counselor. And, dads, the same goes for you, even though others may focus on how the mother is feeling."

– Susan E. Waisbren, PhD

Year One: Considerations for the Galactose-Restricted Diet

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information.

Your baby is growing and is likely already eating some solid foods. You can advocate for your baby by educating family, friends, daycare, and other care providers about their diagnosis and dietary restrictions. Your child's doctor may want to periodically conduct a dietary assessment or refer you to a dietician to ensure your baby is getting adequate amounts of important vitamins and nutrients. If levels are too low, your child's doctor may recommend additional supplements.

GALNET REGISTRY DATA

27% of participants experienced vitamin D deficiency.[2]

GUIDELINE RECOMMENDATIONS

Your child should have a dietary assessment done once a year to determine calcium and vitamin D intake.[1]

Calcium and vitamin D supplements should be added as needed to help ensure healthy bones.[1]

Laboratory Follow-Up

Your child's doctor will likely monitor the levels of harmful metabolites such as Gal-1P in the blood. It has been shown there is high variability in metabolite levels among individuals, therefore each person serves as their own reference when tracking changes over time.[1]

PARENT TIP

IT CAN BE SCARY WHEN YOUR BABY MUST HAVE BLOOD DRAWN, BUT THERE ARE THINGS YOU CAN DO TO ADVOCATE FOR YOUR CHILD AND MINIMIZE THEIR DISCOMFORT. REQUEST THAT AN EXPERIENCED PEDIATRIC PHLEBOTOMIST COLLECT YOUR BABY'S SAMPLE. ACCORDING TO THE HARVARD HEALTH PUBLISHING BLOG, IT MAY ALSO BE HELPFUL TO ASK TO HOLD YOUR BABY DURING THE PROCEDURE AND USE A PACIFIER AS SUCKING SOOTHES BABIES.[31] SOME RESEARCH SUGGESTS THAT SUGAR WATER MAY ACTIVATE NATURAL PAIN RELIEF SYSTEMS IN THE BODY.[32] SOME PROVIDERS MAY USE THIS WHEN DRAWING BLOOD FROM INFANTS. ASK IF THIS IS AN OPTION FOR YOUR CHILD.

"We always made sure our daughter was hungry before blood draws. She was so interested in her bottle that the blood draws were a breeze!"

– Parent of a 5-year-old with galactosemia

GUIDELINE RECOMMENDATION

Because Gal-1P levels take a while to decrease after initiation of the galactose-restricted diet, your child should have the level of Gal-1P in their red blood cells measured 3 and 9 months after starting galactose-free formula.[1]

Neurological

It is important to talk with your child's doctor about any concerns you have about your child's health. Some neurological complications that may occur are impaired motor function, impaired coordination (ataxia), involuntary muscle contractions or movements (dystonia), and involuntary rhythmic shaking in one or more body parts (tremor).[2] Your child's doctor may also ask you about seizures. Seizures are not a common occurrence. However, it is still important to be aware so you can discuss them as soon as possible with your child's doctor.[2]

GALNET REGISTRY DATA

52% of participants across age groups experienced neurological complications.[2]

8% of participants across age groups experienced seizures.[2,a]

[a]Information regarding type, severity, and frequency was not provided in this analysis.

GUIDELINE RECOMMENDATION

During your clinic visits, your child's doctor may ask you about any seizures or seizure-like activity since the last visit. If seizures do occur, your doctor may want to assess the electrical activity of the brain. This is usually assessed with an electroencephalogram (EEG) using electrodes taped to your baby's scalp. These electrodes are not painful to your child.[1]

PARENT TIP

IT CAN BE SCARY TO WITNESS A SEIZURE IN YOUR CHILD. IT CAN BE HELPFUL TO REVIEW SEIZURE FIRST AID INFORMATION, SO YOU FEEL PREPARED IF NEEDED. THE EPILEPSY FOUNDATION IS A GREAT RESOURCE FOR SEIZURE FIRST AID. PLEASE VISIT THEIR WEBSITE FOR MORE INFORMATION: ***WWW.EPILEPSY.COM/LIVING-EPILEPSY/EPILEPSY-AND/PARENTS-AND-CAREGIVERS/ABOUT-KIDS/SEIZURE-FIRST-AID.***[33]

Speech and Language

Speech and language delays, disorders, and/or impairments are the most common complication in children with galactosemia.[2] The severity of speech disorder varies widely, and can make it difficult to communicate with people, work, and socialize with peers. There are specific speech disorders that occur more frequently in people with galactosemia.[31] Motor speech disorders are caused by impairments in the ability to plan, program, or control the neuromuscular movements needed for speech.[31] In a study with 33 participants,

motor speech disorders including apraxia, dysarthria, or both occurred in 27% of people with galactosemia.[34] In addition to motor speech disorders, some children and teens also had breathy, harsh voice quality with inadequate breath support for speaking long phrases and sentences, indicating vocal cord dysfunction.[34] People with galactosemia may have expressive language complications only or mixed receptive-expressive language complications, as described in **Table 4**.[34]

TIPS FROM THE EXPERTS

*ASK YOUR CHILD'S DOCTOR ABOUT SPEECH CLINICAL TRIALS FOR CHILDREN WITH GALACTOSEMIA SUCH AS **BABBLE BOOT CAMP**. THESE TRIALS CAN BE A RESOURCE FOR YOUR CHILD'S SPEECH DEVELOPMENT.*

Table 4: Common speech and voice complications observed in people with galactosemia.[34,35]

Complications	Characteristics
Apraxia	• Inconsistent consonant and vowel errors • Slow or halting transitions from one sound to the next sound • Difficulty with smooth speech and placing emphasis on the correct syllables (e.g., ***con*tent** [happy] and ***con*tent** [subject matter]) • Not due to muscular weakness or an anatomical difference
Dysarthria	• Consistent consonant and vowel errors • Difficulty with smooth speech, emphasis, or slow rate • Due to muscular weakness, decreased range of motion, or an anatomical difference
Receptive language	• Inability to take in and understand information
Expressive language	• Inability to express language (put thoughts into understandable words and sentences)
Vocal cord (vocal fold) dysfunction	• Harsh, breathy, strained, or unstable voice • Decreased ability to speak long phrases and sentences

GALNET REGISTRY DATA

Language delays were the most common of the developmental delays amongst study participants.[2]

66% of participants across all ages experienced speech and language disorders.[2]

It is important to ensure your child is meeting appropriate milestones as they grow so you can get them the support they need as early as possible. In young children who have not yet learned how to speak, screening will assess hearing and how well your child communicates with gestures, body language, and facial expressions.[1]

TIPS FROM THE EXPERTS

EVEN IF YOUR CHILD IS MEETING MILESTONES, THEY MAY QUALIFY TO RECEIVE SERVICES TO SUPPORT DEVELOPMENT. CLASSIC OR CLINICAL VARIANT GALACTOSEMIA AUTOMATICALLY QUALIFIES CHILDREN FOR EARLY INTERVENTION IN SOME STATES. START SPEECH THERAPY AS SOON AS POSSIBLE. ONE EXPERT RECOMMENDS ASSESSMENT AS EARLY AS 2-3 MONTHS, AS SPEECH THERAPY MAY MINIMIZE OR PREVENT SERIOUS SPEECH AND LANGUAGE DISORDERS.

GUIDELINE RECOMMENDATION

Your child should be screened for speech and language delays at ages 7-12 months. Your child's doctor may refer you to a speech-language pathologist for additional testing if your child is not meeting appropriate milestones.[1]

Navigating Early Intervention

Developmental delays can occur in children with galactosemia.[1] Experts suggest parents proactively seek early intervention for their child with galactosemia because of the known complications associated with this medical diagnosis. Intervention is likely more effective at preventing an issue rather than resolving an issue once it has occurred. Recent data from the ongoing Babble Boot Camp study demonstrated that children exposed to proactive activities and routines designed to prevent speech and language disorders had higher babbling complexity and greater expressive vocabulary.[36]

GALNET REGISTRY DATA

52% of participants experienced global developmental delays in infancy/childhood.[2]

In the U.S., infants and toddlers with galactosemia may qualify for early intervention provided free of charge through either county or school programs, depending on state regulations. Contact the school your child will attend for kindergarten to connect with the appropriate agency and start the referral process. You can also find a list with contact information on the CDC website: www.cdc.gov/ncbddd/actearly/parents/state-text.html. Once your child has been evaluated and if they are found to be eligible for the program, you will work with a service coordinator to develop an Individualized Family Service Plan, or IFSP. This is a customized legal document covered by the Individuals with Disabilities Education Act (IDEA) that maps out the support and services your child will receive to help them meet age-appropriate developmental milestones.[37]

An informal Facebook poll provided to members of a galactosemia-specific Facebook group indicated that many parents are not aware of this service and on average, children began intervention at 28 months of age. The following are typical areas of focus for early intervention[37]:

- Physical/motor skills (e.g., crawling, walking, and reaching)
- Cognitive skills (e.g., talking, listening, and understanding others)
- Self-help or adaptive skills (e.g., eating)
- Social or emotional skills (e.g., playing, interacting with others)

PARENT TIP

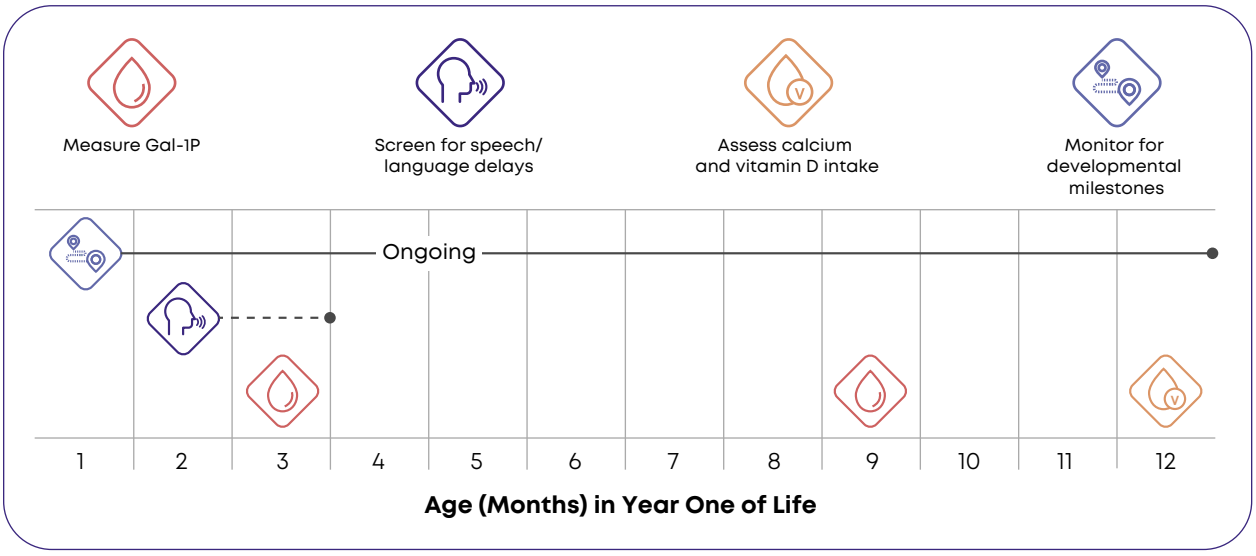
THE CDC OFFERS A DEVELOPMENTAL MILESTONE TRACKER APP THAT CAN BE DOWNLOADED TO YOUR DEVICE TO TRACK YOUR CHILD'S PROGRESS AS THEY GROW.[38] THIS CAN BE A USEFUL TOOL WHEN HAVING CONVERSATIONS WITH YOUR PEDIATRICIAN AND THE EARLY INTERVENTION SERVICE COORDINATOR. SEARCH CDC MILESTONE TRACKER IN YOUR APP STORE.

Early results from the ongoing Babble Boot Camp study suggest that with specific parent training, early intervention beginning with babies as young as 8 weeks of age may minimize or prevent serious speech and language disorders.[36,39]

PARENT TIP

HANG IN THERE. SOME OF THE SYMPTOMS OF GALACTOSEMIA CAN GET BETTER WITH THERAPY AND INTERVENTION. SPEECH AND LANGUAGE CAN BE IMPROVED UPON. THERE ARE REAL BENEFITS TO ALL OF THESE APPOINTMENTS. SPENDING QUALITY TIME WITH YOUR CHILDREN, READING, PLAYING, ENJOYING LIFE, AND TEACHING THEM ABOUT THE WORLD ARE ALSO IMPORTANT.

Year One: Monitoring and Assessments[1]



Early Years

(Ages 1-3)

Early Years (Ages 1-3)

Introduction

By the time your child reaches age 1 year, you may find yourself already juggling appointments with more than one specialist. If not, you may be wondering if you are missing something. Resources from the community such as this handbook can provide insights into what other people with galactosemia have experienced. This can help prepare you for what's ahead as you advocate for your child. Remember that no two children are the same, so your child's experiences as they grow may differ from those of others.

> "Having multiple doctors and therapists caring for your child can be overwhelming, but it can also bring a lot of peace knowing so many people have eyes on development and symptoms that might arise. We were able to find a physical therapist in the same building as an occupational therapist, and they worked as a team to care for our child. Small wins like that make all the difference when you are juggling multiple symptoms, therapies, and doctors' appointments."
>
> *– Parent of two children with galactosemia*

PARENT TIP

FOR RECURRENT APPOINTMENTS WITH SPECIALISTS, TRY AND SCHEDULE FOR A CONSISTENT DAY AND TIME. THIS CAN HELP YOU REMEMBER AND ALSO MAKE IT EASIER TO MANAGE YOUR SCHEDULE TO ACCOMMODATE THESE APPOINTMENTS.

Early Years: Considerations for the Galactose-Restricted Diet

As your child continues to explore new foods in the early years, continue reading food labels. It is important to educate care providers about your child's symptoms and food restrictions. You can review the ***Reading Food Labels*** section in ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter.

Your child's doctor may want to periodically conduct a dietary assessment to ensure your child is getting adequate amounts of calcium and vitamin D. If levels are too low, they may recommend additional supplements.

GUIDELINE RECOMMENDATIONS

Your child should have a dietary assessment done once a year to determine calcium and vitamin D intake.[1]

Calcium and vitamin D supplements should be added as needed.[1]

Laboratory Follow-Up

Now that your child has fully transitioned to solid food on a galactose-restricted diet, your child's doctor may want to monitor the Gal-1P metabolite levels in your child's red blood cells with a blood draw.[1] This is done to establish your child's baseline levels which can be used as a point of reference moving forward.[1] It has been shown there is high variability in metabolite levels among individuals, therefore each person serves as their own reference when tracking changes over time.[1] According to one expert, Gal-1P levels commonly decline from birth until age 6-12 months. Two or three measurements after age 1 year are generally considered baseline.

PARENT TIP

AS YOUR CHILD GETS OLDER, THEY MAY START TO EXPERIENCE ANXIETY WHEN GETTING THEIR BLOOD DRAWN. STUDIES HAVE DEMONSTRATED THAT VIBRATING DEVICES SUCH AS THE BUZZY® MAY HELP TO CONTROL PAIN IN CHILDREN.[40]

GUIDELINE RECOMMENDATION

Your child should have the level of Gal-1P in their red blood cells measured yearly after their first year **until** their baseline has been established.[1]

Neurological[1,2]

Neurological complications can occur in children with galactosemia. Some neurological complications that may occur are impaired motor function, impaired coordination (ataxia), involuntary muscle contractions or movements (dystonia), and involuntary rhythmic shaking in one or more body parts (tremor). Your child's doctor will screen for these complications to monitor any changes over time and ensure the earliest possible intervention. Your child's metabolic geneticist or neurologist will complete a full neurological exam and look at muscle strength and tone, coordination, walking, balance, and assess for tremors. They will also ask if your child has experienced any seizures. For more information about seizures, please see ***page 36*** in the ***First Year of Life*** chapter. Depending on your child's symptoms, your child's doctor may recommend physical or occupational therapy.

Buzzy is a registered trademark of MMJ Labs LLC.

GALNET REGISTRY DATA

52% of participants across all ages experienced neurological complications.[2]

24% of participants who experienced tremor were between age 1 and preschool age.[2]

GUIDELINE RECOMMENDATION

Your child may be screened about every 6 months for neurological complications from age 2-3 years. This usually involves a clinical exam to look for any disorders.[1]

Developmental Follow-Up

It is important to continue monitoring your child to ensure they are meeting appropriate developmental milestones. Pediatricians use pediatric screening to assess development in early childhood. Your child's doctor will use these tools in conjunction with your reported observations regarding your child's development to assess whether intervention is needed. Depending on the domains impacted, support services may include further evaluation with a physical or occupational therapist. These specialists can evaluate your child's specific strengths and determine whether therapy would be beneficial. Based on results of these scales, your doctor might refer you to see a physical therapist and/or an occupational therapist to support your child's continued growth in all areas.

GUIDELINE RECOMMENDATION

Your child's motor development (development of bones and muscles and how well your child uses them to move) should be assessed at age 2-3 years.[1] Examples of motor abnormalities include clumsiness and difficulty with coordination.

GALNET REGISTRY DATA

52% of participants experienced global developmental delays in infancy/childhood.[2]

Speech and Language

Continue monitoring your child's speech and language development during this stage. Studies have shown early intervention, such as speech therapy, can be effective at helping your child catch up.[41-43] Screening and follow-up assessment, if needed, may include hearing, receptive, expressive, and pragmatic language, a structure-function and motor speech examination looking at respiration, resonance, voice, and speech intelligibility for children not meeting age-expected milestones (see **Table 5** for more detail).[1] Apraxia Kids (www.apraxia-kids.org) is a reliable, evidence-based resource for speech and language information.

Table 5: Common speech and language complications assessed during screening[34,35]

Complications	Characteristics
Receptive language	Inability to take in and understand information
Expressive language	Inability to put thoughts into understandable words and sentences
Pragmatic language	Inability of an individual to use language appropriately in social situations (for example, waiting for others to finish speaking before beginning to speak)
Resonance	Amount of air and vibrations in the nose and mouth during speech
Hypernasality	Excessive air and vibrations passing through the nose (sounds like the quintessential telephone operator)
Hyponasality	Too little air and vibrations passing through the nose (sounds like a person with a bad cold)
Articulation	How correctly each sound is produced by the tongue, lips, jaws, and vocal cords

GALNET REGISTRY DATA

Language delays were the most common of the developmental delays amongst study participants.[2]

66% of participants across all ages experienced speech and language disorders.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for speech and language delays at ages 2 and 3 years.[1]

"By 24 months of age, we expect a child to have 50+ words and be able to put together 2+ word phrases (e.g., 'Mommy shoe') that unfamiliar listeners can understand."

– Nancy Potter, PhD, CCC-SLP

According to an expert in speech-language pathology, children with delays in speech and language often act out their frustration with not being able to effectively communicate. This acting out may include crying, screaming, or hitting. Introducing basic sign language, a picture-exchange system, or a low- or high-tech augmentative and alternative communication device often facilitates more appropriate behavior. Ask your speech-language pathologist for help with alternative communication strategies.

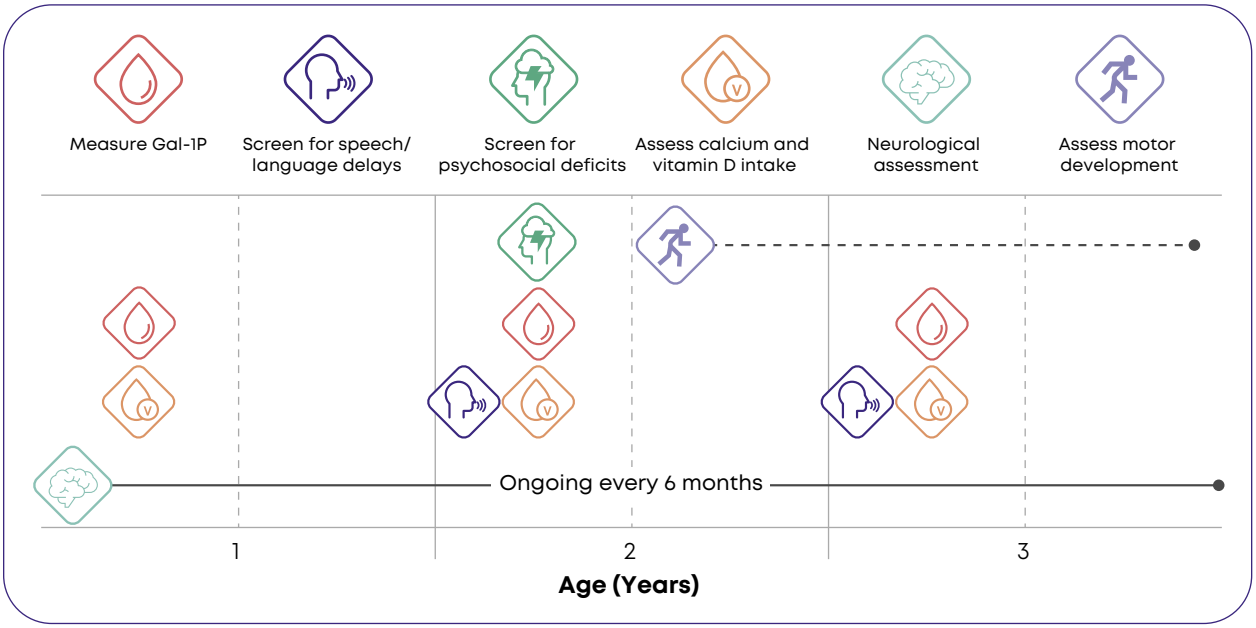
Psychosocial and Mental Health[1]

Emotional and mental health and behavioral problems have been observed in people with galactosemia. Assessments for mental health, autism spectrum disorders, and/or sensory disorders can help to determine appropriate next steps.

GUIDELINE RECOMMENDATION

Your child should be screened for behaviors on the autism spectrum, sensory integration challenges, depression, and anxiety at age 2 years using standardized tools such as the Behavior Assessment System for Children (BASC-3).[1]

Early Years: Monitoring and Assessments[1]



Preschool

(Ages 3-5)

Preschool (Ages 3-5)

Introduction

Depending on your child's birthday and the rules of your school district, they may be starting kindergarten at age 5 or 6 years. Please refer to the ***Primary School (Ages 5-10)*** chapter if your child is entering kindergarten.

For many children, preschool is their first experience with more formal education. Some children during this period are not attending formal preschool. Regardless, as your child starts to learn more about the world, it is an exciting time. Remember, children develop at their own pace. Celebrate with them when they learn new things, make a new friend, or come home excited about an activity at school.

> "It is important to think about all the things your child has learned to do. It doesn't really matter if it takes them more time, as long as they keep learning something new."
>
> *– Susan E. Waisbren, PhD*

Preschool: Considerations for the Galactose-Restricted Diet

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information.

Your child's doctors may want to periodically conduct a dietary assessment to ensure your child is getting adequate amounts of calcium and vitamin D. If levels are too low, they may recommend additional supplements.

GUIDELINE RECOMMENDATIONS

Your child should have a dietary assessment done once a year to determine calcium and vitamin D intake.[1]

Calcium and vitamin D supplements should be added as needed.[1]

Laboratory Follow-Up[1]

Your child's doctor may want to continue monitoring the Gal-1P metabolite levels in your child's red blood cells to establish baseline levels. It has been shown there is high variability in metabolite levels among individuals; therefore each person serves as their own reference when tracking changes over time. One expert noted they may continue laboratory assessments yearly following establishment of baseline. More frequent assessments may be needed as new symptoms or complications occur (e.g., seizures or other neurological problems). In addition, monitoring Gal-1P levels may provide a measure of how your child's diet and adherence is being maintained over time. At this age, they may also monitor galactitol levels in your child's urine.

GUIDELINE RECOMMENDATION

Your child should have the level of Gal-1P in their red blood cells measured yearly after their first year **until** their baseline has been established.[1]

Neurological[1,2]

As with younger age groups, neurological complications can occur in preschool-age children with galactosemia. Some neurological complications that may occur are impaired motor function, impaired coordination (ataxia), involuntary muscle contractions or movements (dystonia), and involuntary rhythmic shaking in one or more body parts (tremor). Your child's doctor will screen for these complications to monitor any changes over time and ensure the earliest possible intervention. Your child's metabolic geneticist or neurologist will complete a full neurological exam and look at muscle strength and tone, coordination, walking, balance, and assess for tremors. They may also ask you if your child has experienced any seizures. For more information about seizures, please see ***page 36*** in the ***First Year of Life*** chapter. Depending on your child's symptoms, your child's doctor may recommend physical or occupational therapy.

GALNET REGISTRY DATA

52% of participants across all ages experienced neurological complications.[2]

27% of participants across all ages experienced general motor disorder (clumsiness, coordination difficulties), most frequently reported at preschool age.[2]

GUIDELINE RECOMMENDATION

Your child should be screened about every 6 months for neurological or motor involvement from age 2-3 years.[1] This usually involves a clinical exam to look for any disorders.

Developmental Follow-Up

Children with galactosemia are at risk for developmental delays, including motor and cognitive.[2] It is important to continue monitoring your child to ensure they are meeting appropriate developmental motor milestones. Pediatricians use pediatric screening to assess development in early childhood. Your child's doctor will use these tools in conjunction with your reported observations regarding your child's development to assess whether intervention is needed.

Your child's doctor can also refer your child for testing to assess intellectual quotient (IQ). These are measures of how well your child performs a set of tasks as compared to typically developing children their age. This testing uses standardized tools such as the Weschsler Preschool and Primary Scale of Intelligence (WPPSI).[1] Knowing how your child performs on these measurements can help identify strengths and any needs for special educational support services. Educational strategies matched to a child's strengths are most effective. Strategies focused on visual learning, hands-on experiences, and un-timed evaluations generally help children with galactosemia reach their full potential in the classroom. Please refer to the ***Navigating Preschool Special Education*** section later in this chapter for more on how to navigate early special education services if needed.

GALNET REGISTRY DATA

11% of participants experienced motor delays in infancy/childhood.[2]

40% of participants experienced cognitive delays in infancy/childhood.[2]

GUIDELINE RECOMMENDATION

Your child's development and cognitive abilities should be assessed at age 4-5 years.[1]

Speech and Language

It is important to assess your child's speech and language to ensure school readiness and determine whether there is a need for continued speech therapy. If your child has already been receiving speech therapy, check in with their speech-language pathologist to discuss progress and assess whether any changes to their plan are needed. Screening and follow-up assessment, if needed, may include hearing, receptive, expressive, and pragmatic language, a structure-function and motor speech examination looking at respiration, resonance, voice, and speech intelligibility for children not meeting age-expected milestones.[1] **Table 6** provides additional detail.

Table 6: Common speech and language complications assessed during screening[34,35]

Complications	Characteristics
Receptive language	Inability to take in and understand information
Expressive language	Inability to put thoughts into understandable words and sentences
Pragmatic language	Inability of an individual to use language appropriately in social situations (for example, waiting for others to finish speaking before beginning to speak)
Resonance	Amount of air and vibrations in the nose and mouth during speech
Hypernasality	Excessive air and vibrations passing through the nose (sounds like the quintessential telephone operator)
Hyponasality	Too little air and vibrations passing through the nose (sounds like a person with a bad cold)
Articulation	How correctly each sound is produced by the tongue, lips, jaws, and vocal cords

Services are provided through most school systems if the student's disability/disabilities adversely affect educational performance and cannot be addressed through general education classes alone. If your child does not qualify or needs more help, you may consider seeking private therapy to address your child's needs. Your insurance may cover this additional support.

GALNET REGISTRY DATA

Language delays were the most common of the developmental delays amongst study participants.[2]

66% of participants across all ages experienced speech and language disorders.[2]

GUIDELINE RECOMMENDATION

Persistent speech disorders can contribute to reading and writing disabilities.[44]
Your child should be screened for speech and language delays at age 5 years.[1]

By age 4-5 years, children with speech difficulties increase their awareness that their speech differs from that of their peers and may show avoidance behaviors to avoid speaking, especially in public.[45] It can be easy to misinterpret avoidance behaviors as inattentive listening or failure to follow directions and miss that primarily the child is avoiding failure.

According to an expert in speech-language pathology, children who have delays in speech and language often act out their frustration with not being able to effectively communicate. This acting out may include crying, screaming, or hitting. Introducing basic sign language, a picture-exchange system, or a low- or high-tech augmentative and alternative communication device often facilitates more appropriate behavior. Ask your speech-language pathologist for help with alternative communication strategies.

Psychosocial and Mental Health

Mental health, emotional health, and behavioral challenges have been observed at increased prevalence in people with galactosemia.[2]

GALNET REGISTRY DATA

44% of participants across all ages experience mental health and behavioral problems.[2]
22% of participants across all ages experienced anxiety.[2]
8% of participants who experienced anxiety disorders were preschool age.[2]
6% of participants exhibited behaviors on the autism spectrum.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for autism spectrum disorders, sensory integration challenges, depression, and anxiety using standardized tools such as the Behavior Assessment System for Children (BASC-3) at age 4-5 years.[1]

Navigating Preschool Special Education

All states in the United States offer special education services for preschoolers who qualify through funding provided by Section 619 of the Individuals with Disabilities Education Act (IDEA).[37] Children can be assessed for eligibility at age 3 years once they transition from early intervention programs.

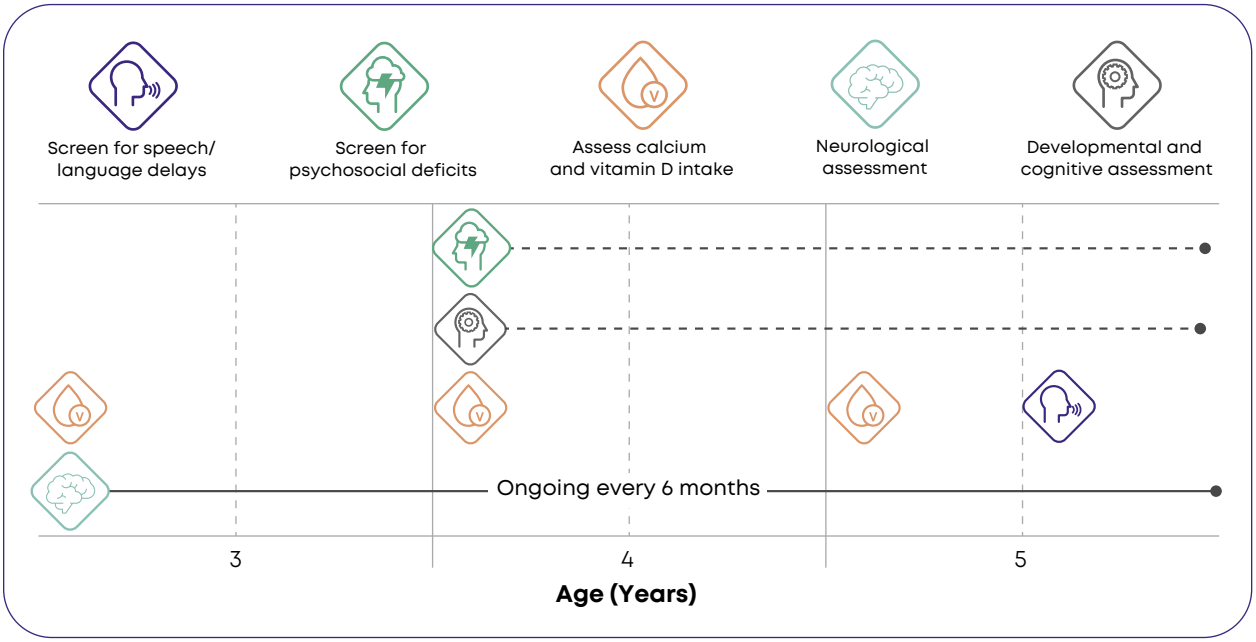
Not all children who are eligible for early intervention are automatically enrolled in preschool services, so it is important to check your state's requirements. The screening for these programs is free and is provided by your local school district. If your child needs services, reach out to your district's director of special education. If you experience difficulties, you can contact your state's 619 coordinator. A list of current coordinators by state can be found on the Early Childhood Technical Assistance Program website (https://ectacenter.org/contact/619coord.asp). Once your child's eligibility for services has been determined, the school district will work with you to develop an individualized education program (IEP). The IEP will outline the services that may help address your child's identified needs. Apraxia Kids is another good source of IEP information (www.apraxia-kids.org/iep-resource-guide/).

In some cases, you may find it difficult to qualify your child for services. Keep advocating and educating the school district (including superintendent, administration, and teachers) about galactosemia. It is important to try to work with your school district to meet your child's needs before bringing in additional personnel, but if your child is not getting the support needed, you may contact an advocate and consider getting an outside second opinion. In many states, school districts are required to pay for an outside evaluation for a second opinion if services are denied. If the previous steps are not effective, consider consulting an attorney who specializes in school law.

Advice on Navigating Special Education from Families in the Galactosemia Community

- ✔ The school and your IEP team are working with you and your child toward the same goal, student success.
- ✔ Educate the school system about galactosemia, including that the only current available standard of care is diet restriction and proactive early intervention.[36]
- ✔ Consider partnering with a student advocate to ensure services are provided.
- ✔ Know your rights. Check the education agency website for your state.
- ✔ Students with an IEP are not required to retake state-mandated testing, so advocate for your child.
- ✔ Advocate for summer programming to support your child.
- ✔ Ask for more frequent communication regarding evaluations for your child through the school system.
- ✔ The more specific you are and the more you vocalize, the more support your child will likely receive.

Preschool: Monitoring and Assessments[1]



Primary School

(Ages 5-10)

Primary School (Ages 5-10)

Introduction

Depending on your child's birthday and the rules of your school district, your 5-year-old may still be in preschool. Please refer to the ***Preschool (Ages 3-5)*** chapter if your 5-year-old has not yet entered kindergarten.

Entering primary school is an exciting step for children. It is also likely one of the first times they will be in a less stringently supervised environment. It can be a scary transition, but one of the best ways to cope is teaching your child to advocate on their own behalf, when possible. It can be helpful to describe ways to advocate when there is a need. Practicing such scenarios can give a child confidence to express what they need when they need it.

Joining us at the conference?
Look for the **Mighty G program** for youth (ages 9-11).
The activities are for all family members,
with or without galactosemia.

Primary School: Considerations for the Galactose-Restricted Diet

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information).

Your child's doctor may want to periodically conduct a dietary assessment to ensure your child is getting adequate amounts of calcium and vitamin D. If levels are too low, they may recommend additional supplements.

GUIDELINE RECOMMENDATIONS

Your child should have a dietary assessment done once a year to determine calcium and vitamin D intake.[1]

Calcium and vitamin D supplements should be added as needed.[1]

Neurological[1,2]

As with younger children, neurological complications can occur in school-age children with galactosemia. Some neurological complications that may occur are impaired motor function, impaired coordination (ataxia), involuntary muscle contractions or movements (dystonia), and involuntary rhythmic shaking in one or more body parts (tremor). Your child's doctor will continue screening for these complications to monitor any changes over time and ensure the earliest possible intervention. Your child's metabolic geneticist or neurologist will complete a full neurological exam and look at muscle strength and tone, coordination, walking, balance, and assess for tremors. They may also ask you if your child has experienced any seizures and ask questions about thinking and memory. For more information about seizures, please see ***page 36*** in the ***First Year of Life*** chapter. Depending on your child's symptoms, they may recommend physical or occupational therapy.

GALNET REGISTRY DATA

52% of participants across all ages experienced neurological complications.[2]

35% of participants who experienced tremor were between preschool age and the second decade.[2]

GUIDELINE RECOMMENDATION

Your child should be screened about every 6 months for neurological involvement from age 2-3 years. This usually involves a clinical exam to look for any disorders.[1]

Developmental Follow-Up

To ensure your child is receiving necessary therapies, you and your child's doctor will want to periodically assess your child's cognitive development. Using standardized tools such as the Wechsler Intelligence Scale for Children (WISC) can help identify your child's strengths and where additional support might be needed.[1]

GALNET REGISTRY DATA

40% of participants experienced cognitive delays in infancy/childhood.[2]

Some studies have found people with classic galactosemia (CG) may have lower levels of executive function.[1] Some of the skills included in executive function are displaying self-control, consistently following multiple-step directions, remaining focused despite

distractions, and planning to meet goals.[46] These functions are rapidly developed between the ages of 3 to 5 years and adolescence. In children, standardized tools such as the Behavior Rating Inventory of Executive Function (BRIEF) are used to assess executive function. If the BRIEF or other screening tool indicates an abnormal result, your provider will follow-up with appropriate intervention (e.g., psychotherapy or medication).[1]

GUIDELINE RECOMMENDATION

Your child's cognitive development and executive function should be assessed at age 8-10 years. There should be a specific focus on processing speed and visual spatial comprehension.[1]

Speech and Language

If your child has been receiving speech therapy, check in with their speech-language pathologist to discuss progress and assess whether any changes to their plan are needed. If your child has not yet required services for speech, screening remains important during this time. As with younger age groups, it is important to monitor and, if needed, assess your child's speech and language to determine whether intervention is necessary. Screening and follow-up assessment at this stage may include hearing, receptive, expressive, and pragmatic language, a structure-function and motor speech examination looking at respiration, resonance, voice, and speech intelligibility for children not meeting age-expected milestones (see **Table 7** for more detail).[1] Slower processing times, a symptom of a language disorder, often become more apparent at this age. Your child may withdraw as instructions and conversations move too quickly, impacting comprehension and participation in academic and social situations. This withdrawal may be interpreted as shyness.

Table 7: Common speech and language complications assessed during screening[34,35]

Complications	Characteristics
Receptive language	Inability to take in and understand information
Expressive language	Inability to put thoughts into understandable words and sentences
Pragmatic language	Inability of an individual to use language appropriately in social situations (for example, waiting for others to finish speaking before beginning to speak)
Resonance	Amount of air and vibrations in the nose and mouth during speech
Hypernasality	Excessive air and vibrations passing through the nose (sounds like the quintessential telephone operator)
Hyponasality	Too little air and vibrations passing through the nose (sounds like a person with a bad cold)
Articulation	How correctly each sound is produced by the tongue, lips, jaws, and vocal cords

GUIDELINE RECOMMENDATION

Your child should be screened for speech and language delays at age 5 years.[1]

Psychosocial and Mental Health

Emotional and mental health and behavioral challenges have been observed at increased prevalence in people with galactosemia.

GALNET REGISTRY DATA

44% of participants across all age groups experienced mental health and behavioral problems.[2]

37% of participants experienced anxiety between preschool and their second decade of life.[2]

7% of participants experienced attention-deficit hyperactivity disorder (ADHD).[2]

6% of participants exhibited behaviors on the autism spectrum.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for psychosocial deficits including autism spectrum disorders, sensory integration problems, depression, and anxiety using standardized tools such as the Behavior Assessment System for Children (BASC-3) at age 8-10 years.[1]

Bone Health

Bone health may be affected in people with classic galactosemia.[1] Although not fully understood, factors that may contribute include vitamin D and calcium deficiencies that can be associated with the galactose-restricted diet, decreased physical activity, and primary ovarian insufficiency (POI) in girls and women. The ovaries produce estrogen and other essential hormones that play a role in puberty, bone health, heart health, and brain function.[47] Hormones can affect your bone health,[48] so you may consult with an endocrinologist if you are concerned about your child's bone health.

GALNET REGISTRY DATA

27% of participants across all age groups experienced diminished bone mineral density.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for bone mineral density from age 8-10 years and again after puberty if normal.[1] Some metabolic geneticists may check as early as age 4-5 years.

Reaching Puberty and Hormonal Complications

According to the GalNet registry, 80% of girls with CG develop POI,[2] which means they may not naturally begin menstruating without therapeutic intervention and may experience delays in the development of secondary sex characteristics (e.g., breast development, widening of hips).[1] Some girls with CG undergo normal spontaneous pubertal development followed by irregular menstrual cycles and eventual complete cessation of menstrual cycles. POI can also lead to fertility issues later in life.[1] The ovaries produce estrogen and other essential hormones that play a role in puberty, bone health, heart health, and brain function.[47]

GALNET REGISTRY DATA

49% of female participants experienced delayed/induced puberty.[2]

POI was reported in 80% of participants.[2]

Because some hormonal replacement therapies may help induce puberty when not reached spontaneously, it is important to start discussing options and referrals as early as possible in your child's development.[1] Some doctors may start this conversation between age 9 and 12 years. One small study (22 participants) found that 28% of girls as young as age 8 years worried about possible infertility.[49] Your child's doctor may refer your child to a pediatric endocrinologist, pediatric or adolescent gynecologist, or reproductive endocrinologist to determine the therapy type and age at which to start treatment. Although data are limited, evidence suggests that most boys with CG do not experience the same deficiencies in sex hormones or delayed puberty and do not appear to have reduced fertility.[1]

Navigating School Services

As your child enters primary school, it is important to ensure appropriate support services are in place, if needed. If your child already participated in a preschool special education program, some of the processes for acquiring primary school services will be familiar.

If your child was not enrolled in a preschool program, the first step is obtaining an evaluation for special education. The school provides this service for free, or you can choose to seek an evaluation from an independent provider (be sure to coordinate with the school before scheduling).[37] Once the evaluation is complete, the **school's individualized education plan (IEP)** team decides whether your child is eligible for special education and, if so, you and your child's therapists will work with their teachers to provide input to develop an IEP. The IEP outlines the services that may help address your child's needs. Your child must have an IEP to receive special education services at school.[37] The IEP will change over your child's school career, so you will want to stay informed. You are an important member of the IEP team. Playing an active role and continuously advocating for your child throughout the process will help ensure your child receives the support needed to succeed with their learning goals in school. It is important to remember that other services not directly related to your child's learning may not be provided by the school (e.g., physical therapy).[37]

Your child's rights to access special education, including speech therapy, are covered by the **Individuals with Disability Educational Act (IDEA)**.[37] Under IDEA, there are 13 different eligibility categories. Children with galactosemia are often classified as **"other health impaired" (OHI)**. The OHI designation enables a child to access any service needed without necessarily meeting the handicapping criteria for that specific disorder. If your child does not qualify for special education, their rights at school are covered by **Section 504 of the Rehabilitation Act**.

The 504 plan outlines specific details about how the school will support your child with their disability (this includes any condition that limits daily activity in a major way). It ensures your child a safe diet and inclusion in activities. These plans are different from IEPs because they are not part of special education. One of the major differences is that 504 plans outline accommodations that the school will provide for your child. For example, a child with attention-deficit hyperactivity disorder (ADHD) who is easily distracted may be seated at the front of the classroom. Unlike the IEP process, there is no universal set of guidelines about the format of 504 plans, and it is largely left to the individual school. Schools are required to put their policies regarding 504 plans in writing. Ask to see a copy of your school's policies when you are ready to put a plan in place for your child.

Once a 504 plan and/or IEP is put in place, you will have a dedicated team within the school comprised of a case manager (often a social worker or therapist), a school psychologist, and special education teachers and administrators.

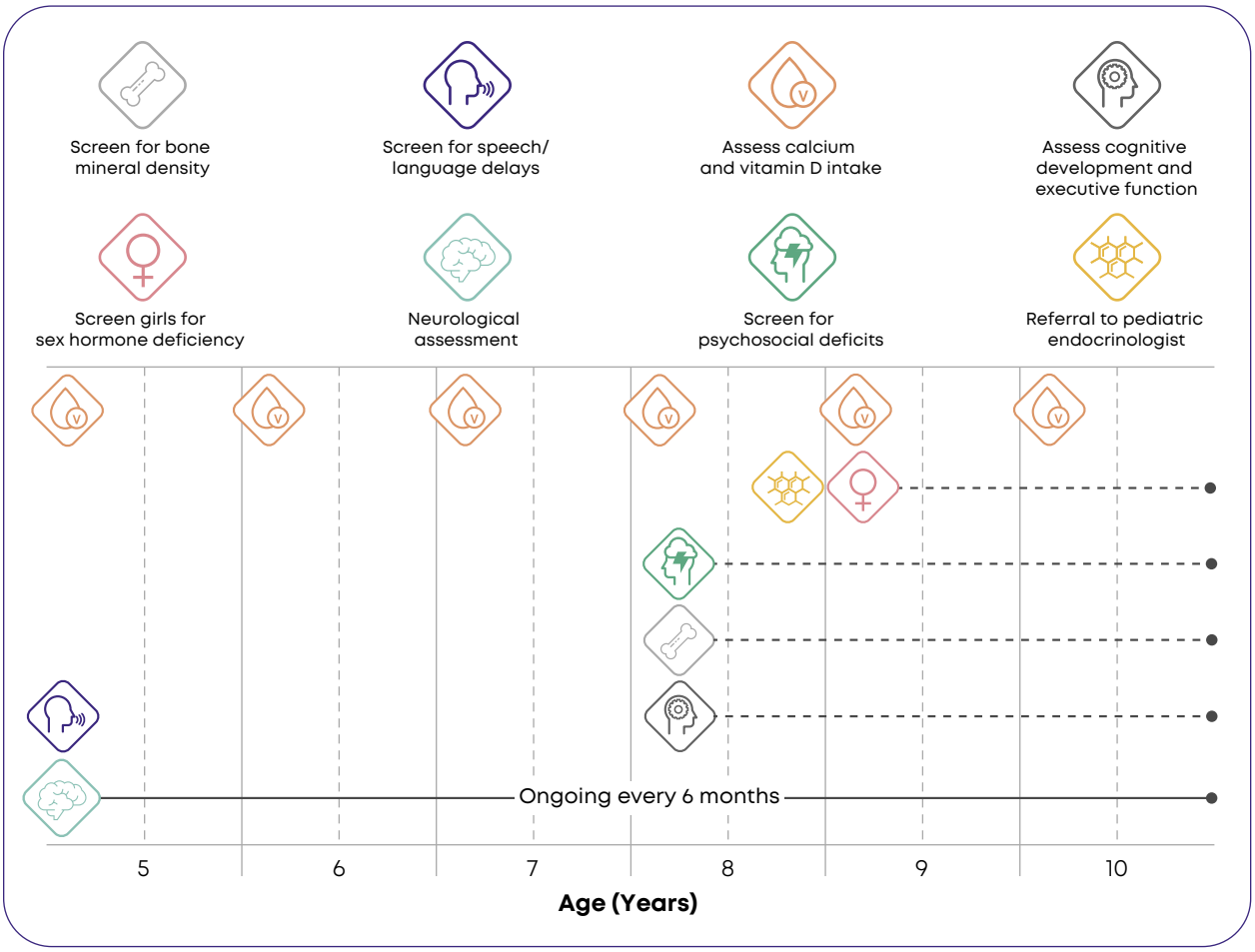
"For my daughter, school is a struggle. Memory is hard. Everything takes 3-4 times as long, so remember to be patient as your child learns at their pace. We receive lots of services in and outside of school. It's important to be a constant advocate for your child while they're in school so they receive the support they need."

– Parent of a 7-year-old with galactosemia

Advice About Education and Advocating from Families in the Galactosemia Community

- ✔ When it comes to advocating for access to educational support services, you are the expert on galactosemia. Educate school administrators and teachers about galactosemia, as you work together as a team to provide your child with the support they need to meet their goals.
- ✔ Prior to the IEP meeting, write down what you think may be affecting your child's educational performance and bring this up at the meeting. Try and meet with the special education teacher before your IEP meeting to align on strategies and concerns ahead of time.
- ✔ Some accommodations may include shortened assignments, preferred seating, having tests read aloud, and taking tests in a different room.
- ✔ Consider requesting extra time for taking exams, extended deadlines for homework assignments, etc.
- ✔ Remember that you and your child have options. If your school is not providing what your child needs, work with the district to find a school placement where their needs will be met.
- ✔ Make sure you advocate for the classroom environment that is best for your child. This could mean having an aide present for extra support, or a separate space for certain lessons for more individualized attention, or a smaller classroom.
- ✔ Parents must approve changes to the IEP/504, so it is important to consider any changes before signing. It's also okay to take the documents home after the meeting to give time to review them before signing.

Primary School: Monitoring and Assessments[1]



Adolescence

(Ages 10-18)

Adolescence (Ages 10-18)

Introduction

Adolescence is a time full of physical, emotional, and mental changes. An adolescent is in the process of shaping social interactions and building self-confidence. They may be aware of differences from their peers. They can participate in their education and healthcare plans more actively. Around the age of 12 years, they may become more interested in and knowledgeable about their medical condition, as well as their educational needs, and have a desire to be more independent. The adolescent may test the rules or refuse to adhere to the diet. This is normal. Continue to give a clear message about expectations. It is also important to remember that young people scare themselves the most. So, by doing their best to maintain rules and expectations, parents provide security for the adolescent. Some find that planning ahead for the transition to independent adult life during high school can be helpful.

Joining us at the conference?
Look for on- and off-site activities for these groups:
Mighty G program is for youth (ages 9-11)
G-Force program is for teens (ages 12-18)

Adolescence: Considerations for the Galactose-Restricted Diet

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information).

GUIDELINE RECOMMENDATIONS

Your child should have a dietary assessment done once a year to determine calcium and vitamin D intake.[1]

Calcium and vitamin D supplements should be added as needed.[1]

Adolescents may take an active role in their diet and meal planning. They may learn how to read food labels, advocate for themselves at restaurants, and educate their peers about their galactosemia and diet restrictions. This can inspire confidence and prepare them for adult life.

Neurological[1,2]

As with younger age groups, neurological complications can occur in adolescents with galactosemia. Some neurological complications that may occur are impaired motor function, impaired coordination (ataxia), involuntary muscle contractions or movements (dystonia), and involuntary rhythmic shaking in one or more body parts (tremor). Your child's doctor will screen for these complications to monitor any changes over time and enable the earliest possible intervention. The metabolic geneticist or neurologist will complete a full neurological exam and look at muscle strength and tone, coordination, walking, balance, and assess for tremors. They will also ask if your child has experienced any seizures and ask questions about thinking and memory. For more information about seizures, please see ***page 36*** in the ***First Year of Life*** chapter. Depending on your child's symptoms, your child's doctor may recommend physical or occupational therapy.

GALNET REGISTRY DATA

52% of participants across all ages experienced neurological complications.[2]

35% of participants who experienced tremor were between preschool age and the second decade.[2]

GUIDELINE RECOMMENDATION

Your child should be screened about every 6 months for neurological involvement from age 2-3 years. This usually involves a clinical exam to look for any disorders.[1]

Developmental Follow-Up

Your adolescent is going through a lot of changes, so it is important to ensure cognitive development is on track through regular assessments.

Identifying strengths and challenges using standardized tools such as the Wechsler Intelligence Scale for Children (WISC), can help establish appropriate support services.[1] Tools such as the Cambridge Neuropsychological Test Automated Battery (CANTAB), the NIH Cognition Toolbox, or other standardized tests, can be used to assess executive function.[1]

GALNET REGISTRY DATA

40% of participants experienced cognitive delays in infancy/childhood.[2]

GUIDELINE RECOMMENDATION

Your child's cognitive development and executive function should be assessed at age 12-14 years. There should be a specific focus on processing speed and visual spatial comprehension.[1]

Some studies have found people with classic galactosemia (CG) may have lower levels of executive function.[1,2] Executive functioning skills include displaying self-control, consistently following multiple-step directions, remaining focused despite distractions, and planning to meet goals.[46] In adolescents, standardized tools such as the Behavior Rating Inventory of Executive Function (BRIEF) are used to assess executive function.[1] If the BRIEF or other screening tool indicates an abnormal result, your child's doctor will follow-up with appropriate intervention (e.g., psychotherapy or medication).

Speech and Language

Continue working with your speech-language pathologist as long as needed and reach out to Apraxia Kids at www.apraxia-kids.org for additional support. For more information on speech and language issues, see the ***Primary School (Ages 5-10)*** chapter.

Psychosocial and Mental Health

Emotional health, mental health, and behavioral challenges have been observed in people with galactosemia. Adolescents may be resistant to psychotherapy or counseling. However once engaged with a therapist, most find it helpful. Sometimes, talking with another adult (e.g., teacher, clergy, or coach) can be helpful. Peer support is sometimes the best option. The Galactosemia Foundation provides opportunities for social support. Another good resource is Next Step, a peer support program for young adults with rare genetic diseases. They offer in-person programs as well as a variety of virtual conferences throughout the year (www.nextstepnet.org).

GALNET REGISTRY DATA

55% of participants experienced anxiety after they turn 10 years old.[2]

44% of participants across all age groups experienced mental health and behavioral problems.[2]

Boys and men with galactosemia experienced mental health and behavioral problems at a higher frequency as they grew older.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for psychosocial deficits including autism spectrum disorders, sensory integration, depression, and anxiety using standardized tools such as the Behavior Assessment System for Children (BASC-3) at age 12-14 years.[1]

Bone Health

Bone health may be affected in people with galactosemia.[1] Although not fully understood, factors that may contribute include restricted diet, decreased physical activity, and primary ovarian insufficiency (POI) in girls and women.

GALNET REGISTRY DATA

27% of participants across all age groups experienced diminished bone mineral density.[2]

10% of participants across all age groups experienced fractures.[2,a]

[a]Number of fractures did not differ from that in general population but the median age at which they occurred in individuals with galactosemia was younger.[2]

GUIDELINE RECOMMENDATION

Your child should be screened for bone mineral density after puberty is complete and every 5 years thereafter.[1]

Endocrinology Follow-Up

Girls and women with CG often have deficiencies in sex hormones and loss of ovarian function.[50] The loss of ovarian function is a condition known as primary ovarian insufficiency (POI).[51] 80% of girls and women with CG develop POI,[2] meaning they may lack menstrual

periods and development of secondary sex characteristics (e.g., breast development, widening of hips) without intervention (hormone replacement therapy).[1] The ovaries produce estrogen and other essential hormones that play a role in puberty, bone health, heart health, and brain function.[47]

GUIDELINE RECOMMENDATION

Girls and women should be screened for deficiencies in sex hormone production if they have not developed secondary sex characteristics by age 12 years or have not started their menstrual period by age 14 years.[1] Screening should include follicle-stimulating hormone (FSH) and 17-beta-estradiol. Your daughter's doctor may refer you to a pediatric endocrinologist for evaluation.[1]

If you have not already done so, consider talking to your daughter's doctor about referral to a pediatric endocrinologist. As your daughter enters adolescence, they may notice their body is different from their peers'. This could impact self-esteem and socialization.[1] One small study (22 participants) found that 28% of girls as young as age 8 years worried about possible infertility.[49] Talk with your daughter and her doctors about how you can best support her. Your daughter's doctor may suggest hormone therapy to induce puberty. They will likely consider when to start therapy based on her hormone levels (specifically FSH), growth, and psychological maturity.[1] Anti-Müllerian hormone (AMH) is a well-recognized estimate of the number of follicles or eggs remaining in the ovary.[52] Girls or women with undetectable or very low AMH are at significant risk of POI (early-onset menopause).[53] For women who are still ovulating, however, it does not predict their current fertility.[54] If your daughter is interested in pregnancy, you may want to consider referral to a reproductive endocrinologist to discuss the limitations in natural conception, as well as fertility treatment options and options for fertility preservation. Additionally, during your daughter's reproductive window, your daughter's doctor may discuss contraception counseling if pregnancy is not desired.

GALNET REGISTRY DATA

49% of female participants experienced delayed/induced puberty.[2]

POI was reported in 80% of participants.[2]

84% of participants with POI reported the use of hormone replacement therapy (HRT).[2]

Median age of start of HRT was 16 years (ages ranged from 11-45 years).[2]

GUIDELINE RECOMMENDATIONS

For puberty inducement, a low-dose estrogen in a stepwise escalating dose is used, then later combined with cyclic progesterone on a monthly basis for regular withdrawal.[1]

Once regular menstrual periods have been established, annual monitoring for menstrual abnormalities, loss of menstrual periods, and symptoms of POI should be conducted.[1]

Despite limited data, fertility does not appear to be affected in boys and men with galactosemia.[1]

Advocating for Your Adolescent in the School System

As your adolescent approaches adulthood, it can be scary to think about allowing them to make safe and healthy decisions independently. You can help prepare your child for independence by teaching them self-advocacy. Self-advocacy is an important skill for your adolescent to learn.[55] If your child has an individualized education plan (IEP) in place, then by the time they are 16 years old, you will begin drafting a transition plan with their IEP team, which may include a case manager (often a social worker or therapist), a school psychologist, and special education teachers and administrators.[37] This transition plan will map out specific goals for your child to accomplish to prepare them for independent adult life.

Encourage your child's participation in IEP or other meetings with the school regarding their disability. Share your knowledge about laws and regulations so they understand how their rights are protected. This will help prepare them for adulthood when they will be making decisions about their own future.

Outside of the IEP in the school system, many parents seek opportunities for their child to gain work/study experiences through local businesses and programs. It may be helpful to seek advice from a counselor who specializes in working with students with learning disabilities. Explore private schools in the area that might have such a resource person on staff who might be willing to talk with you. They may have insights into universities (e.g., tutoring centers) and community programs that can provide support. The Center for Autism Research's CAR Autism Roadmap website (www.carautismroadmap.org) is a good resource for supporting the transition from high school.

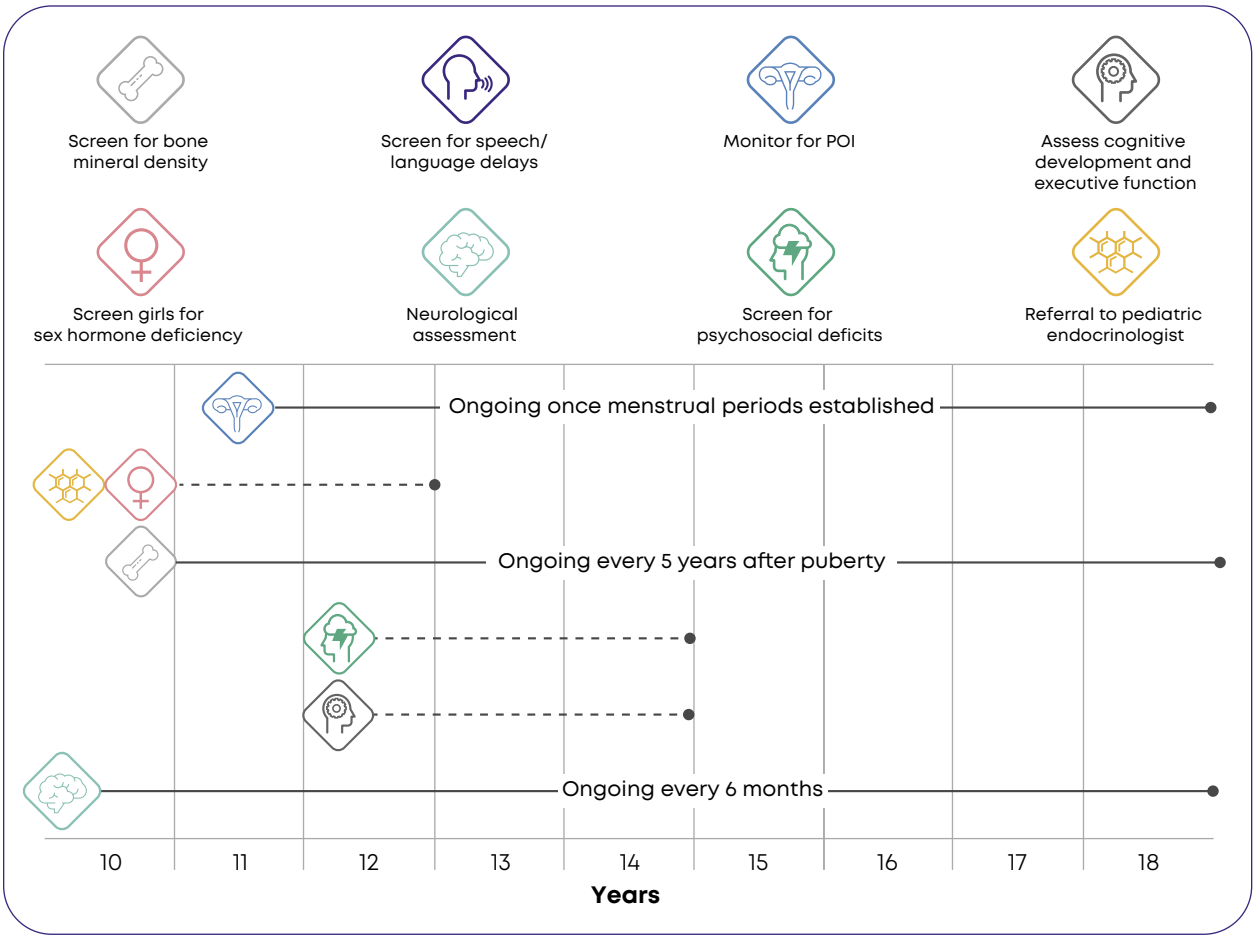
PARENT TIPS

ACCOMMODATIONS CAN BE MADE FOR UNTIMED TESTING OR EXTRA TIME FOR STANDARDIZED TESTS AS WELL AS LOWER COURSE LOADS IN HIGH SCHOOL AND COLLEGE.

SOME STATES OFFER CAMPS TO HELP CHILDREN DEVELOP SOCIAL SKILLS. THESE CAMPS CAN ALSO PROVIDE VALUABLE GUIDANCE FOR PARENTS REGARDING OPTIONS FOR COLLEGE PROGRAMS THAT PROVIDE SUPPORT.

WHEN LEARNING HOW TO DRIVE, IT MAY BE NECESSARY TO HAVE A GPS REQUIREMENT IN THE VEHICLE.

Adolescence: Monitoring and Assessments[1]



Adult Life

(Age 18 and older)

Adult Life (Age 18 and Older)

Introduction

While some adults with classic galactosemia live independently, you may be concerned about your or your child's ability to do so as an adult. It is important to remember that galactosemia will not prevent you or your child from living a fulfilling life. Despite challenges, one study indicated most adults feel their coping with galactosemia is "good" or "very good".[49] The same study indicated only a small minority of adults (8%) reported that galactosemia had a negative effect on family life.[49] Remember that every person is different. Not everyone is ready to graduate high school at the age of 18. In fact, many states offer the opportunity to extend high school graduation until the age of 21 for students who are receiving special education services. Check if this is an option if you have concerns about readiness.

Joining us at the conference?
Look for on- and off-site activities for these groups:
Generation G program is for young adults with galactosemia no longer in high school (Recommended ages 19-25)
Agers program is for adults with galactosemia (Recommended ages 25+)

Adult Life: Considerations for the Galactose-Restricted Diet

Please see ***The Galactose-Restricted Diet (Current Standard of Care)*** chapter for detailed information).

One study of over 400 adults with galactosemia in 12 countries found nearly a third of individuals did not have continued care with a dietician at their healthcare center (defined as no contact for at least 2 years).[56] Maintaining contact with a dietician in adulthood can help people take control of their nutrition. A dietician may assist by customizing a plan and ensuring there are no major nutrient deficiencies.

Neurological[1]

As in childhood, it is still important that adults with galactosemia are periodically monitored for neurological concerns. This is important for tracking any changes in symptoms over time. Physical therapy, occupational therapy, or medical treatment is needed for specific neurological signs or symptoms.

GALNET REGISTRY DATA

52% of participants across all ages experienced neurological complications.[2]

41% of participants experienced tremor after the second decade of life.[2]

GUIDELINE RECOMMENDATION

Adults should be screened annually for neurological involvement. This usually involves a clinical exam to look for any disorders.[1]

Developmental Follow-Up

Although studies have found there is no evidence of a decline in cognitive function in adults with galactosemia, it may be important to screen periodically to ensure any issues are caught early. Standardized tools include the Cambridge Neuropsychological Test Automated Battery (CANTAB) or other similar tools.[1]

GUIDELINE RECOMMENDATION

Executive function should be assessed in young adults (age 18-20 years).[1]

Speech and Language

Continue working with your speech-language pathologist as long as needed. For more information on speech and language issues, see the ***Primary School (Ages 5-10)*** chapter.

Psychosocial and Mental Health

While social adjustment challenges in people with galactosemia may be more likely to occur before the age of 20, mental health or psychiatric concerns may be more prevalent after a person's 20s.[2]

Adults with galactosemia may have difficulty navigating socially (e.g., managing relationships and engaging in social activities).[1] It can be helpful for adults with galactosemia to remain connected to their support network and others who may be facing similar difficulties. Peer support networks often provide an opportunity for exchange of information such as coping skills and problem-solving strategies. Adults can also find support from mental health professionals.

GALNET REGISTRY DATA

44% of participants across all age groups experienced mental health and behavioral problems.[2]

13% of participants experienced depression most commonly after they turned 20.[2]

55% of participants experienced anxiety after they turned 20.[2]

GUIDELINE RECOMMENDATION

Adults should be screened for mental health issues with validated questionnaires that include scales for anxiety and depression (e.g., National Institutes of Health [NIH] Patient-Reported Outcomes Measurement Information System [PROMIS], Beck Anxiety Inventory [BAI], and Beck Depression Inventory [BDI]).[1]

Bone Health

As with younger age groups, bone health may be affected in adults with classic galactosemia (CG).[1] Although not fully understood, factors that may contribute include restricted diet, decreased physical activity, and primary ovarian insufficiency (POI) in girls and women. POI in women may be a contributing factor to impaired bone health. Therefore, it is important to monitor bone mineral density in adulthood. This is usually done using dual-energy X-ray absorptiometry (DEXA) scan ordered by a doctor.[1]

GUIDELINE RECOMMENDATION

Both men and women with galactosemia should be screened for bone mineral density every 5 years.[1]

GALNET REGISTRY DATA

27% of participants across all ages experienced diminished bone mineral density.[2]

10% of participants experienced fractures.[2,a]

Median age of participants with fractures was 24 years (age range of 6 to 59 years).[2]

[a]The number of fractures was not higher than in the general population however, the median age was relatively young. It is not known if bone health issues are a secondary result of the diet and vitamin D/calcium deficiency, a result of the disease, or both.[2]

GUIDELINE RECOMMENDATION

Adults should have a comprehensive dietary evaluation and optimize their calcium and vitamin D intake. In women, hormonal status should be evaluated, and hormone replacement therapy considered. Adults should also participate in regular exercise and have any skeletal problems and clinically significant fractures assessed.[1]

Endocrinology and Fertility in Women with Galactosemia

By now, you or your daughter likely have an established relationship with a reproductive endocrinologist. Please refer to the ***Adolescence (Ages 10-18)*** chapter for more information on endocrinology and POI. There are significant benefits to continuing hormone replacement therapy in adults with POI as evidence supports a role in bone protection and cardiovascular health.[57] Because of the frequency of POI in women with galactosemia, most but not all women with CG may experience fertility challenges in adulthood.[1] It is important to reach out to a reproductive endocrinologist early when considering family planning. Although the chances of pregnancy are reduced in women with CG, natural pregnancy is a possibility for at least some women with galactosemia.[1] The most established and successful treatment option for women with galactosemia who have POI and want to become pregnant is egg donation. While long-term studies of pregnancy outcomes in women with CG are scarce, small cohorts suggest no adverse effects on the infant or mother. In addition to discussion of pregnancy, it may also be helpful to explore options for potential fertility preservation if desired.

GUIDELINE RECOMMENDATIONS

Women should be monitored annually for menstrual abnormalities, loss of menstrual periods, and symptoms of POI.[1]

Women should ask about referral to a reproductive endocrinologist if they desire pregnancy and have been unable to conceive naturally or desire additional counseling about fertility treatment options including oocyte (egg) donation.[1]

"The journey to parenthood isn't the same for everyone. What I feel is most important is that people know their options and have a reproductive healthcare provider that helps them navigate their choices."

– Jessica B. Spencer, MD, MSc

Cataracts

As in newborns, the development of cataracts has been observed in adults.[1] Cataracts are a condition in which the lens of the eye becomes cloudy and often results in vision problems, such as blurriness and defective night vision.[30] Cataracts can be surgically removed and the lens replaced with an artificial version. Experts tend to agree that if no issues are detected in the eyes shortly after birth and adherence to a galactose-restricted diet is good, eye exams can be conducted on an as-needed basis.

GALNET REGISTRY DATA

11% of participants experienced cataracts in adulthood.[2]

GUIDELINE RECOMMENDATION

Adults who do not adhere to a galactose-restricted diet should have more frequent ophthalmological screening.[1]

Adult Life Skills

Note: This section is intended for adults with galactosemia and the parents of those individuals who are experiencing complications into adulthood.

Although you or your adult child may experience challenges in adulthood, proactive planning can help alleviate some of these challenges. At this stage, it is likely you are working with your or your child's care team on implementing a transition plan from any individualized education plan (IEP),[37] with specific goals intended to prepare for independent adult life.

It can be helpful to start practicing self-advocacy skills and start expanding a support network. Exploring volunteer opportunities for you or your child can start the process.

If you or your child has cognitive or other developmental challenges, you may be worried that finding employment and living independently could be a challenge. The Americans with Disabilities Act does not require employers to hire applicants with disabilities, but it does provide protection against discrimination.[58] Parents can encourage their child to learn about their protected rights to advocate for themselves. Many states and the federal government also offer career counseling and housing programs for people with disabilities.[58-60] Contact your state Department of Rehabilitation to learn about training and job programs that might assist with job preparation and employment.

PARENT TIPS

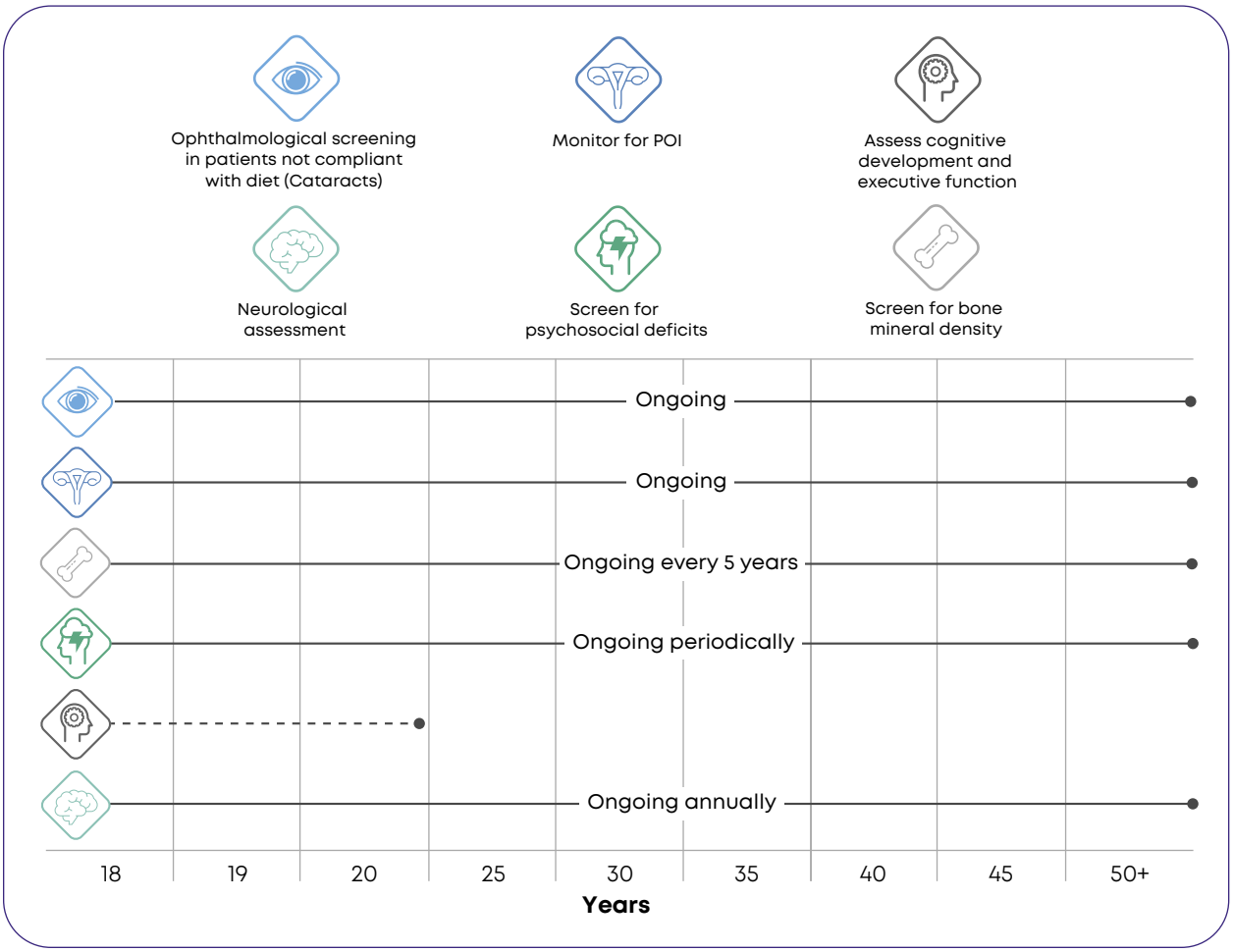
ACCOMMODATIONS CAN BE MADE FOR UNTIMED TESTING OR EXTRA TIME FOR STANDARDIZED TESTS AS WELL AS LOWER COURSE LOADS IN HIGH SCHOOL AND COLLEGE.

WHEN LEARNING HOW TO DRIVE, IT MAY BE NECESSARY TO HAVE A GPS REQUIREMENT IN THE VEHICLE.

Special Needs Trust Funds

Some parents choose to establish a plan for their adult child's care should they no longer be able to manage the process. It can provide security for parents to ensure that their child will be cared for in their absence. Special Needs Trusts provide a mechanism to preserve your child's eligibility for needs-based government benefits such as Medicaid and Supplemental Security Income.[61] These trusts allow a trustee to supplement a beneficiary's government benefits. These supplemental funds can cover the cost of a caregiver, as well as medical and dental expenses.

Adult Life: Monitoring and Assessments[1]



Resources

Resources

Advocating for Your Child: Breakdown of symptoms and specialists

Symptom	Specialist
Abnormal liver enzymes, blood clotting problems, signs of infection	Neonatologist
Brain injury	Neurologist
Cataracts and vision concerns	Ophthalmologist
Feeding complications	Feeding specialist or speech-language pathologist
Hypoglycemia	Neonatologist
Anxiety	Mental health professional
Delayed appearance of secondary sex characteristics	Pediatric endocrinologist
Depression	Mental health professional
Difficulty with attention span, trouble organizing tasks, forgetfulness[38]	Mental health professional
Difficulty with socialization, avoidance of eye contact, lack of facial expressions, restricted or repetitive behaviors, developmental delays[38]	Mental health professional
Diminished muscle coordination, signs that your child is not aligning muscular action with proper distances (over-stepping or -reaching; dysmetria), involuntary muscle contractions, involuntary and rhythmic shaking in one or more body part (tremor)	Neurologist
Fractures	Endocrinologist
Seizures	Neurologist
Speech and language issues	Speech-language pathologist

Websites

The internet can be an incredible resource for information, but also for *misinformation*. Always discuss anything you learn from the internet with your child's care team. We've started a list here of reputable resources.

- Galactosemia Foundation: **www.galactosemia.org**
- Go Dairy Free website: **www.godairyfree.org**
- Understood (nonprofit) website: **www.understood.org**
- National Center for Learning Disabilities website: **www.ncld.org**
- Apraxia Kids (**www.apraxia-kids.org**):
  - List of summer camps for speech and language: **www.apraxia-kids.org/summer-speech-language-camps-for-apraxia/**)
- Video: Understanding Galactosemia Symptoms & the Lifelong Impacts of Speech and Language (**https://www.youtube.com/watch?v=tsquul7DE9A&t=9s**)
- Daily Med website: **https://dailymed.nlm.nih.gov**
- Centers for Disease Control and Prevention website (support for navigating early intervention): **https://www.cdc.gov/ncbddd/actearly/parents/states.html**
- Early Childhood Technical Assistance Program website (list of 619 coordinators): **https://ectacenter.org/contact/619coord.asp**
- Center for Autism Research's CAR Autism Roadmap website: **www.carautismroadmap.org**

Mobile Applications

Accessing information on the fly is made easier with the use of mobile applications on your device.

- CDC Milestones App

Glossary

Allele: An allele is a version of a gene. An individual inherits two alleles of each autosomal gene, one from each parent.

Autosomal: Trait that is not sex-linked and can occur in males and females

Ataxia: Impaired coordination

Babble Boot Camp: An early intervention program of activities and routines designed for infants and toddlers during the pre-speech and very early speech and language stages

Behavior Rating Inventory of Executive Function (BRIEF): Standardized tool used to assess executive function in children and adolescents

Cambridge Neuropsychological Test Automated Battery (CANTAB): Standardized tool used to assess cognitive function including attention and psychomotor speed, executive function, memory, and emotional and social cognition

Carrier: Individual with only one copy of a mutated gene who does not have the disease

Cognitive: Relating to intellectual activity such as thinking, reasoning, and remembering

Dystonia: Involuntary muscle contractions or movements

Enzyme: Enzymes are proteins that facilitate biochemical reactions. They may be involved in the breakdown or generation of biochemical substances such as proteins, sugars, and molecules in the body.

Fermentation: Chemical breakdown of a material by microorganisms

Fine motor skills: Skills that involve small muscle coordination such as writing, grasping small objects, and buttoning clothing

Gene: Unit of genetic information that is passed from parent to child

Gross motor skills: Skills that involve large muscle coordination such as walking, running, and kicking

Hormones: Chemical messengers that coordinate organ function in the body

Inherited disorder: Condition caused by genetic changes which can be passed from parent to child

Metabolic: Chemical reaction converting food to a form of energy available to cells in the body

Metabolic disorders: Disorders that arise from abnormal chemical reactions in the body that convert food into energy

Metabolite: A metabolite is produced from the metabolism or breakdown of a substance.

Mutations: Changes in the genetic code within a gene

Rare disease: Orphan Drug Act defines a rare disease as less than 1 in 200,000 people in the United States[62]

Recessive mutation: Mutation that must be inherited from both parents in order for that genetic trait or disease to be expressed

Residual: Refers to the amount of enzyme activity that remains, despite a mutation

Sepsis: A potentially life-threatening infection resulting in a strong immune response that causes damage to the individual's own tissues

Standard of care: Treatment accepted by medical experts as the current treatment for a disease

Tremor: Involuntary rhythmic shaking in one or more body parts

Wechsler Intelligence Scale for Children (WISC): Standardized tool used to assess intellectual ability in children and five cognitive domains

Wechsler Preschool and Primary Scale of Intelligence (WPPSI): Standardized tool used to assess cognitive development for young children

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Notes

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Notes

This galactosemia handbook presents current information for each life stage, including various symptoms that may appear. Cited recommendations are based on the international clinical guideline, peer-reviewed data, expert insights, and first-hand experience from the galactosemia community.
As you review this handbook, please keep in mind that every person develops differently. Consult with your or your child's care team regarding any specific concerns you have or any questions regarding potential therapeutic options or approaches. The information presented here is not meant to replace advice from a healthcare provider and does not constitute a medical diagnosis or advice.

Galactosemia Foundation
Linked for Life

JAGUAR GENE THERAPY

This document was developed by the Galactosemia Foundation and Jaguar Gene Therapy, with support from experts in galactosemia and members of the galactosemia community.

Version 1.0.