

Classic Galactosemia: a comparison of “best practices” around the world

Instructions: The goal of this project is to compare standard or “best” practices from as many different doctors and countries as possible with regard to the diagnosis, treatment, and outcome of patients with classic galactosemia. Please enter your response in the space allotted beneath each question. Feel free to expand the form to enter more information, as needed. Email your completed survey as an MS Word attachment within 2 weeks of receipt to Judy Fridovich-Keil at jfridov@emory.edu. Thank you!!!

ABOUT YOU

Name/ Date:

Position:

Address:

Phone number:

Email:

ABOUT YOUR COUNTRY

Country name:

Healthcare system:

DIAGNOSIS OF CLASSIC GALACTOSEMIA

How are patients with classic galactosemia ascertained in your country (clinical symptoms vs. newborn screening)?

At what age are most galactosemic patients diagnosed?

How is a diagnosis of classic galactosemia confirmed in your country?

What is the frequency of births affected by galactosemia in your country?

Are all affected infants ascertained, or do you believe some are missed? What happens to affected infants who are “missed”?

Comments:

EARLY INTERVENTION – THE FIRST YEAR AND BEYOND

What is the standard of care for early intervention (for symptomatic vs. pre-symptomatic patients)? Please describe.

What parameters are followed to ascertain how well a patient is responding to intervention?

Who oversees early intervention (specialist, general pediatrician, other)?

How often are affected infants seen for follow-up during the first year?

How often are affected children seen for follow-up after the first year? What are the most important parameters followed in childhood?

Comments:

DIETARY CONSIDERATIONS

What is the prescribed diet for infants and older children with classic galactosemia (e.g. soy formula, elemental, other)?

For toddlers or older children, how strictly is galactose intake restricted? Examples: restriction of dairy products only vs. restriction of dairy as well as other sources (e.g. tomatoes).

How well do most patients/ families adhere to the prescribed diet?

Is dietary restriction of galactose continued indefinitely, or is the diet relaxed for older patients? If yes, how, and when?

Is the diet perceived by patients and families to be a significant hardship? Why or why not?

Comments:

PUBERTY AND THE TEEN YEARS

How frequently are teenage patients followed (e.g. yearly)?

What are the most important components of follow-up?

Any special considerations with regard to follow-up for this group (e.g. concerns about ovarian insufficiency in girls)?

How well do most teenage patients adhere to the prescribed diet?

Comments:

LONG-TERM FOLLOW-UP AND CARE

How frequently are adult patients followed (e.g. yearly)?

Who oversees long-term follow-up (specialist, general internist, other)?

What are the most important components of follow-up for adults?

Any special considerations with regard to follow-up for this group (e.g. concerns about pregnancy and lactation in women)?

Comments:

CONSIDERATIONS FOR OLDER ADULTS (over 50 yrs)

Are there any special considerations for follow-up or intervention in older patients (>50 yrs old)?

For women on long-term hormone replacement are hormones eventually reduced or eliminated to simulate natural menopause? Why or why not?

Comments:

OUTCOMES

What are the outcomes you observe? Please complete the table below, and then in the space following the table please list and explain any other long-term complications you see in your patients that were not described in the table.

NOTE: When estimating each outcome frequency, please consider only patients who are old enough to assess accurately with regard to that specific outcome. For example, if some of your patients are infants, please do not include those patients as “no speech problems” in your estimate. Similarly, when calculating the frequency of ovarian insufficiency in your patient population, please consider only your female patients who are old enough to demonstrate clear evidence of ovarian function or insufficiency. For each outcome parameter please state the number of patients evaluated for that parameter; this number may vary from parameter to parameter since not every patient is appropriate to evaluate for every parameter. Thank you!

Outcome Issue	Approximate % of your patients with classic galactosemia who experience this outcome (please put an “X” in the correct box)				# patients evaluated for this parameter
	0%≤X<25%	25%≤X<50%	50%≤X<75%	75%≤X≤100%	
Social problems					
Cognitive Problems					
Ovarian Insufficiency					
Tremor/Ataxia					
Reduced Bone Density					
Visual Impairment					
Speech problems					
Growth (weight/height)					
Anemia (low RBC)					
Liver Problems					
Kidney Problems					
Vitamin D Deficiency					

Other outcome issues?

Do you see any patterns that might help to explain why some patients experience certain complications while others do not? Examples: age at diagnosis, GALT genotype, adherence to diet, hemolysate gal-1P values, early palliative intervention (e.g. speech therapy), other? Please explain.

ANYTHING ELSE?

Please use this space to make any further comments or to explain any potentially unusual aspects of how patients with classic galactosemia are diagnosed or treated in your country.

THANK YOU!! THANK YOU!! THANK YOU!! THANK YOU!! THANK YOU!! THANK YOU!! THANK YOU!!