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Anxiety and stress in parents as well as the burden of raising a child with classical galactosemia

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Summary

Aim. The aim of the study was to measure anxiety and stress in parents of children with classical galactosemia compared to parents of healthy children and to determine the association of anxiety and stress with the burden of raising an affected child among Polish parents.

Material and methods. The study involved 94 parents of children with classical galactosemia and 87 parents of healthy children. Anxiety and stress were measured using the Polish adaptations of the State-Trait Anxiety Inventory (STAI) and the Perceived Stress Questionnaire (PSQ). The burden of raising a sick child was assessed using the Galactosemia Quality of Life Survey.

Results. Parents of children with classic galactosemia showed increased levels of anxiety and stress. Compared to fathers, mothers of such children manifested significantly higher levels of anxiety, emotional tension and general, external and intrapsychic stress. In mothers, the level of general stress correlated positively with the age of the child with classic galactosemia. Differences in parenting style, social sphere and anxiety about the future state of health of the children were found between Polish and Dutch parents.

Conclusions. The emotional state of parents of children with classical galactosemia warrants the need for development of dedicated intervention and prevention programs to improve the quality of life and mental health of families caring for patients with inborn metabolic diseases.

Key words: anxiety, stress, classical galactosemia

Introduction

Classical galactosemia (CG) (OMIM 230400) is a rare disorder of galactose metabolism caused by a deficit in galactose-1-phosphate uridyltransferase activity [1]. As a result of the enzyme block, toxic metabolites accumulate, causing damage to many systems and organs, in particular the central nervous system, liver, kidneys, lenses, and ovaries [2]. Despite restrictive dietary treatment, CG is characterised by a varied clinical course with late complications. They represent a major clinical problem and their pathomechanism remains unclear. Late complications include cognitive deficits, intellectual disability, abnormal development of active speech, neurological symptoms (including impaired balance and coordination, brachybasic gait, hand tremors, scanning speech), ovarian dysfunction, and abnormal skeletal mineralisation [3, 4]. Problems in emotional-social functioning, such as difficulty in establishing and maintaining relationships with peers, social withdrawal, symptoms of anxiety and depressed mood, are also observed in CG patients [5, 6]. Due to the variety of ailments, patients require systematic, multidisciplinary care throughout their lives. Like other congenital disorders of biochemical metabolism, due to the nature of the condition and the multi-profile therapeutic regimen, as well as the prognostic uncertainty regarding the occurrence of long-term complications, CG poses a challenge not so much for professionals, but especially for the parents of the affected child.

The aim of the study is to assess the anxiety and stress of parents of children with classic galactosemia compared to parents of healthy children, and to determine the association of anxiety and stress with the burden of raising an affected child among Polish parents. In addition, the responses of Polish and Dutch parents regarding the burden of raising children with galactosemia were compared.

The following research questions were posed:

1. Does the level of perceived anxiety and stress differ between parents of healthy and CG children?
2. Is there a correlation between the age of the CG child and the level of anxiety and stress perceived by the parents?
3. Are there differences in anxiety and stress levels between mothers and fathers of children with classic galactosemia?
4. Is there a relationship between the level of anxiety and stress and the perceived severity of burdens associated with raising a child with CG? If so, what is the nature of this relationship?
5. Are there differences in the responses of Polish and Dutch parents regarding the burden of raising a child with CG?

Material and methods

Study participants

The analysis included 94 parents (55 females and 39 males, aged between 25 and 69, with an average age of 38 years) of children with CG aged 3–18. The comparison group consisted of 87 parents (51 females and 36 males, aged between 24 and 69, with an average age of 39 years) of healthy children aged 3–18.

The size of the population of children suffering from CG aged 3–18 in 2013–2018 (*n* = 109) was estimated on the basis of the medical records of the Metabolic Disease Clinic of the Institute of Mother and Child in Warsaw. The method of selection of the study group was based on systematic random selection. Of the 109 children with CG aged 2–18 years, every second child was drawn, and then, to ensure the group was representative in terms of gender and age, successive subjects were selected using a stratified sampling method. The algorithm for systematic random selection of the study group is shown in Figure 1. The diagnosis of CG and the age of the patient were used as inclusion criteria. Exclusion criteria were co-occurring disorders/diseases that

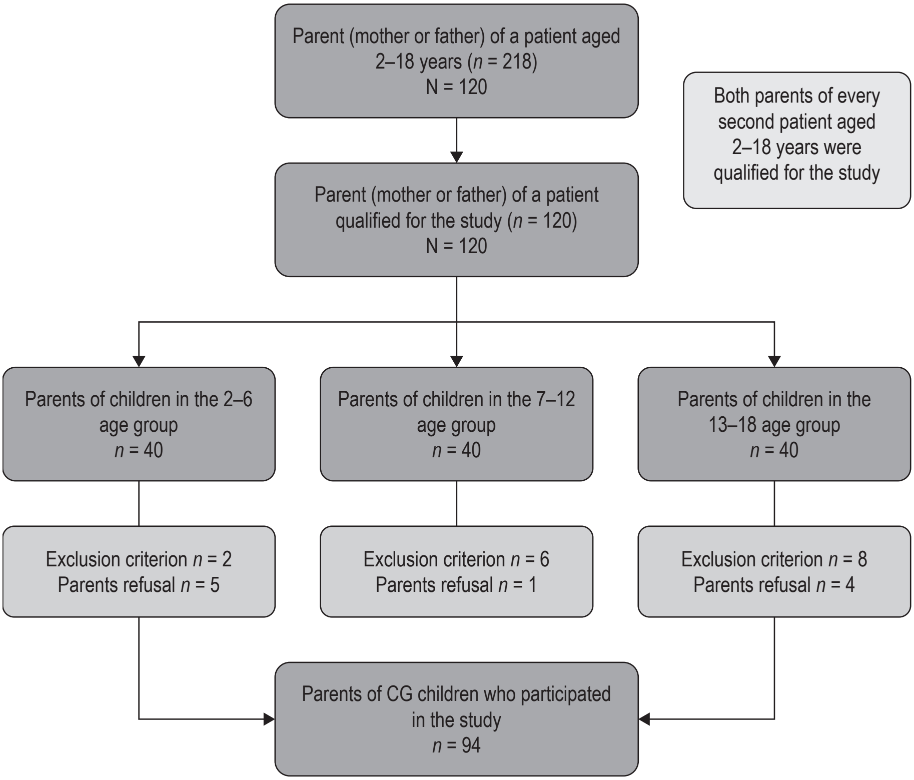

Figure 1. **Algorithm for selection of the study group**

were not long-term complications of CG and factors that prevented the completion of self-report questionnaires. A non-random purposive sampling method was used to ensure a comparable diversity of parents of healthy children according to the following criteria: place of residence, age and sex of child.

Research tools

To assess parents' socio-emotional variables, the following instruments were used: the *State-Trait Anxiety Inventory* (STAI) [7] and the *Perceived Stress Questionnaire* (PSQ) [8]. The STAI and the PSQ are tools commonly used in research and clinical practice, both in healthy and somatically ill populations. The STAI is based on the distinction between anxiety understood as a temporary and situationally conditioned state of an individual and anxiety being a relatively permanent element of personality. The STAI is used to survey people over the age of 15. It consists of two scales: L-*trait* and L-*state*, each of which contains 20 statements. The person under examination is asked to respond to each statement by determining the intensity of his feelings on a scale of 1 to 4. The Polish version of the inventory is highly reliable. The Cronbach's internal consistency coefficient was 0.82 to 0.92 for the L-*state* and 0.86 to 0.92 for the L-*trait*. The theoretical relevance of the test was examined by evaluating the correlations of the results with data obtained from other tools that examine similar areas. Raw scores for each subscale range from 20 to 80 points and can be converted using either sten or centile scores. The survey takes about 15–20 minutes.

The PSQ is a tool available and used in Poland to measure the structure of stress experiences of adolescents over 16 and adults. It makes it possible to estimate the overall level of perceived stress and its three dimensions: "emotional stress" (understood as a sense of restlessness, excessive nervousness, irritability, fatigue), "external stress" (related to experiencing unpleasant sensations arising from social interactions) and "intrapsychic stress" (characterised by a subjective sense of personal defectiveness and lack of competence). An additional dimension of the PSQ is the "lie scale". It is used to verify the statements of respondents. The authors of the test assumed that some respondents may tend to improve their image or be characterised by low levels of criticism and poor insight. The PSQ consists of 27 items. The respondent is to relate to them and express an opinion on a 5-point scale, the extreme answers of which are: "True"/"Not True". The internal consistency coefficients for the three dimensions of stress range from 0.70 to 0.81. The reliability of the "lie scale" was (0.57). Both factor and theoretical accuracy of the tool were confirmed. The time allotted for the survey was not strictly defined, being about 20 minutes for most of the respondents.

The burden of raising a child with CG was assessed using the *Galactosemia Quality of Life Survey* by Bosch et al. [9]. The author's permission was obtained to use 19 questions related to the experiences of parents of children with CG. The questions address, among other items, the sense of burden on parents, as well as on the family as a whole, associated with raising a child with CG, feelings of isolation and exclusion, differences in raising a child with CG and healthy siblings, and parents' concerns about the development of their offspring. The responses obtained in Poland and the Netherlands were compared. The results obtained in the study from the Netherlands were taken from the source paper [9].

An original questionnaire was used to obtain demographic data and assess socioeconomic status. The study was approved by the Bioethics Committee.

Statistical analysis

The conformity of the results' distribution with a normal distribution was tested using the Shapiro-Wilk test. In order to compare parents of CG and of healthy children in terms of quantitative variables, Student's *t*-test analysis was performed for independent samples.

Pearson's *r* correlation analysis was carried out to establish the relationship between the quantitative variables. The analyses included a Bonferroni significance level correction for multiple comparisons. IBM SPSS Statistics version 26 was used for the analyses. A level of $p = 0.05$ was considered statistically significant.

Results

The characteristics of parents of galactosemia-affected and of healthy children are shown in Table 1. There were no differences between the groups in terms of age, sex, place of residence, marital status or education. Among parents of CG children, the proportion of unemployed parents was higher than in the group of parents of healthy children. Mothers of children with CG were more likely to give up work to care for their child. Parents of children with CG rated their material situation worse than parents of healthy children.

Table 1. **Characteristics of a sample of parents of children with CG and parents of healthy children**

Variables	**Parents of children with CG** ($n = 94$)	**Parents of healthy children** ($n = 87$)	***p***
Age, *M* (*SD*)	37.81 (6.11)	39.08 (8.78)	0.257[a]
Sex, *n* (%)			
Women	55 (58.5)	51 (58.6)	0.988
Men	39 (41.5)	36 (41.4)	
Place of residence, *n* (%)			
Countryside	41 (43.6)	38 (43.7)	0.989

table continued on the next page

City, below 50,000 residents	28 (29.8)	28 (32.2)	
City, 51,000–100,000 residents	7 (7.4)	8 (9.2)	
City, 101,000–250,000 residents	12 (12.8)	8 (9.2)	
City, 251,000–500,000 residents	4 (4.3)	3 (3.4)	
City, above 501,000 residents	2 (2.1)	2 (2.3)	
Marital status, *n* (%)			
Marriage	88 (93.6)	76 (87.4)	0.348
Cohabitation	3 (3.2)	5 (6.7)	
Single parent	3 (3.2)	6 (6.9)	
Education, *n* (%)			
Primary	4 (4.3)	6 (6.0)	0.218
Vocational	4 (4.3)	2 (2.2)	
Secondary	48 (51.1)	55 (63.2)	
Tertiary	38 (41.3)	24 (27.6)	
Occupational situation, *n* (%)			
Professionally active	71 (75.5)	86 (98.9)	<0.001
Unemployed	23 (24.5)	1 (1.1)	
Mothers giving up their jobs due to the need to take care of a child, *n* (%) Financial situation, *n* (%)	25 (47)	6 (11.6)	<0.001
Very bad	3 (3.2)	1 (1.1)	0.016
Bad	8 (8.5)	3 (3.4)	
Neither good nor bad	38 (40.4)	23 (26.4)	
Good	39 (41.5)	43 (49.4)	
Very good	6 (6.4)	17 (19.5)	

[a] – p-value for Student's t-test for independent samples. For the other variables, the analysis was performed using either the χ^2 test of independence or Fisher's exact test.

The responses of only one of the parents (either mother or father) were included.

Comparison of perceived anxiety and stress levels in parents of CG children and parents of healthy children

Significant differences were found between the groups in terms of anxiety and stress levels for all analysed variables (except the "lie scale", Table 2). Mothers of CG children showed significantly higher levels of anxiety as a state (weak effect) and as a trait (strong effect) compared to mothers of healthy children. They were also found to have higher levels of emotional tension (strong effect), external stress (strong effect), intrapsychic stress (moderate effect), and general stress (strong effect) than mothers of healthy children.

Table 2. **Comparison of anxiety and stress levels in mothers of healthy and CG children**

	CG children (n = 55)		**Healthy children** (n = 51)				**95% CI**		
	M	***SD***	***M***	***SD***	***T***	***p***	***LL***	***UL***	**Cohen's *d***
Level of anxiety									
state anxiety	39.45	9.93	35.49	9.35	2.11	**0.037**	0.24	7.69	0.41
trait anxiety	47.84	11.01	37.80	5.94	5.90	**<0.001**	6.59	13.42	1.18
Level of stress									
PSQ – emotional tension	22.65	5.79	17.73	4.32	4.94	**<0.001**	2.95	6.91	0.97
PSQ – external stress	21.62	6.14	15.02	4.88	6.10	**<0.001**	4.45	8.74	1.20
PSQ – intrapsychic stress	21.64	10.04	16.00	5.22	3.66	**<0.001**	2.58	8.70	0.74
PSQ – lie scale	19.96	4.19	18.76	4.17	1.48	**0.143**	-0.41	2.81	0.29
PSQ – general stress	64.82	17.03	48.75	12.72	5.53	**0.001**	10.31	21.84	1.08

n – group size; *M* – mean; *SD* – standard deviation; *t* – test value; *p* – statistical significance; *LL* and *UL* – the upper and lower limits of the confidence interval; Cohen's *d* – standardised effect size

Significant differences between the groups were found for anxiety as a trait and emotional tension, intrapsychic stress, general stress, and for the lie scale (Table 3). Fathers of CG children showed significantly higher levels of anxiety as a trait (moderate effect) compared to the group of fathers of healthy children. They also demonstrated higher levels of emotional tension (moderate effect), intrapsychic stress (moderate effect), general stress (moderate effect), and higher scores on the lie scale (weak effect) than fathers having healthy offspring.

Table 3. **Comparison of anxiety and stress levels in fathers of healthy and CG children**

	CG children (n = 39)		**Healthy children** (n = 36)				**95% *CI***		
	M	***SD***	***M***	***SD***	***t***	***p***	***LL***	***UL***	**Cohen's *d***
Anxiety level									
State anxiety	34.67	9.01	33.03	6.99	0.87	0.385	-2.09	5.37	0.21
Trait anxiety	38.72	10.58	34.17	7.11	2.17	**0.033**	0.37	8.74	0.51
Stress level									
PSQ – emotional tension	18.49	5.35	15.06	4.64	2.96	**0.004**	1.12	5.74	0.69
PSQ – external stress	16.51	6.75	14.28	4.92	1.63	0.108	-0.50	4.97	0.38
PSQ – intrapsychic stress	15.56	6.32	12.86	4.26	2.15	**0.035**	0.20	5.20	0.51
PSQ – lie scale	18.28	4.34	16.08	4.62	2.12	**0.037**	0.14	4.26	0.49
PSQ – general stress	50.31	16.70	41.19	12.52	2.36	**0.021**	1.28	14.95	0.62

n – group size; *M* – mean; *SD* – standard deviation; *t* – test value; *p* – statistical significance; *LL* and *UL* – the upper and lower limits of the confidence interval; Cohen's *d* – standardised effect size

Correlations between the age of child with CG and parents' levels of perceived anxiety and stress

The mothers' levels of emotional tension, external stress, intrapsychic stress, and general stress correlated positively at a weak to moderate level with the age of the affected child, meaning that the older the child was, the greater the stress. In the fathers' group, the correlations between the child's age and ratings of anxiety, stress and care were not significant.

Table 4. **Pearson's correlations between child age and anxiety and stress levels of mothers and fathers of CG children**

	Age of a child			
	Mother		**Father**	
	r	***p***	***r***	***P***
state anxiety	0.03	0.859	0.11	0.523
trait anxiety	0.23	0.095	0.25	0.132

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PSQ – emotional tension	0.30	**0.027**	0.11	0.519
PSQ – external stress	0.28	**0.039**	0.13	0.442
PSQ – intrapsychic stress	0.41	**0.002**	0.21	0.207
PSQ – lie scale	-0.06	0.661	-0.11	0.491
PSQ – general stress	0.35	**0.008**	0.13	0.418

r – Pearson's correlation coefficient; *p* – statistical significance

Anxiety and stress levels according to the gender of the parent of a child with CG

Statistically significant differences in perceived levels of anxiety and stress were found between parents. Mothers of CG children were characterised by significantly higher levels of anxiety as a state (moderate effect) and as a trait (strong effect), higher levels of emotional tension (strong effect), external stress (strong effect), intrapsychic stress (moderate effect), and general stress (strong effect) compared to fathers of CG children.

Table 5. **Comparison of anxiety and stress levels by gender of parent**

	Mother (n = 55)		**Father (n = 39)**				**95% *CI***		
	M	***SD***	***M***	***SD***	***T***	***p***	***LL***	***UL***	**Cohen's *d***
Level of anxiety									
state anxiety	39.45	9.93	34.67	9.01	2.39	**0.019**	0.81	8.76	0.50
trait anxiety	47.84	11.01	38.72	10.58	4.02	**<0.001**	4.61	13.62	0.84
Level of stress									
PSQ – emotional tension	22.65	5.79	18.49	5.35	3.55	**0.001**	1.83	6.50	0.74
PSQ – external stress	21.62	6.14	16.51	6.75	3.81	**<0.001**	2.45	7.76	0.80
PSQ – intrapsychic stress	21.64	10.04	15.56	6.32	3.33	**0.001**	2.46	9.69	0.70
PSQ – lie scale	19.96	4.19	18.28	4.34	1.89	**0.062**	-0.09	3.45	0.40
PSQ – general stress	64.82	17.03	50.31	16.70	4.10	**<0.001**	7.49	21.53	0.86

n – group size; *M* – mean; *SD* – standard deviation; *t* – test value; *p* – statistical significance; *LL* and *UL* – the upper and lower limits of the confidence interval; Cohen's *d* – standardised effect size

Analysis of semi-structured interviews with parents of CG children

Parents of Polish and Dutch children with CG responded similarly to the interview questions. Half of the parents perceived CG in terms of a burden on the family system, with the majority reporting anxiety related to the illness, CG's negative impact on interactions with the child, and a sense of personal burden related to caring for the offspring. Most parents of both Polish and Dutch girls declared concerns about fertility.

The greatest discrepancies were evident in the way the children were brought up, the social sphere and the anxiety about the future health of the children with CG. Almost 70% of Dutch parents treat their CG child in the same way as their healthy offspring. In Poland, only 46% of parents gave such an answer. Differences were also noted in questions relating to the social sphere: Polish parents experienced exclusion due to their child's illness to a greater extent (36% vs 21%; $p = 0.019$), had less belief in a happy life for children with CG (68% vs 86%; $p = 0.002$), and expressed significantly more concern about their child's health (43% vs 11%; $p < 0.001$).

Table 6. **Distribution of responses of Dutch (n = 97) and Polish (n = 94) parents of children with CG in a semi-structured interview, expressed as a percentage**

		Totally agree		Agree		Disagree		Totally disagree	
		N	**P**	**N**	**P**	**N**	**P**	**N**	**P**
1	Galactosemia as a genetic disease is a burden to our family	14	20	32	26	43	41	11	13
2	The care of my child with galactosemia is a great burden	19	18	41	36	30	37	10	9
3	I feel isolated as a parent of a galactosemic child	4	12	17	24	57	42	22	22
4	It bothers me that my child suffers from galactosemia	54	49	32	32	13	14	1	5
5	One can live a good life with galactosemia	22	14	64	54	12	27	2	5
6	I treat my child with galactosemia the same way as my healthy child	37	20	32	25	30	39	1	16
7	I watch over my child with galactosemia more than over my healthy child	17	30	42	35	32	17	9	18
8	Galactosemia affects the contact with my child	41	42	32	23	18	14	9	21

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9	I raise my child with galactosemia in a different way than my healthy child	3	22	20	26	41	28	36	24
10	My child's friends know what galactosemia is	8	20	43	31	39	48	10	1
11	My child suffers from unclear speech	13	17	25	29	42	35	20	19
12	Hospital visits are a burden to our family	2	17	33	25	45	39	20	19
		Almost always		**Often**		**Some-times**		**Almost never**	
		H	**P**	**H**	**P**	**H**	**P**	**H**	**P**
13	It bothers me to go to the hospital for follow up	10	6	16	14	46	45	28	35
14	I find it difficult when my child needs to have a blood test	30	10	20	14	30	39	20	37
15	My child with galactosemia is jealous of his/her healthy sibling	0	11	8	17	47	53	45	19
16	I worry about the future of my child	15	33	23	16	47	27	15	24
17	I worry about the results from blood tests and urine tests	11	5	7	11	32	49	50	35
18	I worry my child may become ill	4	17	7	26	48	17	41	40
19	I worry my female child might suffer from infertility	20	37	35	19	35	32	10	12

N – Netherlands; P – Poland

Analysis of the relation between parents' anxiety and stress levels and the burden of raising a child with an illness

Higher anxiety as a state and as a trait was associated with a greater burden on parents in relation to the child's illness itself, the demands of care, the need for regular hospital visits, a sense of isolation, and a higher level of anxiety due to the child's suffering from a chronic disease (positive correlations ranging from weak to strong) (Table 7). The higher the sense of state anxiety and trait anxiety, the greater the impact of the disease on contact with the child, and the higher the anxiety about the child's future, including possible additional diseases or diseases resulting from the underlying illness as well as suffering from infertility (positive and moderate correlations). In addition, state anxiety was associated with worrying about the child's blood test results.

Moderate negative correlations were noted between the severity of state and trait anxiety and the opinion on the possibility of living a good life despite suffering from CG, and with the assessment of the knowledge about galactosemia of the child's friends, as well as between state anxiety and treating a CG child the same as a healthy one.

Positive correlations at weak to moderate levels were found between all dimensions of stress (emotional tension, external stress, intrapsychic stress) and overall stress intensity and the burden on the family because of the illness itself, caregiving, the need for regular hospital visits, a sense of isolation, with higher levels of anxiety due to the child's suffering from a chronic disease and concerns about the possibility of long-term consequences of galactosemia, such as infertility or developmental disorders. The higher the intensity of the parent's stress, the greater the impact of the child's illness on contact with the child (positive and moderate/strong correlations). Emotional tension was positively correlated at weak to moderate levels with stress associated with hospital visits and worry about the child's future and blood test results. Worrying about blood test results was positively and moderately correlated with higher levels of external and intrapsychic stress, and worrying about blood test results was correlated with increased intrapsychic stress and overall stress levels (weak and positive correlations). Negative correlations at a moderate to strong levels were found between all dimensions of stress as well as the total score and the opinion of the ability of a child with galactosemia to live a good life, the assessment of the knowledge about galactosemia of the child's friends, and treating a child with the disease the same as healthy offspring (except for external stress – a non-significant relationship).

Table 7. **Pearson's correlation coefficient between levels of anxiety and stress and the burden of raising a child with classical galactosemia**

	State anxiety	Trait anxiety	**Emotional tension**	**External stress**	**Intrapsychic stress**	**General scale**
Galactosemia as a genetic disease is a burden to our family	0.45**	0.53**	0.53**	0.49**	0.52**	0.54**
The care of my child with galactosemia is a great burden	0.46**	0.45**	0.50**	0.42**	0.48**	0.49**
I feel isolated as a parent of a galactosemic child	0.54**	0.51**	0.56**	0.49**	0.49**	0.53**
It bothers me that my child suffers from galactosemia	0.37**	0.39**	0.46**	0.29**	0.41**	0.40**
One can live a good life with galactosemia	-0.44**	-0.43**	-0.56**	-0.42**	-0.47**	-0.51**

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I treat my child with galactosemia the same way as my healthy child	-0.32*	-0.27	-0.42**	-0.26	-0.41**	-0.36*
I watch over my child with galactosemia more than over my healthy child	0.20	0.08	0.21	0.12	0.24	0.20
Galactosemia affects the contact with my child	0.41**	0.45**	0.55**	0.45**	0.56**	0.55**
I raise my child with galactosemia in a different way than my healthy child	0.11	0.06	0.04	0.09	0.10	0.08
My child's friends know what galactosemia is	-0.42**	-0.44**	-0.42**	-0.30**	-0.37**	-0.39**
My child suffers from unclear speech	0.31**	0.29**	0.36**	0.27*	0.33**	0.33**
Hospital visits are a burden to our family	0.32**	0.25*	0.24*	0.16	0.16	0.16
It bothers me to go to the hospital for follow up	0.19	0.07	0.06	-0.05	-0.01	-0.02
I find it difficult when my child needs to have a blood test	0.04	0.06	0.12	0.11	0.14	0.13
My child with galactosemia is jealous of his/her healthy sibling	-0.05	-0.05	0.14	0.03	0.12	0.14
I worry about the future of my child	0.31**	0.43**	0.47**	0.39**	0.44**	0.44**
I worry about the results from blood tests and urine tests	0.23*	0.19	0.25*	0.21	0.28*	0.26*
I worry my child may become ill	0.30**	0.41**	0.39**	0.38**	0.40**	0.42**
I worry my female child might suffer from infertility	0.32*	0.32*	0.48**	0.39*	0.42**	0.45**

*$p < 0.05$; **$p < 0.01$

Discussion

A galactose-restricted diet is the only available treatment for CG, which is effective in saving lives in the neonatal period, but does not prevent long-term complications [10]. The medical aspects of CG are well described, unfortunately, less attention has been paid to psychosocial factors. A consequence of the paucity of in-depth research addressing the psychosocial issues associated with CG is the lack of adequate therapeutic interventions for patients and their families. In recent years, the main focus was on the area of health-related quality of life (HRQoL) [9, 11–13].

In our study, we attempted to analyse the emotional condition of parents of children with CG. We found significant differences between parents of healthy and sick children in terms of anxiety and stress levels for all analysed variables. Parents of CG children manifested significantly higher levels of anxiety. They were also characterised by higher levels of emotional tension, external stress, intrapsychic stress, and general stress. Higher levels of emotional variables can be explained on many levels. CG belongs to the group of rare diseases. The very nature of these diseases, linked to the lack of familiarity with such diseases among most medical professionals, including, for example, in primary care and education, generates an increased risk of emotional burden within the family.

Another factor is the inability to predict the child's future functioning, even if dietary recommendations are followed. In our study, emotional tension was correlated with worry about the child's future. CG has symptoms that affect cognitive, emotional, social, motor, physical, speech, and language spheres and affect basic aspects of life, such as education, work, social interactions, and daily activities [5]. CG patients have less educational achievements than their healthy peers [12]. Deficits in attention, memory, executive function, visuospatial, and language skills are observed in the majority of patients, regardless of intellectual functioning [1 ,4, 5, 14, 15]. Difficulties become particularly apparent when the child reaches school age, increasing the family burden associated with the illness. Cognitive disorders determine patients' educational attainment and consequently affect the sphere of emotional and social functioning. The presence of a number of specific limitations in the process of adaptation to education is not only related to reduced cognitive processes. School and work activities are also hampered by speech disorders as well as resting or intention tremors [16, 17]. In view of the above, parents' anxiety and stress may also stem from concern about their child's independence in adult life. Another important source of anxiety for parents are ideas about the future of their chronically ill offspring after the death of their caregivers. This is particularly evident in CG patients who require the support of another person in their daily functioning in the area of securing material needs, interacting with institutions as well as implementing medical recommendations. An increase in the impact of the child's illness on contact with the parent as the stress intensified was documented.

Higher levels of anxiety and stress in parents of adolescents with CG may be explained by the overlap of normative-developmental difficulties arising from adolescence with symptoms of the underlying disease [19]. Adolescence is characterised not only by adolescents' oppositional attitude towards existing patterns, but also sometimes by rebellion against their own health [20]. During adolescence, a full understanding of the nature of the disease along with its consequences emerges. The knowledge of the course of the condition and subsequent prognosis can exacerbate anxiety in adolescents and lead to depression, while rebellion and denial of authority contribute to non-adherence to treatment recommendations or experimentation with treatment. It is also important to bear in mind the occurring shift in taking responsibility for the implementation of the treatment process from the parent to the adolescent, which triggers anxiety in the caregivers due to the loss of control over the implementation of medical recommendations.

The observed relationship between parental gender and perceived anxiety and stress can be seen in the context of the prevailing socio-cultural, patriarchal division of social roles, especially outside large agglomerations. In the past, society shifted the blame for the child's disability or illness onto the mother, which contributed to her low self-esteem and social ostracism. The results of Gray's qualitative study [18] suggest that a child's intellectual disability often results in the family returning to a traditional pattern of relationships between the partners. The researcher indicated that a child's disability significantly affects mothers' emotional wellbeing and careers, ultimately confining them to a traditional gender role. Mothers not only take on the burden of implementing medical and educational recommendations, but also burden themselves with social responsibility for their child's behaviour.

The traditional view of the division of marital responsibilities places greater responsibility on women for the development and upbringing of their offspring. Many of them choose to sacrifice their professional and personal ambitions in favour of looking after the child on a full-time basis. Our own study showed that up to 74% of the parents of CG patients resided in rural areas or towns up to 50,000 inhabitants. It is therefore necessary to take into account the considerably reduced access to specialised assistance to support the ill child's development on a daily basis (neurologist, special educator, sensory integration therapist, psychologist, psycho-dietician), but also to support the emotional wellbeing of their parents (psychologist, psychotherapist, psychiatrist). Nearly half of the mothers gave up their careers due to their child's illness. The higher levels of anxiety and stress in women can be explained, among other things, by the depletion of psychological resources resulting from an excess of responsibilities focused on providing care. In addition, higher levels of anxiety and stress may be the result of temperamental variables revealed by gender [21], which is biologically determined on the one hand and culturally reinforced on the other.

Interestingly, in the study presented here, significant differences were noted between groups of parents on the lie scale. Fathers of CG children scored higher than fathers of healthy children. The lie scale recognises individuals who attribute highly desirable social behaviour to themselves and present themselves in a highly favourable light while hiding their faults. The high scores may be related to the fear of social exclusion often experienced by families who have a child with a rare disease [8].

In the Netherlands, Hoedt et al. [13] observed that parents of children with CG rated themselves comparably to parents of healthy children in HRQoL. Unfortunately, these studies did not differentiate the results by the gender of the respondent. Research by Bosch et al. [9] showed that up to 86% of Dutch parents of children with CG ($n = 97$) felt anxious about their child's illness, 46% perceived it as a burden on the family, 60% saw caring for a child with a disease as a huge burden, and 21% experienced a sense of social isolation [9]. This survey revealed similar problem areas. Differences were noted in responses to questions related to feelings of isolation, exclusion, parenting methods, and anxiety about the future and health. These differences can be linked to the health policies of European Union countries. The most important piece of legislation on rare diseases is the EU Council Recommendation of 8 June 2009 on an action in the field of rare diseases. This document sets out the recommended health policy of the member states for people affected by a rare disease, with the overarching aim of harmonising interventions. National action plans in individual EU countries were to be established no later than by the end of 2013. This task was completed in Austria, Belgium, Croatia, Cyprus, the Czech Republic, France, the Netherlands, and the UK, among others. Pending completion of data collection in own research (2018), the National Plan for Rare Diseases in Poland has not been implemented. There is therefore a lack of systemic solutions, including interdisciplinary action, to regulate the situation of families of children with rare diseases.

Attention to the emotional and social situation of parents seems particularly relevant in rare diseases, which are associated with a greater sense of alienation of carers, low public awareness of the disease, limited access to specialised medical care or to reliable educational and information sources. These factors exacerbate the emotional, behavioural and social costs to the parents and thus interfere with the adaptation to the child's chronic illness. The frequent feelings of anxiety, excessive nervousness, frustration, and fatigue experienced by parents in the face of external demands may be due to the depletion of personal resources caused by the illness, as well as a lack of understanding and empathy from the surroundings. It is also worth bearing in mind that when parents of children with CG are diagnosed with symptoms of anxiety and stress, prompt intervention through psychoeducation, psychotherapy and, if indicated, psychiatric pharmacotherapy, offers an opportunity to effectively improve their psychological state.

Limitations of the study

The study had several important limitations. It did not take into account factors such as psycho-social aspects of emotional support being an important predictor of parents' emotional state. The differences between mothers and fathers of CG children found in the study should be interpreted with caution, especially due to the small size of the group of fathers. The authors are aware of the need for a follow-up study of a prospective nature.

The *Galactosemia Quality of Life Survey* used in the study should be regarded as a tool through which very fragmentary and preliminary data were obtained. A full understanding of the issue of parents' attitudes toward the disease and the limitations it imposes on the family requires the acquisition or development of a validated tool.

Despite the significant limitations of the study, it is worth noting that this is the first broad study of parents of Polish CG patients. Moreover, the starting point for their recruitment were virtually all patients with classical galactosemia receiving specialised care in metabolic paediatrics in the <18 age group in Poland.

Conclusions

Parents of children with CG pay high emotional, behavioural and social costs resulting from caring for an ill child. The identification of factors affecting the emotional state of the family is essential for the development of intervention and prevention programmes to improve quality of life and mental health. These programmes should offer comprehensive activities, including workshops and support groups for parents, individual, couples and family therapy, as well as training in relaxation techniques and stress management skills. It is also important to elaborate information materials on the disease for professionals in various fields, such as teachers, psychologists and primary care physicians, which would allow better understanding of the specific developmental needs of CG patients and the challenges their families face.

The concluding objective is to create an environment in which families of children with CG can function with a greater sense of safety, stability and support, which will significantly contribute to improving their quality of life and overall wellbeing.

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