The Classical Galactosemia European Care Pathway

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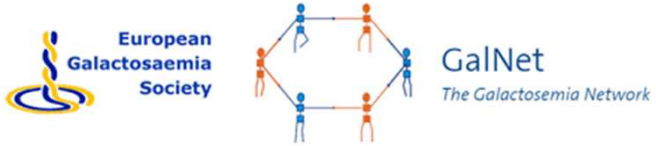


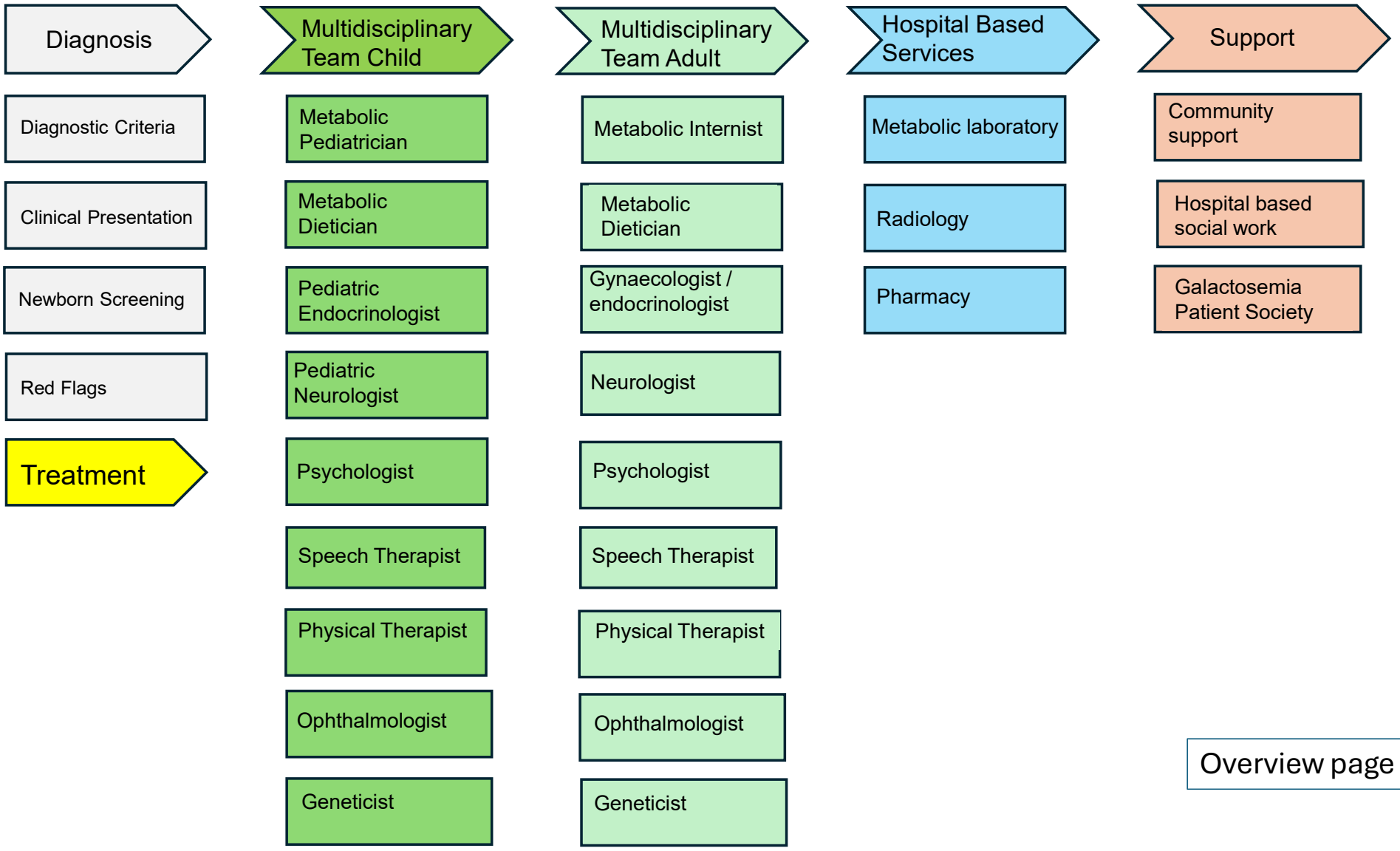
Based on
Welling L, Bernstein LE, Berry GT, Burlina AB, Eyskens F, Gautschi M, Grünewald S, Gubbels CS, Knerr I, Labrune P, van der Lee JH, MacDonald A, Murphy E, Portnoi PA, Õunap K, Potter NL, Rubio-Gozalbo ME, Spencer JB, Timmers I, Treacy EP, Van Calcar SC, Waisbren SE, Bosch AM; Galactosemia Network (GalNet**). International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up.** J Inherit Metab Dis. 2017 Mar;40(2):171-176. doi: 10.1007/s10545-016-9990-5.

Reviewed by the International Galactosemia Network and the European Galactosemia Society

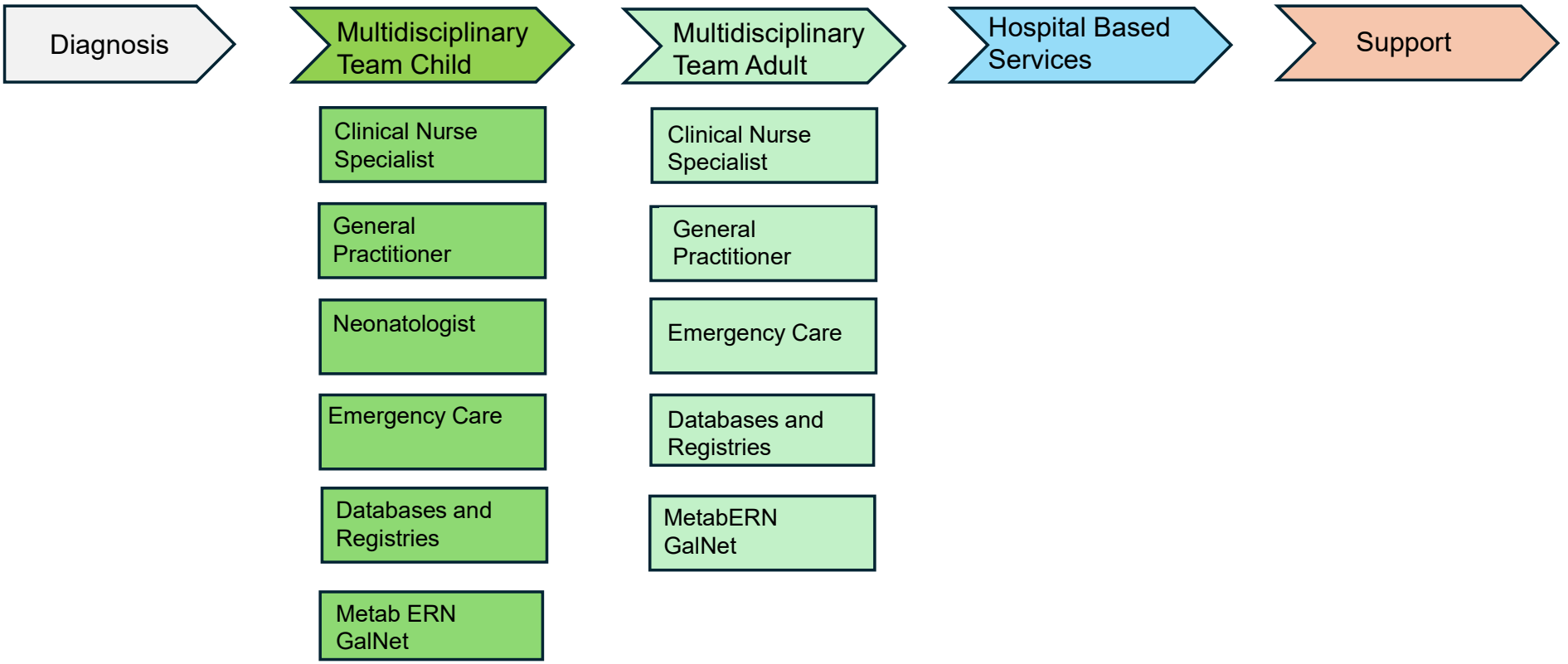
Classical Galactosemia: Clinical Characteristics

Orphacode: 79239
Disease definition: Classical galactosemia is a life-threatening inherited metabolic disease with onset in the neonatal period. Infants usually develop feeding difficulties, lethargy, and severe liver disease.

Orphanet Summary
When ingesting breast milk or lactose-containing formula, infants develop feeding problems, failure to thrive, and signs of liver damage (jaundice, bleeding tendency, hypoglycemia). In the absence of appropriate treatment (galactose restriction), sepsis (E-coli) and neonatal death may occur. Despite adequate treatment, long-term complications appear including cognitive impairments, motor deficits, ovarian dysfunction with reduced fertility in women and diminished bone density.



Overview page 1



Overview page 2

Diagnosis

Diagnostic Criteria

Clinical Presentation

Red Flags

Newborn Screening

Diagnostic Criteria	Gold standard: GALT enzyme activity in red blood cells <15% or Presence of 2 disease causing variants in the *GALT* gene
Clinical Presentation	When ingesting breast milk or lactose-containing formula, infants develop feeding problems, failure to thrive, and signs of liver damage (jaundice, bleeding tendency, hypoglycemia). In the absence of appropriate treatment (galactose restriction), sepsis (E-coli) and neonatal death may occur. Despite adequate treatment, long-term complications appear including cognitive impairments, motor deficits, ovarian dysfunction with reduced fertility in women and diminished bone density.
Red Flags	Infants: early onset feeding problems, failure to thrive, signs of liver damage (jaundice, bleeding tendency, hypoglycemia, neonatal cataracts, sepsis (commonly *ecoli*). Later childhood and adult presentations: signs of liver disease, cataracts, developmental delay, cognitive impairments, motor deficits, speech delay/dyspraxia, ataxia, ovarian dysfunction.
Newborn screening	In some countries, CG patients are identified through NBS, but other countries chose not to include CG in their NBS program. NBS prevents development of critical illness and death, but does not change frequency of long-term complications.

Treatment

Treatment	
Dietary Treatment	Clinicians should immediately commence a galactose restricted diet (e.g., soy-based, casein hydrolysate or elemental formula) if classical galactosemia is suspected in an infant, without waiting for confirmation of the diagnosis. Clinicians should treat patients with a red blood cell GALT enzyme activity below 10 % and/or pathologic variations on both alleles of the GALT gene, including p.S135L, with a galactose-restricted diet. There is not enough evidence to conclude whether patients with 10–15 % red blood cell residual GALT activity should or should not be treated. Treatment is a life-long galactose-restricted diet that eliminates sources of lactose and galactose from dairy products, but permits galactose from non-milk sources that contribute minimal dietary galactose. Small amounts of galactose present in specific mature cheeses and caseinates are acceptable. Allowed are any amount and type of fruits, vegetables, legumes, unfermented soy-based products, mature cheeses (with galactose content <25 mg/100 g), and the food additives sodium or calcium caseinate. Although higher in galactose, all fermented soy-based products can be allowed in the small quantities that are typically used in the diet. **See: Metabolic Dietician**
Supportive treatments	Depending on longterm complications of individual patients: Among others but not exclusively, see specific sections for infomation • Speech therapy • Physical therapy • Cognitive support • Psychosocial support • Hormonal therapy • Occupational therapy

Multidisciplinary Team Child

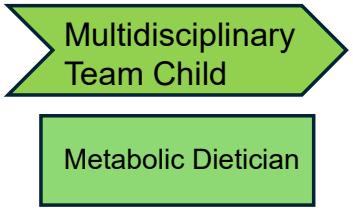
Metabolic Pediatrician

Multidisciplinary Team Child	Metabolic Pediatrician : follow up
• Growth	Growth must be evaluated at every outpatient clinic visit
• Puberty development	Girls with CG should be screened for hypergonadotropic hypogonadism if they reach the age of 12 years with insufficient secondary sex characteristics or if they reach the age of 14 years with no regular menses. Screening should include follicle-stimulating hormone and 17-beta-estradiol. Girls should be referred to a pediatric endocrinologist. We recommend that girls and women with CG, who have gone through puberty and established regular menstrual periods, should be monitored annually for menstrual abnormalities, secondary amenorrhea, and symptoms of primary ovarian insufficiency (POI). We do not recommend routine endocrinology follow-up in males. **See further information under "Pediatric Endocrinologist"**
• Speech and language development	All children with CG should be screened for speech and language delay at ages 7–12 months, 2, 3, and 5 years (consider combining with cognitive screening).If children show low or borderline speech and language development, full assessments should be conducted. See further information under "Speech Therapist"
• Cognitive and Social development	Clinicians should refer patients for testing of developmental quotient (DQ) and intellectual quotient (IQ), to obtain a well-validated measure of development and cognitive abilities. At minimum, testing should be done at ages 2-3, 4,5, 8-10.12-14, further according to needs. **See further information under "Psychologist"** It is recommended to screen children for psychosocial deficits, including autism spectrum disorders, sensory integration problems, depression and anxiety, using standardized questionnaires such as the Behavior Assessment System for Children, Second Edition (BASC-2) in English or a similar tool in other languages, at age 2 years in combination with screening for speech and language delays and in combination with developmental testing at ages 4–5, 8–10, and 12–14 years.

Multidisciplinary Team Child

Metabolic Pediatrician

Multidisciplinary Team Child	Metabolic Pediatrician : follow up
• Motor development	Clinicians should screen patients with CG for neurological involvement by clinical examination from the age of 2– 3 years. Such screening should include examination for ataxia, tremor, dysmetria, and dystonia. If a specific neurological deficit is noted, monitoring of progression with a designated scale is advised. Clinicians should ask patients or caregivers about onset of seizure and seizure-like activity since previous examination and perform an EEG, if indicated.
• Cataract	Clinicians should refer all patients to an ophthalmologist for evaluation of cataract at the time of diagnosis. Ophthalmological follow-up is necessary in patients with a cataract at diagnosis until it has fully resolved. We recommend performing ophthalmological screening in all patients who are non-compliant with diet.
• Biochemical follow up	**Red blood cell Gal-1-P levels:** At diagnosis, and after 3 and 9 months of dietary galactose restriction. Yearly after the first year of life until an individual baseline has been established In case of increase in galactose intake and concern about intoxication. **Calcium and Vitamin D:** annual dietary assessment with measurement of plasma total 25-OH-vitamin D levels.
• Bone Health	Bone mineral density (BMD) should be assessed by age appropriate dual-energy X-ray absorptiometry (DXA) scan from age 8–10 years. With evidence of reduced bone density (Z-score≤−2.0), follow-up according to current pediatric bone health guidelines is advised. Without evidence of reduced bone density, we recommend performing a repeat dual-energy X-ray absorptiometry scan when puberty is complete and thereafter every 5 years. Treatment should be started according to WHO FRAX recommendations. We recommend comprehensive dietary evaluation, optimization of calcium intake if needed, monitoring and if necessary supplementation of vitamin D, hormonal status evaluation and hormone replacement therapy consideration, as well as regular exercise and assessment of skeletal problems and clinically significant fractures in all patients with CG. Supplementation of vitamin K might be beneficial when combined with an adequate intake of calcium and vitamin D, but currently there is not enough evidence to recommend the routine use of vitamin K. At present there is not enough evidence to justify routine determination of bone turnover markers in patients with CG.



Multidisciplinary Team Child	Metabolic Dietician
Dietary treatment	Clinicians should immediately commence a galactose restricted diet (e.g., soy-based, casein hydrolysate or elemental formula) if classical galactosemia is suspected in an infant, without waiting for confirmation of the diagnosis. Clinicians should treat patients with a red blood cell GALT enzyme activity below 10 % and/or pathologic variations on both alleles of the GALT gene, including p.S135L, with a galactose-restricted diet. There is not enough evidence to conclude whether patients with 10–15 % red blood cell residual GALT activity should or should not be treated. Treatment is a life-long galactose-restricted diet that eliminates sources of lactose and galactose from dairy products, but permits galactose from non-milk sources that contribute minimal dietary galactose. Small amounts of galactose present in specific mature cheeses and caseinates are acceptable. Allowed are any amount and type of fruits, vegetables, legumes, unfermented soy-based products, mature cheeses (with galactose content <25 mg/100 g), and the food additives sodium or calcium caseinate. Although higher in galactose, all fermented soy-based products can be allowed in the small quantities that are typically used in the diet. We do not advice a galactose restricted diet in the non-galactosemic mother carrying a child with galactosemia
Diet analysis	We recommend an annual dietary assessment of calcium and vitamin D intake with measurement of plasma total 25- OH-vitamin D levels. Both calcium and vitamin D should be supplemented as necessary following the age-specific recommendations for the general population.

Multidisciplinary Team Child

Pediatric Endocrinologist

Multidisciplinary Team Child	
Pediatric Endocrinologist	We advise referral to an endocrinologist who is part of a Classical Galatosemia Center of Expertise. Girls with CG should be screened for hypergonadotropic hypogonadism if they reach the age of 12 years with insufficient secondary sex characteristics or if they reach the age of 14 years with no regular menses. Screening should include follicle-stimulating hormone and 17-beta-estradiol. We recommend considering follicle stimulating hormone level, growth, and psychosocial maturity of the individual girl, for determination of age at start of treatment. For puberty inducement, a low dose estrogen in a step-wise escalating dose is used, then later combined with cyclic progesterone for regular withdrawal bleeds. We recommend that girls and women with CG, who have gone through puberty and established regular menstrual periods, should be monitored annually for menstrual abnormalities, secondary amenorrhea, and symptoms of primary ovarian insufficiency (POI). Changes in menses or POI symptoms should be evaluated with a serum folliclestimulating hormone level. Anti-Müllerian hormone measurement is not helpful in determining which women will undergo POI, but may be helpful in identifying women at risk for imminent POI when it is undetectable. Imaging by pelvic ultrasound or MRI is not recommended unless otherwise clinically indicated. We recommend not using anti-Müllerian hormone and ovarian imaging routinely for follow-up as these have not been shown to accurately predict pubertal development or fertility outcome. We recommend providing counseling about adequate birth control methods for women who do not desire pregnancy. While combined oral or transdermal contraceptives may provide cycle control, bone protection, and attenuate hot flashes, they may fail to provide adequate birth control in women with very elevated follicle-stimulating hormone levels. Fertility preservation may not be successful. Currently, fertility preservation techniques are not yet readily used in everyday practice. We recommend fertility preservation should only be offered with appropriate institutional research ethics review board approval to girls with classical galactosemia at a young pre-pubertal age. We do not recommend routine endocrinology follow-up in males.

Multidisciplinary Team Child

Pediatric Neurologist

Multidisciplinary Team Child	
Pediatric Neurologist	Clinicians should screen patients with CG for neurological involvement by clinical examination from the age of 2 to 3 years. Such screening should include examination for ataxia, tremor, dysmetria, and dystonia. If a specific neurological deficit is noted, monitoring of progression with a designated scale is advised. It is suggested to screen adult patients annually and to record progression, if any. Pediatric patients could be screened more frequently (every 6 months) in order to identify potentially modifiable neurological problems. We do not recommend routine brain and spinal cord imaging in the follow-up of patients with CG. In those patients with significant or progressive neurological symptoms and signs, imaging may be warranted to (1) determine if a second condition is present or (2) further define the development and progression of neuroradiology findings in individual patients.

Multidisciplinary Team Child

Psychologist

Multidisciplinary Team Child	
Psychologist	Clinicians should refer patients for testing of developmental quotient (DQ) and intellectual quotient (IQ), to obtain a well-validated measure of development and cognitive abilities. At minimum, testing should be done at: Age 2 to 3 years: to assess early speech/language and motor development in time for early intervention, using a standardized test instrument such as the Bayley Scales of Infant and Toddler Development (BSID) or a similar measure. Age 4 to 5 years: to assess school readiness and need for occupational therapy and speech-language therapy, using a standardized test instrument such as the Wechsler Preschool and Primary Scale of Intelligence (WPPSI) or a similar measure. Age 8 to 10 years: to assess cognitive development, specific areas of strengths and weaknesses, and the need for special therapies, using a standardized test instrument such as the Wechsler Intelligence Scale for Children (WISC) or a similar measure. Age 12 to14 years: to assess cognitive development and specific areas of strengths and weaknesses, and to assess the need for special therapies, using a standardized test instrument such as the Wechsler Intelligence Scale for Children (WISC) or a similar measure. Age 15 years and older: according to needs, specific questions. For obtaining a measure of functioning when formalized testing is not possible or when additional assessments are needed between formalized testing points, we recommend using a validated parent/informant questionnaire, such as the Adaptive Behavior Assessment System (ABAS) or a similar measure. We recommend a clinical assessment of executive function, if feasible in the clinic, with specific attention to processing speed and visual spatial comprehension. In children (8 to 10 years) as a first screening use the Behavior Rating Inventory of Executive Function (BRIEF), and in adolescents (12 to 14 years) and in young adults (18 to 20 years) use the Cambridge Neuropsychological Test Automated Battery (CANTAB), the Amsterdam Neuropsychological Tasks program (ANT) or a similar measure, with follow-up, as needed.

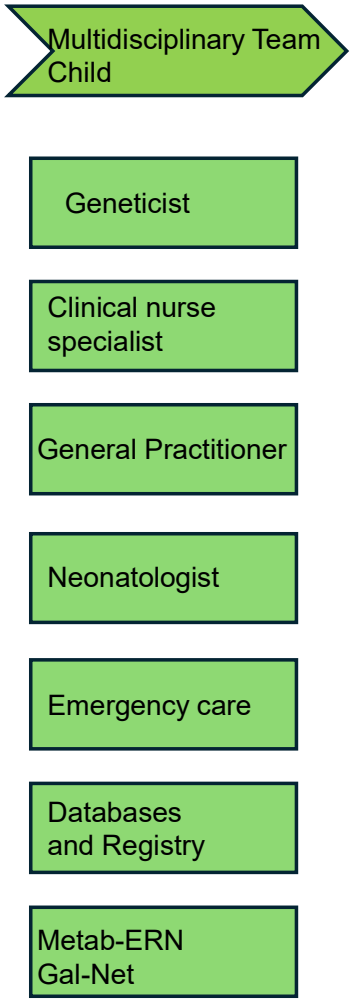
Multidisciplinary Team Child

Language and Speech Therapist

Physical Therapist Rehabilitation

Ophthalmologist

Multidisciplinary Team Child	
Language and Speech Therapist	All children with CG should be screened for speech and language delay at ages 7 to12 months, 2, 3, and 5 years. If children show low or borderline speech and language development, full assessments should be conducted. We recommend that an assessment of speech and language includes hearing screening, a brief assessment of pre-linguistic communication (<2 years of age) and expressive, receptive, and pragmatic language use, structure-function examination, motor speech (observation of respiration, resonance, voice, articulation), and speech intelligibility for all children not meeting age appropriate milestones. We recommend a cognitive evaluation as well if a disorder is suspected. For children who are not meeting age appropriate speech or language milestones, we recommend treatment based on guidelines for treatment of speech, language, and voice disorders in the general population. Therapy should begin during the first year of life and include modeling and training of gestural communication to increase infant and toddler language development. Play-based milieu for language development is recommended during the second year of life. Individual speech therapy focused on high repetition of a small number of targets should begin during the second year of life and continue as needed throughout the preschool and elementary school years. Respiration, phonation, and resonance deficits should also be addressed.
Physical Therapist Rehabilitation	Referral for support in case of motor problems
Ophthalmologist	Clinicians should refer all patients to an ophthalmologist for evaluation of cataract at the time of diagnosis. Ophthalmological follow-up is necessary in patients with a cataract at diagnosis until it has fully resolved. We recommend performing ophthalmological screening in all patients who are non-compliant with diet.



Multidisciplinary Team Child	
Geneticist	Referral for counseling of parents of affected children. Addressing mode of inheritance, recurrence risk, indentification of at risk relatives, cascade testing, reproductive options.
Clinical Nurse Specialist (or pediatrician)	Coordination of care Advise on research and Registry opportunities, consent Pathway and Guidelines education Coordinate and prepare for transition to adult department
General Practitioner	Management of intercurrent illness Coordination of local services and supports Consult metabolic pediatrician if needed
Neonatologist	Diagnose based of clinical symptoms or after newborn screening Consult and refer to Pediatrician for Metabolic Diseases
Emergency care	Management of intercurrent illness Consult metabolic pediatrician if needed
Databases and Registry	Participate in Registry and Databases Metab ERN and GalNet
Metab ERN and GalNet	Participation in Registry and Databases Crossborder consultations through Metab ERN GALNET: https://www.galactosemianetwork.org/ MetabERN: European Reference Network for Metabolic Disorders

Multidisciplinary Team Adult

Metabolic Internist

Multidisciplinary Team Adult	Metabolic Internist : follow up
• Endocrinology	Females must be monitored for hypergonadotropic hypogonadism, POI, and fertility issues by a gynaecologist / endocrinologist. We do not recommend routine endocrinology follow-up in males. **See further information under "gynaecologist / endocrinologist"**
• Speech and language	Refer to speech therapist on indication
• Cognition and Psychosocial situation	Refer to a psychologist on indication Clinicians should refer patients age 15 and for testing of developmental quotient (DQ) and intellectual quotient (IQ) according to needs, and specific questions. We recommend a clinical assessment of executive function, if feasible in the clinic, with specific attention to processing speed and visual spatial comprehension. In adolescents (12–14 years) and in young adults (18–20 years) use the Cambridge Neuropsychological Test Automated Battery (CANTAB), the Amsterdam Neuropsychological Tasks program (ANT) or a similar measure, with follow-up, as needed. We recommend screening adults for mental health issues with validated questionnaires that include brief scales for anxiety and depression, such as the NIH PROMIS Questionnaires, Beck Anxiety Inventory (BAI), Beck Depression Inventory (BDI) or similar measures. With adults, we recommend discussing living situations, work and/or educational situations, satisfaction with social relationships, and sexual intimacy during outpatient clinic visits and to refer for professional consultation, if necessary. **See further information under Psychologist**

Multidisciplinary Team Adult

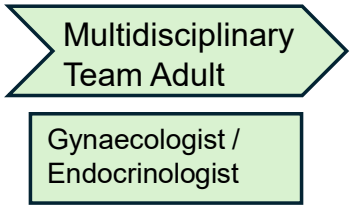
Metabolic Internist

Multidisciplinary Team Adult	Metabolic Internist : follow up
• Neurology	Clinicians should screen patients with CG for neurological involvement by clinical examination. Such screening should include examination for ataxia, tremor, dysmetria, and dystonia. If a specific neurological deficit is noted, monitoring of progression with a designated scale is advised. Clinicians should ask patients or caregivers about onset of seizure and seizure-like activity since previous examination and perform an EEG, if indicated. We do not recommend routine brain and spinal cord imaging in the follow-up of patients with CG. In those patients with significant or progressive neurological symptoms and signs, imaging may be warranted to (1) determine if a second condition is present or (2) further define the development and progression of neuroradiology findings in individual patients.
• Cataract	Ophthalmological follow-up is necessary in patients with a cataract at diagnosis until it has fully resolved. We recommend performing ophthalmological screening in all patients who are non-compliant with diet.
• Biochemical follow up	**Red blood cell Gal-1-P levels:** Yearly after the first year of life until an individual baseline has been established, In case of increase in galactose intake and concern about intoxication. **Calcium and Vitamin D:** annual dietary assessment with measurement of plasma total 25-OH-vitamin D levels.
• Bone Health	Bone mineral density (BMD) should be assessed by age appropriate dual-energy X-ray absorptiometry (DXA) scan. We recommend performing a repeat dual-energy X-ray absorptiometry scan when puberty is complete and thereafter every 5 years. Treatment should be started according to WHO FRAX recommendations. We recommend comprehensive dietary evaluation, optimization of calcium intake if needed, monitoring and if necessary supplementation of vitamin D, hormonal status evaluation and hormone replacement therapy consideration, as well as regular exercise and assessment of skeletal problems and clinically significant fractures in all patients with CG. Supplementation of vitamin K might be beneficial when combined with an adequate intake of calcium and vitamin D, but currently there is not enough evidence to recommend the routine use of vitamin K. At present there is not enough evidence to justify routine determination of bone turnover markers in patients with CG.

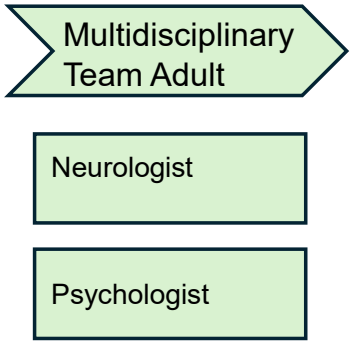
Multidisciplinary Team Adult

Metabolic Dietician

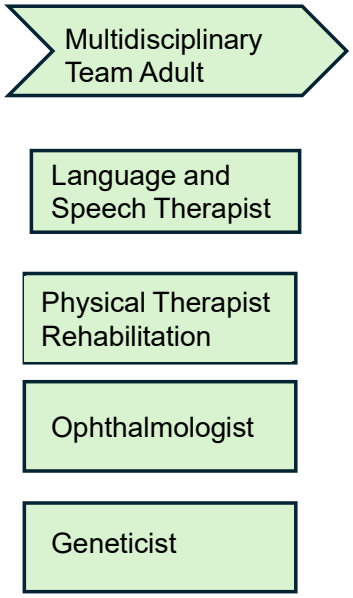
Multidisciplinary Team Adult	Metabolic Dietician
Dietary treatment	Clinicians should treat patients with a red blood cell GALT enzyme activity below 10 % and/or pathologic variations on both alleles of the GALT gene, including p.S135L, with a galactose-restricted diet. There is not enough evidence to conclude whether patients with 10–15 % red blood cell residual GALT activity should or should not be treated. Treatment is a life-long galactose-restricted diet that eliminates sources of lactose and galactose from dairy products, but permits galactose from non-milk sources that contribute minimal dietary galactose. Small amounts of galactose present in specific mature cheeses and caseinates are acceptable. Allowed are any amount and type of fruits, vegetables, legumes, unfermented soy-based products, mature cheeses (with galactose content <25 mg/100 g), and the food additives sodium or calcium caseinate. Although higher in galactose, all fermented soy-based products can be allowed in the small quantities that are typically used in the diet.
Diet analysis	We recommend an annual dietary assessment of calcium and vitamin D intake with measurement of plasma total 25- OH-vitamin D levels. Both calcium and vitamin D should be supplemented as necessary following the age-specific recommendations for the general population.



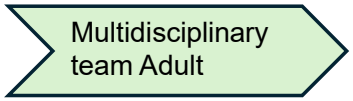
Multidisciplinary Team Adult	
Gynaecologist Endocrinologist Obstetrician	We recommend referral to an endocrinologist / gynaecologist /obstetrician who is part of a Classical Galactosemia Center of expertise, for endocrine, fertility and obstetrical issues. We recommend not using anti-Müllerian hormone and ovarian imaging routinely for follow-up as these have not been shown to accurately predict pubertal development or fertility outcome. We recommend that girls and women with CG, who have gone through puberty and established regular menstrual periods, are monitored annually for menstrual abnormalities, secondary amenorrhea, and symptoms of primary ovarian insufficiency (POI). Changes in menses or POI symptoms should be evaluated with a serum folliclestimulating hormone level. Anti-Müllerian hormone measurement is not helpful in determining which women will undergo POI, but may be helpful in identifying women at risk for imminent POI when it is undetectable. Imaging by pelvic ultrasound or MRI is not recommended unless otherwise clinically indicated. We recommend that women with hypergonadotropic hypogonadism, or primary ovarian insufficiency should be provided counseling and support about their reproductive options and management of irregular or absent menses. Hormone replacement therapy should be initiated with the onset of secondary amenorrhea to reduce the risk of osteoporosis and other complications of primary ovarian insufficiency. We recommend considering a referral to a reproductive endocrinologist for women who desire pregnancy and have been unable to conceive naturally, or for women who desire additional counseling about fertility treatment options including oocyte donation. We recommend providing counseling about adequate birth control methods for women who do not desire pregnancy. While combined oral or transdermal contraceptives may provide cycle control, bone protection, and attenuate hot flashes, they may fail to provide adequate birth control in women with very elevated follicle-stimulating hormone levels. An intrauterine device may provide the lowest failure rate. We do not recommend routine endocrinology follow-up in males.



Multidisciplinary Team Adult	
Neurologist	Clinicians should screen patients with CG for neurological involvement by clinical examination. Such screening should include examination for ataxia, tremor, dysmetria, and dystonia. If a specific neurological deficit is noted, monitoring of progression with a designated scale is advised. Clinicians should ask patients or caregivers about onset of seizure and seizure-like activity since previous examination and perform an EEG, if indicated. We do not recommend routine brain and spinal cord imaging in the follow-up of patients with CG. In those patients with significant or progressive neurological symptoms and signs, imaging may be warranted to (1) determine if a second condition is present or (2) further define the development and progression of neuroradiology findings in individual patients.
Psychologist	Clinicians should refer patients age 15 and for testing of developmental quotient (DQ) and intellectual quotient (IQ) according to needs, and specific questions. We recommend a clinical assessment of executive function, if feasible in the clinic, with specific attention to processing speed and visual spatial comprehension. In adolescents (12–14 years) and in young adults (18–20 years) use the Cambridge Neuropsychological Test Automated Battery (CANTAB), the Amsterdam Neuropsychological Tasks program (ANT) or a similar measure, with follow-up, as needed. We recommend screening adults for mental health issues with validated questionnaires that include brief scales for anxiety and depression, such as the NIH PROMIS Questionnaires, Beck Anxiety Inventory (BAI), Beck Depression Inventory (BDI) or similar measures. With adults, we recommend discussing living situations, work and/or educational situations, satisfaction with social relationships, and sexual intimacy during outpatient clinic visits and to refer for professional consultation, if necessary.



Multidisciplinary Team Adult	
Language and Speech Therapist	Referral for support in case of speech problems
Physical Therapist Rehabilitation	Referral for support in case of motor problems Referral for support in case of movement disorder (tremor, ataxia)
Ophthalmologist	Ophthalmological follow-up is necessary in patients with a cataract at diagnosis until it has fully resolved. We recommend performing ophthalmological screening in all patients who are non-compliant with diet.
Geneticist	Genetic counselling for and affected females and males of reproductive age, re mode of inheritance, recurrence risks, identification of at risk relatives, cascade testing, reproductive options



Clinical nurse specialist

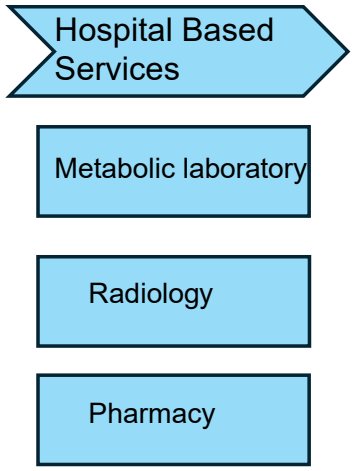
General Practitioner

Emergency care

Databases and Registry

Metab-ERN GalNet

Multidisciplinary Team Adult	
Clinical Nurse Specialist (or pediatrician)	Coordination of care Advise on research and Registry opportunities, consent Pathway and Guidelines education Coordinate and prepare for transition to adult department
General Practitioner	Management of intercurrent illness Coordination of local services and supports Consult metabolic pediatrician if needed
Emergency care	Management of intercurrent illness Consult metabolic pediatrician if needed
Databases and Registry	Participate in Registry and Databases Metab ERN and GalNet
Metab ERN and GalNet	Participation in Registry and Databases Crossborder consultations through Metab ERN GALNET: https://www.galactosemianetwork.org/ MetabERN: European Reference Network for Metabolic Disorders



Hospital based services	
Metabolic laboratory	Confirmation of diagnosis GALT enzyme measurement *GALT* DNA studies Monitoring galactose-1-phosphate uridyltransgerase
Radiology	Dexa scan
Pharmacy	Advice on galactose content medication

Support

Community support

Hospital based social work

Galactosemia patient Society

Support	
Community support	Refer if neccessary: Public Health Nurse Primary Care Social worker Mental Health Social Worker Disability Social Worker
Hospital based social work	For specific needs and advise for community support team
Galactosemia patient Society	The European Galactosemia Society Website also lists National Societies Welcome - Galactosaemia.eu

European Reference Network
MetabERN
European Reference Network for Hereditary Metabolic Disorders

European Galactosaemia Society

GalNet
The Galactosemia Network

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