Galactosemia Foundation

GALACTOSEMIA

Adult Workbook

Tools, Worksheets, and Trackers for Managing Life with Classic Galactosemia

Galactosemia Foundation

How to Use This Workbook

This workbook is the companion to the Adult Handbook. The handbook explains what adult Galactosemia looks like. This workbook gives you the tools to act on it, at appointments, at home, and across the stages of adult life.

Three ways to use each section

Every section is built in three layers. You do not pick a level, you pick a mode, and you can switch modes anytime. Fatigue, anxiety, illness, good days, busy days all change what you can take in. The workbook assumes that.

- Just Tell Me, one or two directives at the top of each section. If today is a hard day, do one of them and close the book. That counts.
- Walk Me Through It, step-by-step, plain language, with the full checklists, scripts, and trackers. Stop whenever you need to.
- I've Got This, a short scan strip at the end of each section. The whole section in one column for when you just want to skim and move.

Also in each section

- Worksheets and checklists matched to the handbook section of the same name.
- Language scripts you can bring into appointments or phone calls.
- Space to write. You are allowed to skip what is not useful.

Use what helps. Leave what doesn't. Progress counts, even when small.

Start Here

Nothing in this workbook is required reading. You do not need to start on page one. Enter wherever your life is right now, you can always come back to the rest later.

If you are...

- Newly transitioning from pediatric to adult care, start with Section 2: Transition to Adult Care.
- Coming back to healthcare after a gap, start with Section 1: Returning, Re-Entering, and Burnout.
- Trying to set up basic adult care, start with Section 3: General Adult Care and Section 3b: Annual Metabolic Follow-Up.
- Dealing with eye or dental issues, see Section 4: Vision and Dental.
- Working on nutrition, energy, or sleep, see Section 5: Nutrition, Movement, Energy, and Sleep.
- Navigating hormones, HRT, or POI, see Sections 6 and 7: Hormonal Health and HRT.
- Thinking about fertility or family-building, see Section 8: Fertility, Infertility, and Family-Building.
- Pregnant, planning a pregnancy, or postpartum, see Section 9: Pregnancy, Prenatal Care, and Breastfeeding.
- Focused on bone health or DEXA, see Section 10: Bone Health and DEXA.
- Tracking a tremor or motor changes, see Section 11: Tremor (includes the Archimedes spiral test).
- Noticing balance changes or worried about falls, see Section 12: Fall Risk and Home Safety.
- A man with CG looking for information that applies to you, see Section 13: Men's Health.
- Dealing with a rare but serious event (E. coli risk, surgery, acute illness), see Section 14: Rare but Serious Events.
- Fighting a denial or learning insurance, see Section 15: Barriers to Care, Insurance.
- Exploring SSI / SSDI, see Section 16: Disability Benefits.
- Thinking about work, reviews, or advancement, see Section 17: Career Growth.
- In a study, registry, or on an investigational drug, see Section 17b: Research and Clinical Trial Participation.
- Looking after your mental and social health, see Section 18: Mental Health and Social Health.

A few ways adults actually use this workbook

- The night before an appointment: flip to the matching section, write down your top two concerns, and bring it with you.
- Once a year: fill in the Annual Metabolic Follow-Up checklist and the tracker tables that apply.
- After a new diagnosis or life change: pick one section and work through just that one.
- When a denial letter shows up: go to Section 15 and use the glossary and appeal prompts.

You do not have to fill this workbook out. You get to use it. Skip what is not yours. Come back when it is.

Returning, Re-Entering, and Burnout

Matched to handbook section: Returning, Re-Entering, and Burnout

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Pick one item from the Starter Checklist below. Just one.
- If it has been over three years, start with finding one primary care provider to contact.
- Write down the one health concern you would raise first. That is the starting point.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Re-Entry Starter Checklist

You do not need to do everything. Pick one or two to begin.

- ☐ Identify one primary care provider to contact
- ☐ Gather any past medical records I already have
- ☐ Write down one health concern I want addressed first
- ☐ Schedule a first appointment (stability and prevention only)
- ☐ Decide whether a trusted person will come with me

What I Want From This Visit

Top concern: _____

One outcome I want: _____

What I do NOT want this visit to include: _____

Advocacy Fatigue Notes

When I have had to repeat myself, what usually helps:

_____

_____

Burnout is not failure. It is ongoing stress. You are allowed to re-enter at your own pace.

I've Got This

Scan and skip. The whole section in one column.

- Pick one provider to contact, not a whole system at once.

- Bring the records you already have; do not chase ones you do not.
- One concern per visit is enough, build back slowly.
- Schedule when your energy is on, not when it is empty.
- If the wait is long, a letter of medical necessity (which your metabolic team or PCP can write) can speed things up.

Transition to Adult Care (Ages ~16-25)

Matched to handbook section: Transition to Adult Care

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Ask your pediatric team for a written summary of your health history.
- Write down one adult provider name to look up this week.
- Put the word "transition" on your calendar, even if only as a note to yourself.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

When to Start

- Ages 14-16: Awareness and discussion
- Ages 16-18: Active planning
- Ages 18-21: Overlapping care with adult providers

Where I Am Now

☐ Just starting
☐ In progress
☐ Delayed or restarting
☐ Unsure

Doctor-to-Doctor Transition Checklist

☐ Asked pediatric team about transition timing
☐ Requested written medical summary
☐ Requested recent labs and imaging
☐ Asked for adult provider recommendation (including transition letter)
☐ Pediatric team agreed to overlap care
☐ Pediatric team sent records to adult providers
☐ Requested full medical records transfer from pediatric metabolic provider
☐ Asked for a full medical records transfer from current to new primary care team
☐ I have my Personal Health Snapshot

Current pediatric provider(s) and specialists:

_____

Transition planning started on: _____

What to Request from Pediatric Teams

- ☐ Diagnosis and genetic confirmation
- ☐ Baseline health summary
- ☐ Lab trends and imaging
- ☐ Medication history and reactions
- ☐ Emergency considerations
- ☐ Growth, development, and neuropsychological evaluations
- ☐ Bone density (DEXA) history
- ☐ Reproductive / endocrine history (POI status if applicable)
- ☐ Speech-language therapy records, if any
- ☐ Care coordination and social work notes

Ask How Records Are Shared

- ☐ Patient portal
- ☐ Printed copy
- ☐ Sent directly provider-to-provider
- ☐ Other: _____

Insurance at Transition

- ☐ Insurance reviewed, including aging-out timelines
- ☐ Confirmed adult metabolic provider is in-network
- ☐ Prescription coverage reviewed for current medications and supplements

HIPAA and Proxy Decision Guide

Now that I am 18, caregivers no longer have automatic access to my medical information. I can choose what to share.

Access Tool	Who I Want to Include	When It Applies

My Choice Right Now

- ☐ HIPAA authorization for: _____
- ☐ Limited medical proxy for: _____
- ☐ Shared scheduling / insurance access only
- ☐ I am handling care independently at this time

Medical Summary Template

Name: _____

Date of birth: _____

Diagnosis: Classic Galactosemia (GALT deficiency)

_____

Current medications / supplements: _____

Allergies: _____

Key past history (procedures, hospitalizations):

_____

Current providers: _____

Emergency contact: _____

Turning 18 is a legal milestone, not a switch. Build skills one at a time.

I've Got This

Scan and skip. The whole section in one column.

- Target ages 16-25; overlap between pediatric and adult teams is normal.
- Always request a full medical records transfer: medical summary, lab trends, DEXA history, reproductive/endocrine records, neuropsych, SLP records.
- Confirm in-network status, PA requirements, and prescription coverage before the first adult visit.
- A peer-to-peer call between pediatric and adult teams is worth asking for.
- HIPAA is a tool, not a wall, you decide who gets access.

General Adult Care

Matched to handbook section: Getting Back to Health: General Adult Care

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Book one primary care visit in the next three months if you have not had a primary care check-up in the last year.
- Put one CG-specific lab or scan on your provider's radar, DEXA, Vit D, or FSH/AMH if applicable.
- Write down the top concern you will bring to the next visit.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Annual Screening Checklist

Left column is the general adult baseline. Right column adds what international guidelines recommend specifically for adults with Classic Galactosemia. Both matter, the CG-specific layer does not replace standard screening; it is in addition.

Ages 18-39: General	**Ages 18-39: CG-Specific Additions**
☐ Blood pressure annually ☐ Cholesterol at least once ☐ Cervical cancer screening as recommended ☐ STI screening as appropriate ☐ Vaccinations up to date	☐ Baseline DEXA scan ☐ Vitamin D and calcium levels yearly ☐ For Women: FSH / estradiol / AMH if assigned female (POI can present early) ☐ Dilated eye exam (cataract check) ☐ Metabolic dietitian visit at least once a year ☐ Baseline neuropsychological evaluation ☐ Speech-language check if clarity or word-finding changes
Ages 40-49: General	**Ages 40-49: CG-Specific Additions**
☐ Continued annual primary care ☐ Cholesterol every 4-6 years ☐ Diabetes screening if risk factors present ☐ Perimenopausal symptom discussion when applicable	☐ DEXA on the schedule your metabolic team sets ☐ Tremor and balance check at annual visit ☐ Cognitive / memory check-in ☐ HRT review (dose, bone, cardiovascular) if on replacement ☐ Continued yearly metabolic follow-up ☐ Cataract monitoring
Ages 50-60: General	**Ages 50-60: CG-Specific Additions**
☐ Colon cancer screening (from age 45) ☐ Breast cancer screening as recommended	☐ More frequent DEXA (every 1-2 years) ☐ Formal fall-risk assessment

☐ Cardiovascular risk assessment ☐ Bone health discussion	☐ Cognitive screening (not only if concerned) ☐ Yearly cataract exam ☐ Cardiovascular risk read in context of long-term HRT and POI history ☐ Tremor tracking as a baseline, not only when new

Appointment Prep

Top concern today: _____

Second concern: _____

Questions to ask: _____

_____

_____

Lab Results Tracking

Date	Test	Result	Reference Range	Notes

My Adult Care Team

International guidelines recommend that adults with Classic Galactosemia be followed by a multidisciplinary team. Use this worksheet to list yours.

Role	Provider Name	Contact / Clinic

Role	Provider Name	Contact / Clinic

Common roles to include: metabolic specialist, primary care, endocrinology (bone/reproductive), OB-GYN or reproductive endocrinology, metabolic dietitian, neurology, mental health, speech-language pathology.

Team Coordination

- ☐ I have a lead provider who coordinates my CG-specific care
- ☐ My providers can share records with each other (release signed)
- ☐ I have asked my metabolic team to send a clinical letter to my PCP
- ☐ Baseline neuropsychological evaluation on file or scheduled

I've Got This

Scan and skip. The whole section in one column.

- General adult screening + CG-specific layer, both matter.
- Multidisciplinary team: metabolic, PCP, endo, OB/GYN, dietitian, neuro, mental health, SLP.
- DEXA, Vit D, FSH/AMH start in the 20s, not at 40.
- One lead provider coordinates, signed releases let the team actually talk.
- Use the Appointment Prep box before every visit; track labs in one place over time.

Annual Metabolic Follow-Up

Galactosemia-specific yearly check, based on international clinical guidelines

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Put one metabolic-team visit on the calendar for this year.
- Write down three questions you want to bring.
- Ask for a copy of the visit summary after the appointment.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Standard adult screening is not enough on its own. International guidance recommends that adults with Classic Galactosemia be seen yearly by a metabolic provider to review the areas below.

Yearly Review Checklist

- ☐ Diet review with metabolic dietitian (galactose restriction, calcium, vitamin D)
- ☐ Bone health, DEXA on recommended schedule
- ☐ Reproductive / endocrine, menstrual history, FSH/estradiol if applicable, POI follow-up
- ☐ Cognitive and speech, any new concerns in memory, word-finding, or speech clarity
- ☐ Motor, tremor, coordination, balance, new neurological symptoms
- ☐ Eye, cataract check
- ☐ Mental health, mood, anxiety, social functioning
- ☐ Review of all medications and supplements

Questions I Want to Bring

_____

_____

_____

Annual Visit Log

Year	Provider	Key Findings	Next Step

Year	Provider	Key Findings	Next Step

Even if you feel fine, one CG-focused visit a year keeps bone, hormone, and neuro changes from going unnoticed.

I've Got This

Scan and skip. The whole section in one column.

- One CG-focused visit a year, even when you feel fine.
- Covers: diet, bone, reproductive/endocrine, cognition/speech, motor, eye, mental health, meds.
- Feeding results into the Visit Log gives you a year-over-year record.
- Coordinate findings with your PCP, do not leave it to chance.

Vision and Dental Care

Matched to handbook section: Getting Back to Health, Vision and Dental

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Book one eye exam if you have not had one in the last 12 months.
- Ask specifically for a dilated or slit-lamp exam to check for cataracts.
- Tell your dentist you have CG.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Eye Care Tracker

- ☐ Last comprehensive eye exam date: _____
- ☐ Asked about dilated / slit-lamp exam (cataracts check)
- ☐ Current prescription recorded
- ☐ Any vision changes noted in the last year

Dental Care Tracker

- ☐ Last dental cleaning and check-up date: _____
- ☐ Next appointment scheduled
- ☐ Any pain, sensitivity, or bleeding noted
- ☐ Fluoride / preventive care plan discussed

Notes for Next Visit

_____

_____

Cataracts are the most common eye finding in Galactosemia. Yearly exams catch small changes early.

I've Got This

Scan and skip. The whole section in one column.

- Cataract check is the CG-specific eye priority, ask for dilated or slit-lamp.
- Yearly eye exam baseline, even if vision feels fine.

- Track exam dates so you do not forget who you saw for what and when.

Nutrition, Movement, Energy, and Sleep

Matched to handbook section: Nutrition, Movement, Energy, and Sleep

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Pick one small daily habit to start this week. Just one.
- Notice one energy pattern today, morning, afternoon, or evening, and write it down.
- Do not try to change everything at once. Sustainable is the goal.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Daily Food and Energy Log

Day	Meals / Snacks	Energy 1-5	Mood 1-5	Notes

Weekly Reflection

Days I felt best: _____

Days I felt worst: _____

Foods that helped: _____

Foods that made things harder: _____

Sleep quality this week (1-5): _____

Movement this week: _____

Sleep Tracker

Date	Bed Time	Wake Time	Quality 1-5	Notes

Date	Bed Time	Wake Time	Quality 1-5	Notes

Movement Log

Date	Activity	Minutes	Fatigue After 1-5

Consistency matters more than intensity. Low-energy days still count.

I've Got This

Scan and skip. The whole section in one column.

- Galactose-restricted diet stays lifelong; it is a foundation, not a phase.
- Calcium, vitamin D, hydration, and sleep are the non-negotiables.
- Rest is metabolic work. It is not laziness.
- Movement: consistency beats intensity. Five days a week of walking counts.
- Change and track one variable at a time so you can see what actually helps.

Hormonal Health

Matched to handbook section: Hormonal Health

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Write down your current hormone symptoms in your own words.
- Ask your OB/GYN or reproductive endocrinologist for baseline FSH, estradiol, and AMH.
- Write the date of your last period if you have one.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Hormonal Symptom Tracker

Check any you have noticed in the past month.

- ☐ Missed or irregular periods
- ☐ Hot flashes or night sweats
- ☐ Sleep disruption
- ☐ Mood changes
- ☐ Vaginal dryness
- ☐ Low libido
- ☐ Fatigue that feels hormonal
- ☐ Bone or joint pain

Menstrual / Cycle Log

Cycle Start	Length	Flow (L/M/H)	Symptoms	Notes

Questions to Ask My Provider

- ☐ Should I see an endocrinologist or reproductive endocrinologist?

- ☐ What do my hormone labs show over time?
- ☐ Is what I'm on replacement, or is it contraception?
- ☐ Does my plan protect bone and cardiovascular health, not only prevent hot flashes?

I've Got This

Scan and skip. The whole section in one column.

- POI is common in CG, do not wait until symptoms are severe to ask.
- Baseline FSH, estradiol, and AMH levels are worth getting early.
- Track cycle and symptoms; patterns show up across months, not weeks.
- HRT is part of this conversation, not a separate topic.
- Endocrine or reproductive endocrinology referral is worth having on the team.

Hormone Replacement Therapy (HRT)

Matched to handbook section: Hormone Replacement Therapy

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Write down the exact name and dose of the HRT you are on, or being offered.
- Ask: is this true replacement, or a contraceptive being used as a substitute?
- Put the next reassessment on your calendar before you leave the appointment.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Treatment-Plan Clarifier

Fill in with your provider. The goal is to know which tool you are being offered and why.

Question	Answer

Key Questions

- ☐ Initiation timing discussed, at physiologic age of puberty if POI is evident, or at onset of secondary amenorrhea
- ☐ Is this true hormone replacement, or a contraceptive being used as a substitute?
- ☐ What is the long-term plan? Bone protection? Cardiovascular? Cycle control?
- ☐ What dose am I on and why?
- ☐ What chemical form of estrogen am I on? They may not all protect bone health to the same extent.
- ☐ When will we reassess?
- ☐ If insurance denies, what are the alternatives?

My HRT Baseline (before starting)

DEXA scan date / result: _____

Estradiol level: _____

FSH: _____

Other baseline labs: _____

Current symptoms I want HRT to address:

_____

Reassessment Tracker

Date	Dose	Symptom Change	Labs	Next Step

Replacement is not the same as contraception. Both are valid tools, know which one you are receiving and sometimes different chemical forms of estrogen.

I've Got This

Scan and skip. The whole section in one column.

- HRT and contraception are different tools with different doses. Know which you have.
- Initiation timing: physiologic age of puberty if POI evident, or at onset of secondary amenorrhea.
- Baseline: DEXA, estradiol, FSH, current symptoms written down.
- Reassess regularly, dose and formulation are not set for life.
- If insurance denies a specific formulation, a letter of medical necessity usually works.

Fertility, Infertility, and Family-Building

Matched to handbook section: Fertility, Infertility, and Family-Building Options

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Write down where you are today, planning, exploring, unsure, or not wanting children. All four are valid.
- If you are under 35 and think you might want children, ask about fertility preservation options.
- Today's answer does not have to be forever.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Where I Am Today

- ☐ I want to know my baseline (regardless of plans)
- ☐ I want to explore pathways to having children
- ☐ I do not want biological children, I want information for health reasons only
- ☐ I am not ready to make any decision today

Baseline I Can Request

- ☐ AMH (Anti-Müllerian Hormone)
- ☐ FSH and estradiol
- ☐ Antral follicle count (ultrasound)
- ☐ DEXA scan
- ☐ Cardiovascular baseline

These tests are mine to request, whether or not I want children.

Options Map, Check what you want to learn more about

- ☐ Timed natural conception with monitoring
- ☐ Fertility preservation (egg or embryo freezing)
- ☐ IVF with my own eggs
- ☐ IVF with donor eggs
- ☐ Donor embryos
- ☐ Gestational carrier / surrogacy
- ☐ Adoption (domestic, international, foster-to-adopt, kinship)
- ☐ Co-parenting or chosen family arrangements

- ☐ Choosing not to pursue parenthood

The Cost Question

- ☐ Employer fertility benefits checked
- ☐ State-mandated coverage reviewed
- ☐ Clinical trials / research studies explored
- ☐ Grants or foundation programs identified
- ☐ HSA/FSA eligibility confirmed

Realistic total cost estimate: _____

Language I Can Use

- "Family planning is not my focus right now. I am here for my overall health."
- "I would like to understand my baseline, but I am not making decisions today."
- "Please don't assume I want to pursue fertility treatment. I want information, not a recommendation."
- "I've decided not to pursue pregnancy. Please keep that out of this visit."
- "This topic is heavy. Can we pace this conversation?"

Every path is valid, including the choice to step away from this conversation.

I've Got This

Scan and skip. The whole section in one column.

- POI shrinks the window, an early conversation preserves more options.
- Pathways: spontaneous, IVF with own eggs, donor egg, surrogacy, adoption, child-free. All valid.
- Reproductive endocrinology referral is the on-ramp to most of these.
- International guidelines recommend fertility counseling for CG adults, ask for it.
- You are allowed to say "I want information, not a recommendation."

Pregnancy, Prenatal Care, and Breastfeeding

Matched to handbook section: Pregnancy, Prenatal Care, and Breastfeeding

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Make sure your OB and metabolic team are talking to each other.
- Keep your galactose-restricted diet throughout pregnancy and breastfeeding.
- Write down your postpartum support plan, one line, one person is a start.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Preconception Checklist

- ☐ Reproductive endocrinology consultation scheduled
- ☐ Hormone therapy review completed (do not stop on own)
- ☐ Genetic testing options for galactosemia & other conditions discussed
- ☐ DEXA baseline obtained
- ☐ Medication review for pregnancy safety
- ☐ Baseline labs drawn (thyroid, vitamin D, B12, iron, hormones)
- ☐ Dietitian familiar with galactose-restricted diet identified

Pregnancy Care Team Builder

Role	Provider Name	Contact	First Visit Date

Roles to fill: primary care, metabolic specialist, OB/GYN (with MFM access), reproductive endocrinologist (if relevant), dietitian, mental health provider, dentist.

Diet During Pregnancy, Nutrition Check

- ☐ Calcium plan in place (dairy-free sources identified)
- ☐ Vitamin D level checked
- ☐ Iron, B12, iodine, choline, DHA reviewed
- ☐ Prenatal vitamin confirmed galactose-free & lactose-free
- ☐ Medical foods reviewed with dietitian if used

Trimester Monitoring Log

Trimester	Labs	Bone Health	Mental Health	Fatigue Plan

Breastfeeding Plan / Supplementation (if chosen)

- ☐ Hydration and calorie plan with dietitian
- ☐ Calcium and vitamin D continued
- ☐ Rest pacing plan with partner / family
- ☐ Backup plan (pumping, combo feeding, formula) written down, no judgment

Postpartum Recovery Checklist

- ☐ Mental health check-in with provider within 6 weeks
- ☐ Hormone therapy adjustment discussion scheduled
- ☐ Repeat DEXA scheduled after weaning
- ☐ Metabolic specialist kept in loop
- ☐ Extended recovery time built into work / life plan

Language I Can Use if the Healthcare Provider Seems to Misunderstand My Goals

- "I'd like provider-to-provider communication between my OB and metabolic team."
- "Please use galactose-free and lactose-free medication formulations when clinically appropriate."
- "Fatigue and cognitive processing differences are part of my baseline."
- "I'd like to continue my galactose-restricted diet throughout pregnancy and breastfeeding."

The goal is not perfection. It is informed, coordinated care with a team that listens.

I've Got This

Scan and skip. The whole section in one column.

- OB and metabolic coordination is non-negotiable during pregnancy.
- Galactose-restricted diet continues. Hydration and calories go up.
- Galactose-free / lactose-free medication formulations whenever possible.
- If newborn status is unknown, breastfeeding is withheld until screening confirms (can pump & freeze milk).
- Postpartum: DEXA, HRT adjustment as necessary, mental health, all on the 6-week radar.

Bone Health and DEXA

Matched to handbook section: Bone Health

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Ask when your next DEXA is due. If you do not know, that is the answer you need.
- Take today's calcium and vitamin D, even one dose counts.
- Write down the current names and doses of your supplements.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Bone Health Baseline

First DEXA date: _____

T-score / Z-score: _____

Vitamin D (25-OH) level: _____

Calcium intake (diet + supplement): _____

Hormone status at time of scan: _____

DEXA Tracker

Date	Site	T/Z Score	Change	Next Scan

Supplement Tracker

- ☐ Calcium citrate (preferred), dose: _____
- ☐ Vitamin D, dose: _____
- ☐ Vitamin K, dose: _____
- ☐ Magnesium citrate, dose: _____
- ☐ Other: _____

Questions to Bring

- ☐ When should my next DEXA be?
- ☐ Is my current vitamin D level protective?
- ☐ Does my HRT / hormone plan support bone protection?
- ☐ Am I on the right form of calcium for my gut?

The best supplement is the one you take consistently.

I've Got This

Scan and skip. The whole section in one column.

- CG affects bone mineral density earlier than the general population.
- Baseline DEXA in the 20s; repeat every 1-2 years depending on T/Z score.
- Calcium citrate is usually better tolerated than carbonate.
- Vitamin D 25-OH is the number to watch, ask for it by name.
- HRT, exercise, and weight-bearing activity all contribute to bone protection.

Tremor

Matched to handbook section: Tremor

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Do one spiral test today and date it.
- If tremor is new or worsening, say those exact words at your next appointment.
- Write down one daily-life task the tremor is interfering with.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Archimedes Spiral Test: Self-Assessment Tool

Disclaimer

This Archimedes Spiral Test sheet is designed for adult use only. It is provided as a personal self-assessment tool to help track tremors or fine motor changes.

- It is not a diagnostic test, nor a measure of disease escalation.
- It does not replace a medical evaluation by a qualified healthcare professional.
- Subtle neurological symptoms can be missed by visual inspection alone, and only a trained professional can provide a full evaluation.

You may use this tool once, occasionally, or yearly as a way to track changes for yourself. If you notice any significant changes or have concerns, please contact your doctor.

The Archimedes Spiral Test is widely used by neurologists to evaluate tremors in conditions such as Parkinson's disease, essential tremor, dystonia, and other movement disorders. You are encouraged to bring your completed spirals to your doctor if you are concerned, they may use it as part of a neurological assessment and advocating for help.

Instructions: Use a pen or pencil.

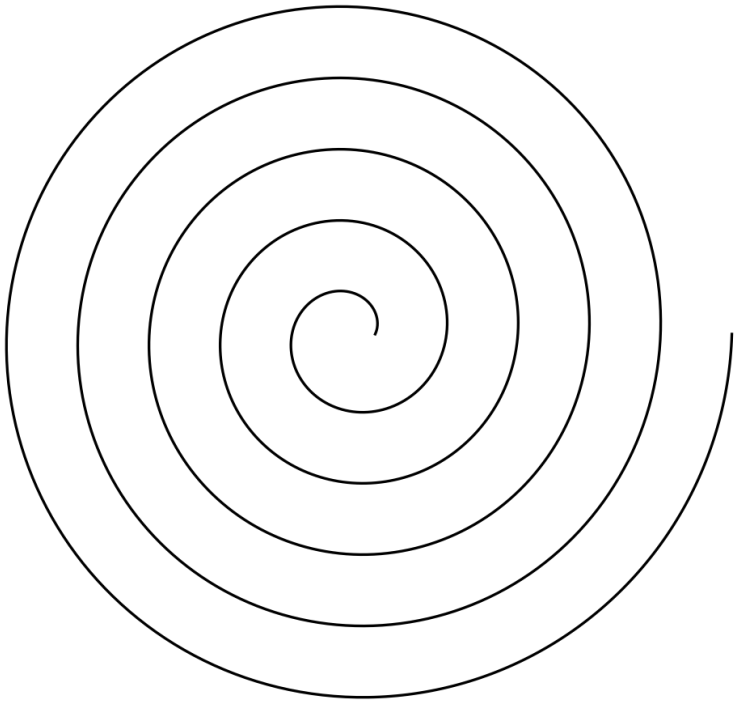
- Place the paper on a hard, flat surface like a table or desk.
- Trace over the spiral below in one smooth motion.
- Perform the test separately with your right and left hands.
- Try again for a second attempt if desired.

This version uses ~6 mm spacing between turns and 5 full turns, which is standard in many clinical protocols.

For personal use only. Not a substitute for medical advice. Many neurological conditions use spiral testing; bring to your doctor if concerned.

Right Hand Spiral Test

Trace over the spiral in one smooth motion, from the outside in or the center out, whichever feels natural.



Date: _____

Time: _____

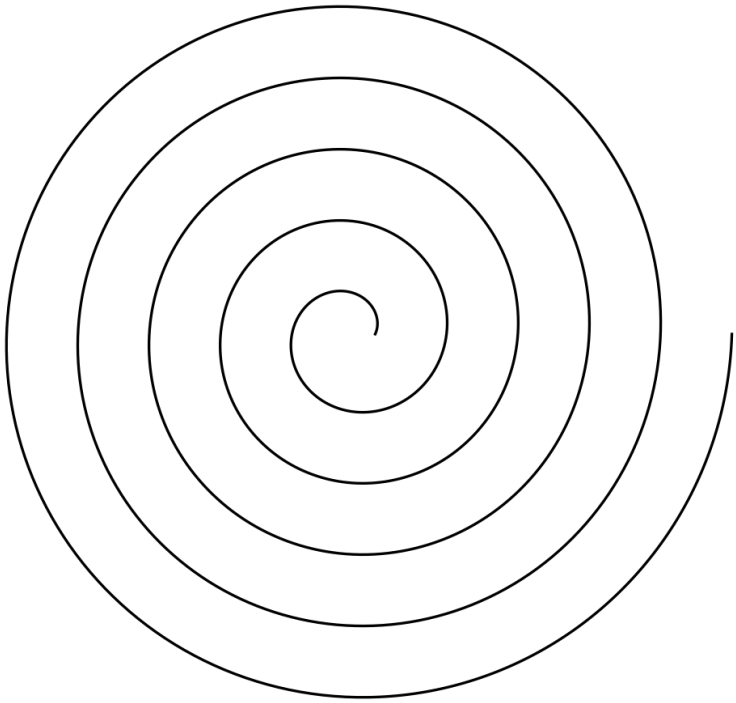
Fatigue level (1-5): _____

Medications taken today: _____

Notes: _____

Left Hand Spiral Test

Trace over the spiral in one smooth motion, from the outside in or the center out, whichever feels natural.



Date: _____

Time: _____

Fatigue level (1-5): _____

Medications taken today: _____

Notes: _____

Symptom Change Tracker

Date	What Changed	Trigger	Daily Impact

Language for Appointments

- "This tremor is new / worsening."
- "This is interfering with daily tasks."
- "This represents a change from baseline."
- "Anxiety may worsen symptoms, but this tremor persists outside anxious situations."

Speech and Swallowing

Speech and motor-speech impairments are common in adults with CG. A speech-language pathologist (SLP) can help with clarity, word-finding, and swallowing.

- ☐ I have noticed changes in speech clarity or word-finding
- ☐ I have asked about a referral to a speech-language pathologist
- ☐ I have a current SLP or know where to get one

SLP name / clinic: _____

I've Got This

Scan and skip. The whole section in one column.

- Spiral test: 6 mm between turns, 5 full turns, repeat both hands yearly.
- Anxiety can amplify tremor but not fully explain it, distinguish in your notes.
- SLP is the right referral for speech clarity, word-finding, and swallowing changes.
- Track daily-life impact, not just the tremor itself.
- "New" and "change from baseline" are the two phrases doctors act on fastest.

Fall Risk and Home Safety

Matched to handbook section: Fall Risk As We Get Older

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Walk through your home and fix one trip hazard today.
- If you have had a near-fall in the last 6 months, mention it at your next visit.
- Write down what you were doing when your balance felt off.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Balance and Mobility Check

- ☐ Any near-falls in the last 6 months
- ☐ Any actual falls in the last year
- ☐ New fear of falling
- ☐ Dizziness, unsteadiness, or vertigo episodes
- ☐ Noticed change in gait or stride

Home Safety Checklist

- ☐ Loose rugs removed or secured
- ☐ Clear paths in hallways and at night
- ☐ Grab bars in bathroom (or planned)
- ☐ Non-slip mats in bath / shower
- ☐ Good lighting on stairs and night routes
- ☐ Supportive footwear indoors
- ☐ Phone reachable if down

What to Ask Provider

- ☐ Gait and balance assessment
- ☐ Vision check for depth / contrast changes
- ☐ Medication review for dizziness or sedation
- ☐ Physical therapy referral for strength / balance work

A single fall or near-fall is worth naming. Prevention is not restriction, it is stability.

I've Got This

Scan and skip. The whole section in one column.

- Neurologic + bone mineral density = real fall risk for CG adults, even under 60.
- Common home fixes: rugs secured, stair lighting, bathroom grab bars, cord management.
- PT referral is worth asking for if balance feels off or falls are happening.
- Report every near-fall, they are data, not embarrassment.
- A single cane or trekking pole is not a downgrade. It is an upgrade.

Men's Health

Matched to handbook section: Men's Health and Galactosemia

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Book one yearly primary care visit and bring your full CG history written down.
- Ask your PCP about bone health screening, DEXA applies to you too.
- Write down your baseline symptoms today so future changes are visible.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

What I Notice In My Body

Check any that apply. Patterns matter, even with normal labs.

- ☐ Ongoing fatigue or low stamina
- ☐ Tremor, coordination, or muscle issues
- ☐ Brain fog or slowed thinking
- ☐ Mood, energy, or libido changes
- ☐ Bone or joint pain, prior fractures
- ☐ Palpitations, dizziness, heat intolerance

Conversation Starter

What feels harder than it used to: _____

What takes longer to recover from: _____

What affects daily life or work: _____

What to Ask

- ☐ Annual primary care with my full history
- ☐ Bone health addressed in routine screening
- ☐ Hormonal labs if symptoms are present
- ☐ Referral to endocrine / neuro if new symptoms persist

Lack of data is not lack of risk. Your experience matters, even before studies catch up.

I've Got This

Scan and skip. The whole section in one column.

- CG research has skewed female, same CG follow-up still applies to men.
- DEXA, neuropsych, mental health, metabolic visits are not gendered.
- Ask for hormonal labs when symptoms are present; men get POI-adjacent patterns too.
- Bring your full history written down, most PCPs have not seen adult CG.
- Lack of data is not lack of risk. Advocate.

Rare but Serious Events

Matched to handbook section: Rare Occurrences

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- Keep the ER Passport Card in your wallet (see next section).
- If a new neurological event happens, go to the ER and use the Event Summary worksheet.
- Practice saying your pivot line out loud before you need it.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

ER Pivot Card, Seizures

Say clearly: "This is a first or changed seizure in a patient with a metabolic disorder. We need a safety plan before discharge."

- ☐ Ask for rescue medication (medication you can take at home to abort or shorten an attack) before discharge
- ☐ Ask for clear seizure precautions in writing
- ☐ Urgent neurology referral
- ☐ Guidance on nasal vs. rectal rescue meds

Safe Discharge Check

- ☐ Discharge paperwork matches verbal instructions
- ☐ Return precautions in writing
- ☐ Follow-up appointments and timelines listed
- ☐ Prescriptions present and understood
- ☐ After-hours plan stated

Cognitive / Behavioral Change Pivot

Use neutral language: "This represents a change from baseline function." Ask for infection check, electrolytes, liver/kidney labs, and neurologic assessment if symptoms persist.

Event Summary Worksheet

Date and time: _____

What happened (neutral description): _____

How long it lasted: _____
Recovery time and symptoms after: _____
Who was with me: _____
What I asked for at discharge: _____
Follow-up dates: _____

Rising Labs Tracker

Date	Test	Result	Prior Baseline	Action

I've Got This

Scan and skip. The whole section in one column.

- Seizure pivot: "first or changed seizure in a patient with a metabolic disorder."
- Cognitive or behavioral pivot: "this represents a change from baseline function."
- Safe discharge: written paperwork must match what you were told verbally.
- Track rising labs, they are the warning signal, not the emergency.
- Carry the ER Passport Card so you do not have to explain in a crisis.

Emergency Room (ER) Passport Card

A wallet-sized card you can print, fold, and carry. If you arrive at an ER unable to speak for yourself, or with a provider who has never seen Classic Galactosemia, this card tells them what matters in the first thirty seconds.

How to Use It

- Print this page at 100% scale (do not 'fit to page', that will shrink the card).
- Cut around the outer dashed border.
- Fold in half along the middle line so the blue 'I Have Galactosemia' side is on the front.
- Fill in the blanks in pen. Update yearly and any time your emergency contact, insurance, or allergies change.
- Laminate if you can, or slide it into a clear ID sleeve. Keep it in your wallet behind your driver's license or insurance card.
- Consider making a second copy for a partner, parent, or roommate.
- Pair this with a medical ID bracelet, phone lock-screen medical info, or Apple Health / Google Health Medical ID.

What to Say When You Hand It Over

"I have Classic Galactosemia. It's a rare metabolic disorder most providers haven't seen. This card has the basics. Please take a moment to read it as soon as possible."

Your Card

Galactosemia Foundation

I HAVE GALACTOSEMIA

Name: _____
Date of Birth: _____
Allergies / Intolerances: _____
Insurance ID: _____
Emergency Contact Name: _____
Phone: _____
Metabolic Provider: _____

Adult with Galactosemia, ultra-rare lifelong metabolic disorder. Please allow time and assess change from baseline.

---------- fold here ----------

GALACTOSEMIA

METABOLIC ALERT, *Not an allergy. Dietary control alone does not eliminate risk. Lactose/galactose fillers may be present and not always flagged on labels. Please assess risk vs. benefit and consider alternatives.*

This patient lives with a condition you may be seeing for the first time. Please assess change from baseline, and partner with them.

- Lifelong metabolic disorder (not pediatric-limited)
- Stress or illness may amplify symptoms
- Tremor, slowed speech, or processing may be baseline
- Please allow time to communicate, do not interrupt
- Multiple findings may be chronic

Thank you for caring for me.

You do not have to explain your whole medical history in an emergency. This card does the first 30 seconds for you.

Barriers to Care, Insurance

Matched to handbook section: Barriers to Care

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- If a denial letter arrived: circle the appeal deadline. That is the only date that matters today.
- If a visit or test needs approval: ask the office to start the prior authorization. You do not have to fight the insurer yourself.
- If a bill or EOB (Explanation of Benefits) is confusing: call the number on the back of your insurance card and say, "Please walk me through this line by line."
- If you lost coverage or had a life change: you have a 60-day Special Enrollment window. Start the Marketplace application today.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Insurance Glossary Quick Reference

You do not need to memorize these, just recognize them when your plan, HR, or a bill uses them.

Premium	The monthly amount you pay to keep your insurance active, whether or not you use care that month.
Deductible	The amount you pay out of pocket before insurance starts sharing costs. Resets each plan year.
Copay	A fixed dollar amount you pay for a visit, prescription, or service (e.g., $30 per PCP visit).
Coinsurance	A percentage of the cost you pay after meeting your deductible (e.g., plan pays 80%, you pay 20%).
Out-of-Pocket Max	The most you will pay in a plan year for covered services. After this, insurance pays 100%.
In-Network	Providers and facilities that have a contract with your insurance, lower cost to you.
Out-of-Network	Providers without a contract with your plan, usually much higher cost or not covered at all.
Formulary	The list of prescription drugs your plan covers, grouped into tiers that determine your copay.
Prior Authorization	Approval your plan requires before it will pay for certain drugs, tests, or procedures. Often needed for HRT, DEXA, specialty medications.
Step Therapy	A rule that says you must try a cheaper drug first before the plan will cover a pricier one. Can be appealed if medically inappropriate.

EOB (Explanation of Benefits)	A statement from your insurer showing what was billed, what they paid, and what you owe. Not a bill.
Claim	A request for payment that your provider (or you) submits to your insurance company.
Denial	When insurance refuses to pay for a service. Always check the reason, most denials can be appealed.
Appeal	A formal request to reverse a denial. You have a legal right to appeal, usually within 60-180 days.
Medical Necessity	The standard insurance uses to decide if a service is covered. Your metabolic team can write letters supporting it.
Specialty Pharmacy	A pharmacy that handles complex or high-cost drugs (some HRT, infusions). Your plan may require you to use one.
SPD (Summary Plan Description)	The full written document describing your employer plan's benefits, rules, and appeal rights. HR must give it to you on request.
COBRA	A law that lets you keep your employer health plan (at full cost) for up to 18-36 months after leaving a job.
Qualifying Life Event	A life change (marriage, job loss, aging off a parent's plan at 26, birth of child) that opens a special enrollment window outside of open enrollment.
HSA / FSA	Tax-advantaged accounts you can use for medical costs. HSA requires a high-deductible plan; FSA is employer-linked and usually use-it-or-lose-it.
Prior Year Deductible Carryover	Some plans count late-year costs toward next year's deductible. Worth asking about if you hit care late in the year.

Terms to Personalize for My Plan

Term	What It Is on My Plan

Examples: my premium $____, my deductible $____, my OOP (out-of-pocket) max $____, my formulary tier for HRT, my plan's PA (prior authorization) phone line, my appeals address.

Referrals, Scripts, and Prior Authorizations Tracker

Many of the things an adult with Classic Galactosemia needs, DEXA scans, HRT, specialty labs, neuropsych evaluations, speech-language therapy, genetic counseling, require either a referral from your PCP, a written script (order) from a provider, or prior authorization from your insurance. Use this tracker to keep them from falling through the cracks.

What typically needs what

- DEXA scan, usually needs a written order from PCP or endocrinology; sometimes needs prior auth if under age 50.
- HRT / estrogen, needs a prescription; the specific formulation often requires prior authorization.
- Specialty labs (FSH, AMH, estradiol, vitamin D, bone markers), need a lab order; some plans require PA.
- Neuropsychological evaluation, needs a referral; almost always requires prior authorization.
- Speech-language therapy, needs a referral; PA often required for adult SLP services.
- Metabolic specialist (adult), needs a referral from PCP on most HMO plans.
- Genetic counseling, needs a referral; may require PA.
- Physical or occupational therapy (for tremor, balance, falls), needs referral; PA common after initial visits.
- Cataract surgery, needs ophthalmology referral and PA.
- MRI / CT / advanced imaging, needs order and almost always PA.

Tracker

Date Requested	What (Referral / Script / PA)	Provider Who Ordered	Status	Auth # or Ref #	Expires

Status shorthand you can use

- Requested, asked provider but not yet sent
- Sent, order or PA submitted to insurance
- Approved, PA approved, good through date listed
- Denied, see appeal tracker below
- Scheduled, appointment or scan booked
- Complete, done and results received

Language I Can Use

Asking my PCP for a referral or script

- "I have Classic Galactosemia. International guidelines recommend I have a baseline DEXA scan and annual metabolic follow-up. Can you place the order today?"

- "I'd like a referral to an adult metabolic specialist, endocrinology, and OB/GYN (or reproductive endocrinology) if not already in place."
- "Can you write a letter of medical necessity for this? I can share language from the clinical guidelines if helpful."

Calling my insurance about prior authorization

- "I'm calling to check the prior authorization status for [service / medication]. My member ID is _____. The ordering provider is _____."
- "What documentation do you need from my provider to approve this?"
- "What is the turnaround time, and how will I be notified of the decision?"
- "If this is denied, what is the appeal process and timeline?"

Appeal Tracker (for Denials)

Date Denied	What Was Denied	Denial Reason	Appeal Filed On	Supporting Docs Sent	Outcome

A first denial is not the end. Many CG-relevant services (HRT formulations, DEXA before 50, neuropsych) are approved on appeal once a letter of medical necessity cites the international clinical guidelines.

Employer Insurance

- ☐ Current plan name and ID
- ☐ Asked HR for Summary Plan Description (SPD)
- ☐ Know open enrollment dates
- ☐ Know what coverage changes with job changes

Marketplace Insurance (ACA)

- ☐ Know my state's marketplace website
- ☐ Know annual open enrollment window
- ☐ Know which qualifying life events trigger special enrollment
- ☐ Subsidy eligibility checked

Medicaid / Medicare

- ☐ Medicaid eligibility checked in my state

- ☐ Medicare age / disability pathway understood
- ☐ Dual-eligible status reviewed if applicable

Dental and Vision Add-Ons

- ☐ Dental plan in place
- ☐ Vision plan in place
- ☐ Standalone vs. bundled compared

I've Got This

Scan and skip. The whole section in one column.

- Know the big three: deductible, out-of-pocket max, formulary.
- HRT, DEXA, neuropsych, and specialist labs often need prior authorization.
- First denials are common and often reversed on appeal, the letter includes your deadline.
- Track claims and PAs in writing, not email threads.
- State insurance commissioner and ombudsman are free escalation paths.
- Life change = 60-day Special Enrollment window on the Marketplace.

Disability Benefits, SSI and SSDI

Matched to handbook section: Disability Benefits

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- If you are considering applying: ask your metabolic specialist for a letter describing diagnosis, functional limits, and prognosis. That letter is the backbone of everything else.
- If a denial letter arrived: note the appeal deadline. Most approvals happen at reconsideration or hearing, not on the first try.
- If overwhelmed: call 211 or a local disability advocate. You do not have to navigate SSA alone.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

SSI vs. SSDI At a Glance

	SSI	SSDI

Fill in with a benefits counselor: based on, work history required, income rules, health insurance link, typical benefit size.

SSDI Medical Documentation Checklist

- ☐ Confirmed diagnosis (GALT deficiency) from metabolic specialist
- ☐ Objective clinical findings (labs, imaging, DEXA)
- ☐ Functional limitations documented (fatigue, cognitive, tremor, POI, etc.)
- ☐ Treatment burden described (appointments, monitoring, restrictions)
- ☐ Prognosis statement from specialist

Step-by-Step Application Tracker

- ☐ Step 1, Confirm coverage type
- ☐ Step 2, Understand plan's disability definition
- ☐ Step 3, Gather medical documentation
- ☐ Step 4, Complete forms carefully; keep copies

- ☐ Step 5, Prepare for follow-up / additional requests
- ☐ Step 6, If approved, track payment start and review dates
- ☐ Step 7, If denied, request reason and start appeal

If Denied

Denial is information. Many approvals happen at appeal or hearing.

- ☐ Read denial letter carefully and underline reason
- ☐ Request reconsideration within the stated window
- ☐ Gather additional documentation the reviewer said was missing
- ☐ Consult a disability attorney or advocate if possible (often paid only on approval)

Denial is not failure. It is a starting point for appeal.

I've Got This

Scan and skip. The whole section in one column.

- SSI is needs-based; SSDI depends on work history. Many CG adults qualify for one or both.
- Specialist letter > generic primary care letter. Ask the metabolic clinic.
- Document functional limits (fatigue, cognitive load, tremor, POI), not just the diagnosis.
- First denials are common. Appeal deadlines are short, circle them immediately.
- Disability attorneys are usually paid only if you win.

Career Growth, Reviews, and Advancement

Matched to handbook section: Career Growth

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- If work feels unsustainable today: write down one accommodation that would help (quieter space, flex hours, remote day, lighter cognitive load). That is your starting ask.
- If a review is coming: list three wins from the past 6 months. That is your draft.
- If you are masking to survive: note it. Masking is labor. It belongs in your energy budget.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

My Role Snapshot

Role / title: _____

Review period: _____

Date of next review: _____

Contribution Tracking

List observable work, not effort or intent.

Contribution	What I Did	Outcome / Impact

Raise / Advancement Prep

- ☐ Contributions documented across the full year, not just recent months
- ☐ Outcomes separated from hours or effort
- ☐ Evidence I can point to (specific examples)
- ☐ Accommodations request written down if needed

Re-Entry Into Work

What I can do now: _____

Conditions that help me do my best work:

_____

Realistic pace for long-term stability: _____

Language You Can Use

- "I'd like to revisit compensation given my contributions this year."
- "I work best with [specific condition]. Would that be possible?"
- "I'd like to separate my contributions from the timeline conversation."

Career growth does not have to be linear to be real. Sustainability matters.

I've Got This

Scan and skip. The whole section in one column.

- Document outcomes, not hours. Impact is what reviews actually reward.
- Accommodations are a negotiation tool, not a confession.
- Three concrete wins > one long narrative.
- Sustainability is a career strategy, not a compromise.
- Non-linear trajectories are still real careers.

Research and Clinical Trial Participation

A place to track studies, researchers, protocols, and investigational drugs

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- If you are in a study right now: write down the study name, the PI, and one phone number or email to reach them. That is enough for today.
- If you are on an investigational drug: keep the drug name, dose, and last dose date somewhere you can find it in an ER.
- If you are thinking about joining a study: you are allowed to ask questions and you are allowed to say no at any time.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

Classic Galactosemia is rare, which means much of what is known about adult CG comes from adults like you choosing to participate in research. Whether you are in a registry, an observational study, a clinical trial, or taking an investigational drug, this section helps you keep it all in one place, and keeps you in the driver's seat.

Types of Research You Might Be Part Of

- Patient registry, a long-term database of people with CG (e.g., the Galactosemia Foundation registry, GalNet, institutional registries).
- Natural history study, tracks how CG changes over time without testing a treatment.
- Observational study, researchers observe a specific outcome (bone, cognition, fertility) without intervening.
- Survey or interview study, one-time or periodic questionnaires about symptoms, quality of life, or experience.
- Clinical trial, tests a specific intervention (drug, device, dietary change). Usually phased I-IV.
- Biobank, contributes a blood, urine, or tissue sample for future research.
- Genetic / genomic study, sequences your genes or variants for research use.

Before I Join, Questions to Ask

- ☐ What is the purpose of this study and what is it hoping to show?
- ☐ Who is running it (institution, PI, sponsor, funder)?
- ☐ Is this peer-reviewed and IRB / ethics-approved? What IRB?
- ☐ What exactly will I be asked to do? (visits, labs, scans, surveys, drugs)
- ☐ How long is the commitment?
- ☐ Are there costs to me? Is travel covered? Is there compensation?

- ☐ What are the known risks and side effects?
- ☐ Can I withdraw at any time, and what happens if I do?
- ☐ Who owns my data and samples? Can they be shared or sold?
- ☐ Will I get my individual results back, or only aggregate findings?
- ☐ Who is my point of contact if something changes with my health?
- ☐ Will my insurance be billed for any procedures?

Studies I Am Currently Participating In

Study / Registry Name	Institution	PI or Lead Researcher	My Role	Start Date	Expected End

Researcher and Group Contacts

Add anyone you want to stay in touch with, principal investigators, coordinators, patient advocates, foundation staff.

Name	Role	Institution / Group	Email	Phone

Protocol Summary Worksheet

Fill one of these out for each active study. You should have this in writing from the study team, if not, ask.

Study title: _____

Protocol number / ClinicalTrials.gov ID (NCT#):

_____

Sponsor / funder: _____

IRB of record: _____

Principal Investigator: _____

Site coordinator name and direct contact:

_____

What I am asked to do (in plain language):

_____

_____

Visit schedule (how often, where): _____

_____

Labs, scans, or procedures involved: _____

_____

Known risks / side effects: _____

_____

What I agreed to share (data, samples, tissue, genetic info):

_____

_____

How I can withdraw and what happens to my data if I do:

_____

_____

Next visit date: _____

Investigational Drug Tracker

If you are on an investigational drug, a drug being studied that is not yet FDA-approved for your condition, track it here. Bring this to every non-study medical appointment, including the ER.

Drug Name / Code	Dose	Schedule	Start Date	Stop Date	Side Effects Noticed

Important if you are on an investigational drug

- ☐ I carry a wallet card or phone note listing the investigational drug and study contact
- ☐ My PCP has been told I am on an investigational drug
- ☐ I know who to call 24/7 if I have a side effect or concern
- ☐ I have a written plan for what to do if I end up in the ER
- ☐ I know what other medications must be avoided while on the investigational drug

24/7 study emergency contact: _____

Visit Log

A running record of study visits, useful for yourself, for taxes/travel reimbursement, and if you ever switch sites.

Date	Study	Visit Type	What Was Done	Follow-Up Needed

My Rights as a Research Participant

- You can ask any question at any time, and you deserve a plain-language answer.
- You can withdraw from a study at any time, for any reason, without affecting your regular medical care.
- You have the right to a copy of the consent form you signed.
- You have the right to know who is paying for the research and who benefits financially.
- You have the right to be told if new information changes the risks.
- You have the right to be treated with respect, by the PI, the coordinator, and every member of the team.

Language I Can Use

- "Can you explain this in plain language, what is the study asking of me?"
- "I'd like a copy of the protocol summary and the consent form before I decide."
- "What happens to my samples if I withdraw?"
- "Will my primary care team and metabolic team be notified of my participation?"
- "I'm considering stepping back from this study, what does that process look like?"

Participating in research is a gift. It is also a choice, not a debt. You are allowed to say yes, no, pause, or leave, on your terms.

I've Got This

Scan and skip. The whole section in one column.

- Track one study per row: name, PI, institution, contact, role.
- Investigational drug? Keep name, dose, and last-dose date ER-accessible.
- Consent is ongoing, you can pause or withdraw without penalty.
- Ask what happens to your samples and data if you leave.
- Loop your metabolic team in on trial participation so records stay unified.

Mental Health and Social Health

Matched to handbook section: Mental Health and Social Health

Just Tell Me

If today is a hard day, do one of these. Close the book. It counts.

- If today feels heavy: text HOME to 741741, or call/text 988. You do not have to be in crisis to use them.
- If you are masking more than engaging: write down one person who does not need you to explain. Text them. That counts as treatment.
- If nothing feels possible: rest is part of treatment, not a reward. Close the book.

☐ **I did one thing. That is enough for today.**

Walk Me Through It

Step-by-step, in plain language. Stop whenever you need to.

What I Am Experiencing Right Now

- ☐ Sleep disrupted for more than 2 weeks
- ☐ Mood shifted persistently, not passing
- ☐ Withdrawing from people I usually feel close to
- ☐ Things that used to feel manageable feel heavy
- ☐ Spending more energy masking than engaging
- ☐ Anxiety driving decisions (avoidance, shrinking life)
- ☐ Intrusive thoughts, hopelessness, or self-harm thoughts
- ☐ Substance use / eating / screen use becoming a coping pattern
- ☐ Feeling emotionally flat or disconnected from myself

Any ONE of these is enough of a reason to reach for support. You do not need to qualify.

Pattern Awareness

Symptoms increase when: _____

What helps even slightly: _____

What makes it worse: _____

Build My Mental Health Toolbox

Clinical Options

- ☐ Therapy (chronic illness / rare disease / grief / neurodivergence)
- ☐ Psychiatry consultation
- ☐ Neuropsychological evaluation

- ☐ Occupational therapy (adult)
- ☐ Speech-language therapy (adult)

Community and Peer

- ☐ Galactosemia Foundation peer community
- ☐ Infertility / grief / chronic illness support groups
- ☐ Couples or partnered counseling

Body-Based Foundations

- ☐ Gentle movement
- ☐ Sleep hygiene
- ☐ Time outdoors
- ☐ Breath / grounding practices

Social Health Anchors

One or two anchor relationships: _____

One person who does not need me to explain:

_____

My script for low-energy weeks: _____

My script for dietary visibility at social events:

_____

Gentle Affirmations

- My mental health is part of my medical health, not a side topic.
- Fatigue is not laziness. It is a physical symptom.
- Pulling back is not failure. I can return to connection at my own pace.
- Needing support means I am paying attention, not that I am weak.
- Being understood by one person is medicine.
- Rest is part of treatment, not a reward.
- I was never supposed to do this alone.

Urgent Help

In the U.S., call or text 988 for the Suicide and Crisis Lifeline. Text HOME to 741741 for the Crisis Text Line. Call 911 or go to the nearest ER if you are in immediate danger. Call 211 for local mental health and social services.

You do not need to be "sick enough" to use these. They exist for you.

I've Got This

Scan and skip. The whole section in one column.

- One symptom for two weeks is enough reason to reach for support. You don't have to

qualify.

- Masking is labor. Budget for it like any other energy cost.
- CG-aware therapy, psychiatry, and neuropsych are all reasonable asks, not overkill.
- Peer community (GF) is medicine too, not a substitute for clinical care.
- 988 (call or text), 741741 (text HOME), 911, and 211 are your crisis menu.

Appendix A, Letter Templates

Two ready-to-use letters you can print, fill in, and hand to a new provider

These are fill-in templates. Bracketed text like [Patient Name] is meant to be replaced. You or your provider can edit on paper, in Word, or by printing a signed copy. Keep a signed copy in your records.

Letter 1, Pediatric to Adult Metabolic Handoff

From the pediatric metabolic team to the new adult provider. Ask your pediatric clinic to complete this on letterhead before your last pediatric visit.

Date: _____

To: [Receiving Adult Provider Name, Title]
Clinic / Institution: _____
Address: _____
Phone / Fax: _____

Re: [Patient Name], DOB [__/__/____], MRN [_____]

Dear Dr. [Receiving Provider],

I am writing to formally transition the care of [Patient Name] to your practice. [Patient Name] has been followed in our pediatric metabolic clinic since [age/year] for Classic Galactosemia (GALT deficiency, OMIM 230400), confirmed by [newborn screening / enzyme assay / genotype: _____].

Diagnosis and Genotype

Classic Galactosemia. Red blood cell GALT activity: _____. Genotype: _____ / _____.

Current Clinical Status

- Dietary status: lifelong galactose-restricted diet, adherence [good / variable / limited].
- Cognitive / developmental: _____.
- Speech / language: _____.
- Motor / tremor / ataxia: _____.
- Endocrine (POI in females / gonadal status in males): _____.
- Bone health / most recent DEXA (date, Z/T-score): _____.
- Mental health / cognitive supports: _____.
- Other active issues: _____.

Current Medications and Supplements

1. _____
2. _____
3. _____
4. _____

Most Recent Labs (date: __/__/____)

- Erythrocyte galactose-1-phosphate (Gal-1-P): _____ mg/dL
- Urinary galactitol: _____
- FSH / LH / estradiol (if applicable): _____ / _____ / _____
- 25-OH vitamin D: _____
- Calcium, phosphorus, alkaline phosphatase: _____ / _____ / _____
- TSH: _____
- LFTs: _____

Recommended Adult Monitoring (per Welling et al. 2017 international guideline)

- Annual metabolic visit: diet review, Gal-1-P, weight, BP, adherence.
- Endocrine: annual FSH/LH/estradiol in females; bone-health labs in all.
- DEXA every 2-5 years; more often if osteopenia/osteoporosis.
- Neurology referral if new/worsening tremor, ataxia, or movement symptoms.
- Speech-language evaluation as needed for adult dyspraxia or communication needs.
- Neuropsychological evaluation at transition and as clinically indicated.
- Mental health screening annually.

Red Flags Requiring Prompt Contact

- Rising Gal-1-P without dietary explanation.
- New or changed seizure activity.
- Acute change from cognitive or motor baseline.
- Pregnancy, coordinate with maternal-fetal medicine and metabolic team.

I have reviewed this transition with the patient and family. Records, imaging, and genetic testing can be released with the attached signed authorization. Please feel free to contact our team with any questions.

Sincerely,

_____

[Pediatric Metabolic Provider Name, Credentials]

[Title / Clinic]

Phone: _____ Fax: _____ Email: _____

Letter 2, Patient Introduction to a New Primary Care Provider

A letter you bring to a new PCP. Fill it in ahead of the visit so you do not have to explain Classic Galactosemia from scratch in the room. You can print it, email it, or upload it to the patient portal before the first visit.

Date: _____

To: Dr. _____ (Primary Care Provider)
Clinic: _____

From: _____
Date of birth: __/__/____ Preferred pronouns: _____
Preferred contact: _____

Dear Dr. _____,

Thank you for taking me on as a patient. I am writing this letter so you have the context you need before our first visit. I live with Classic Galactosemia (CG), a rare inherited metabolic condition. I have written this myself so we can spend our visit on what I actually need, not on explaining the diagnosis.

What Classic Galactosemia Is (in plain language)

Classic Galactosemia is a genetic condition where my body cannot properly process galactose, a sugar found in milk and many foods. I follow a lifelong galactose-restricted diet. Even with perfect diet, my body produces some galactose on its own, so long-term complications are part of the condition, not a sign that I have failed my diet.

Long-Term Issues That Can Affect Adults with CG

- Cognitive differences, slower processing, or executive-function challenges.
- Speech and language differences (adult dyspraxia in some).
- Tremor, ataxia, or other movement symptoms.
- Primary ovarian insufficiency (POI) in most females with CG.
- Low bone mineral density / early osteopenia or osteoporosis.
- Fatigue, anxiety, depression, physical and emotional layers both.

About Me Specifically

Biggest day-to-day CG impact: _____
What I am doing well right now: _____
What I am worried about: _____
How I best receive medical information (written / verbal / both):
_____

Support person you can speak with (if any):
_____

My Current Specialists

- Metabolic / genetics: _____
- Endocrinology: _____
- OB-GYN / reproductive: _____
- Neurology: _____
- Mental health: _____
- Other: _____

What I Need From Primary Care

- Annual physical, age-appropriate general screening, and vaccinations.
- Help coordinating referrals and prior authorizations across my specialists.
- Baseline and follow-up labs: CBC, CMP, lipids, TSH, 25-OH vitamin D, calcium.
- Bone-health monitoring, DEXA referrals when due.
- Mental-health check-ins; referrals to CG-aware therapy when needed.
- A clear plan for sick days: who to call, what counts as urgent.

Medications, Supplements, and Allergies

Medications (include dose): _____
Supplements (calcium, vitamin D, other):
_____
Allergies / intolerances: _____

One Ask

If you are unsure about something CG-specific, please tell me, I would rather hear "I want to check with your metabolic team" than a best guess. I will always share records and reports from my specialists so we can keep the picture unified.

Thank you for reading this. I am looking forward to working with you.

Sincerely,

_____
[Your Name]
Phone: _____ Email: _____

References

Key clinical sources behind this handbook

This handbook is grounded in peer-reviewed research and the International Clinical Guideline developed by the Galactosemia Network (GalNet). The sources below are the primary references for the medical information it contains. Each citation links to the full article.

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