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CLINICAL STUDY

Mood and illness experiences of adults with cystinosis

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Abstract

Background: Cystinosis is an autosomal recessive disorder with an estimated incidence of 1/100,000–200,000 live births. The main complications are renal disease, visual impairment, endocrine abnormalities and growth retardation. *Objective*: Our aim was to describe the mood and illness experiences of adults with cystinosis. *Methods*: Twenty-three patients attending the adult cystinosis clinic at Guy's Hospital, London were asked to complete the Hospital Anxiety and Depression Scale (HADS) questionnaire anonymously. Eighteen months later, 21 patients who were still alive were invited to participate in a semi-structured interview aimed at exploring illness experience. *Results*: Eighteen patients completed the HADS questionnaire (means: depression = 7.2; anxiety = 9.2), and 12 participated in the interviews. Three significant themes emerged: (i) the main physical complaints were tiredness, the impact of short stature and side effects of cysteamine medication, especially halitosis, poor taste and nausea. (ii) Cystinosis has a major impact on relationships, autonomy and social life, including reliance on families for support to self-manage, distress at dependence, social anxiety, reduced social involvement and some positive effects on family cohesiveness. (iii) Patients use a range of individual coping strategies to deal with their illness and medication. *Conclusions*: Adult cystinosis patients reported comparatively high-anxiety and depression scores. Common complaints related to the complications of cystinosis and the side-effects of cysteamine, which impacted on relationships, autonomy and social life. Patients described a wide range of strategies, including benefit finding, for coping with cystinosis.

Keywords

Cystagon, cystinosis, cysteamine, mood, quality of life

History

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Introduction

Cystinosis is a rare autosomal recessive disorder affecting 1/100,000–200,000 live births.[1] It is caused by a defective *CTNS* gene which encodes the protein cystinosin. Cystinosin is necessary to transport the amino acid cystine from lysosomes into the cytoplasm of cells. In cystinosis, intracellular cystine accumulates leading to progressive organ dysfunction.[2,3] Chronic kidney disease (CKD) is a common complication. Other organ manifestations are visual impairment, diabetes mellitus, hypothyroidism, hypogonadism and growth retardation, as well as respiratory and musculoskeletal problems.

The only treatment available is cysteamine (Cystagon®), which reduces intracellular cystine accumulation. It acts by participating within lysosomes and facilitates the conversion of cystine into cysteine and cysteine–cysteamine mixed disulfide, both of which can exit the lysosome.[4] Cysteamine is effective in preventing organ dysfunction and delays the onset of renal failure, hypothyroidism, diabetes and neuromuscular problems, especially if commenced early in life.[5] However, adherence can be difficult to achieve due to frequent dosing and side-effects including body odor, halitosis and nausea.[6,7]

Research to date has primarily focused on the clinical aspects of cystinosis and the search for new therapies with little emphasis on the psychological impact, especially during adulthood. The purpose of this study was to explore the mood and illness experiences of adults with cystinosis.

Materials and methods

Setting

Guy's & St. Thomas' NHS Foundation Trust, London is a tertiary care centre which runs the largest adult cystinosis clinic in the UK. Patients are usually referred from the pediatric clinic at the age of 18 years or renal clinics in other hospitals.

Recruitment

All patients attending the adult cystinosis clinic were sent written information about the study and invited to participate.

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In the 1st phase, all patients ($n = 23$) were sent a Hospital Anxiety and Depression Scale (HADS) questionnaire. In the 2nd phase 18 months later, all patients ($n = 21$) were invited to participate in an interview.

Assessment

Hospital anxiety and depression scale

Anxiety and depression were measured using the well-validated 14-item HADS questionnaire.[8] Seven items assess degree of anxiety and seven assess levels of depression, both with a score range of 0–21. It has been widely used across a range of clinical and general population settings, allowing comparisons between different patient and age-matched groups.

Interview

A topic guide was developed and questions were designed based on the relevant literature and discussions with experts. Depending on patients' preference, the interviews were conducted individually face-to-face in the hospital or over the phone by two interviewers (NB and AM). Interviews were recorded and transcribed verbatim. Psychological counselling was offered in case the need was revealed.

Data analysis

The HADS data were scored to produce individual assessments of anxiety and depression, as well as means and standard deviations for the groups as a whole. The interviews were analyzed using framework analysis which involves five stages: familiarization; identifying a thematic framework; indexing; charting and mapping and interpretation.[9] During the familiarization process interviews were read multiple times and emerging themes were formed. The interviews were coded. Credibility of the codes was checked by a second researcher (CG) who read and coded four randomly selected interviews. Any differences in the codes generated by researchers NB and CG were reconciled with researcher RH. After this, codes were grouped into themes.

Ethics

The study was approved by an Institutional committee. The HADS questionnaires were completed anonymously. Patients who agreed to participate in the interviews gave verbal consent at the beginning and were fully informed that they could withdraw from the interview at any time without giving a reason and without their medical care being affected.

Results

Patient cohort

All the 23 patients (age range 18–47 years, 11 male) were sent a HADS questionnaire. Eighteen patients returned completed questionnaires anonymously. Eighteen months later, 21 patients were still alive and were invited to participate in an interview.

Hospital anxiety and depression scale

The mean anxiety and depression scores (SD) of 18 patients who completed the questionnaire were 9.2 (3.9) and 7.2 (3.4), respectively.

Interviews

Twelve patients (age range 20–47; seven male) agreed to take part in an interview. One participant was a student, one worked full-time, three worked part-time and seven were not working. Eight patients (67%) lived with their parents.

Findings

Three main themes emerged (Table 1).

Theme 1: physical impact

All the 12 patients who participated in the interview emphasized the physical impact of living with cystinosis. Six participants reported feeling different from other people due to their short stature and some reported being teased at school. Four patients complained about photophobia and one participant discussed the effects of being blind. The impact of renal disease, including chronic dialysis and renal transplantation was raised by six participants. Typical complaints were fatigue and absence at school leading to problems with exams and difficulty in holding a full-time job.

The adverse effects of cysteamine, in particular, body odor, halitosis and unpleasant taste were a frequent complaint. Eight participants (66%) recalled incidents of being bullied at school or hurtful comments because of body odor. For nine patients, the taste of the medication and associated nausea affected adherence with treatment.

Theme 2: impact on relationships, autonomy and social life

All the 12 patients emphasized the impact of cystinosis on their family and social life. Ten patients reported relying on their families for assistance and support. Eight patients lived with their parents, of whom four described feeling distressed at their on-going dependence. Three patients expressed a desire to be more independent but felt that their health hindered them from doing so. Three patients stressed that they were responsible for their own self-care and sought assistance only when needed.

The positive effect of cystinosis on family cohesiveness was reported by seven patients. The family bond was strengthened by experiences associated with managing their illness. These participants described feeling fortunate for the support, assistance and understanding of their families.

Eight participants reported that cystinosis had a negative impact on their social life, in particular reduced social involvement through fears about others' responses to them or their own social competence. However, other participants did not experience any problems establishing friendships and felt grateful for the understanding of their friends.

Theme 3: coping strategies

Patients employed different processes to cope with their condition and particularly mentioned the support of family

Table 1. Main themes.

1. Physical impact	
Short stature	*''My school life . . . .I found quite difficult. Because of my physical size I would usually get picked on . . . .you know I would get the usual offensive terms.''*
	''I guess . . . when I was younger kind of being that bit different from people . . . I used to have a real hang up on my height, and you can get teased and stuff at school.''
Visual impairment	*''I always wear sunglasses 'cos when the sun comes out I have to wear them otherwise I will go blind, as the light gets magnified by the crystals. I find that quite difficult.''*
	''It was the cystinosis that caused my blindness. And I think that had the biggest effect on me out of everything.''
Chronic kidney disease	*''I had two kidney transplants and I am now back on dialysis. My kidneys have been the main problem.''*
	''The effects of cystinosis of the kidneys . . . I used to have hospital appearances all the time. Missed out on schooling . . . weren't able to do GCSEs 'cos the kidneys' failed. And I was on dialysis and felt pretty low.''
	''The main problem now is that I can only do part-time work. I couldn't hold down a full time job because I get too tired''
Odour / halitosis	*''The main problem is the smell from the medicine 'cos people don't like it . . . If I don't know them it doesn't hurt me so much, if it's someone I know, that I have told that's it's my medicine and they still say it, then that does upset me.''*
Unpleasant taste / nausea	*''It's been quite tough 'cos I got bullied quite a bit at secondary school 'cos' of the smell . . . .I usually don't take my medication because of it.''*
	''I am not very good with my medication, as my doctor knows. I hate taking it. I hate that I have to do it . . . I purely hate the taste.'' ''Well the drugs . . . it's well known that they taste absolutely minging. So it's really hard to take them. So you just take what you can.'
	''Tablets . . . I can't handle them at all. They make me ill straight away . . . I haven't taken them for about a year. Cysteamine makes me sick. I can't hold it down at all.''
	''I don't take my medicines in the morning, because if I did I would not be able to work because I would be so sick. That's why I would rather have a short life but do more, and my doctors know that.''.
2. Relationships, autonomy and social life	
	''I especially rely on my parents to get the medicines ready and to ensure I get them from the hospital without any problems. I would love to be able to more things by myself, but I don't feel happy about doing that.''
	''[Depend on] *my mum quite a lot at the moment . . . there are a lot of things that I would like to do for myself . . . but at the moment, I can't really.''*
Dependence on family	*''I know that I don't need to depend on people the majority of the time, so I don't. When I am poorly or I need prescription picked up or I need a lift somewhere I don't feel that's a problem, because most of the time I'm not needy like that. Also people ask me when they're ill, so it's a two-way process.''*
Autonomy and self-management	*''I don't work. My mum and dad don't allow me to do no work. I don't go out on my own . . . I depend on my parents a lot . . . Well I can't do much because well I am a small person and very weak, not too strong . . . So I need help because . . . it is tough out there.''*
Support from family	*''The most positive thing I can think of is that my family . . . is very cooperative. My mum always cares for me. If say I want to go out, my dad takes me and I depend on him . . . .they* [the parents] *are always there for me. My mum and dad have been encouraging and supportive throughout their lives and I owe it all to them.''*
	''I don't really have to depend on other people; it's just down to me.''
	''My daughter . . . she is 17, she is my young carer . . . she is brilliant . . . she is there . . . with the medication . . . to nip to the shops for me.''
	''It's hard making friends . . . that is one of the biggest things it's affected because it can make you feel quite alone . . . it has had a bad effect on my social life.''
Impact on social life	*''It makes me a bit more anxious around people because of how I have been treated . . . it also makes it harder to open up to people. I am a lot more reserved because of my condition and I know how some people can react badly to it.''*
3. Coping strategies	
	''I was always deemed very independent, even when I was little. I've always been a 'coper'. You have a choice; you cope or you don't.''
	''There's no reason why I shouldn't lead a normal life now. As long as I keep taking the drugs I feel all right.''
	''I hate that I have to go to the hospital. But I get on with it because people have got it a lot worse, so you just sort of get on with it.''
Resilience	*''I take something called 'crystal breaths' . . . that helps to overpower the smell of the cysteamine. And also brush my teeth regularly and use mouth wash and I chew gum a lot.''*
Adherence with cysteamine	*''Normally I take them before I go to sleep, so the taste passes while I am asleep.''*
Benefit finding	''[It has] *given me some good characteristics like determination, drivev . . . it made me more determined to prove people wrong.''*
	''I think because of my illness, I actually look after myself a lot better. I don't drink alcohol, I don't smoke . . . I have a healthy lifestyle and I think that is something that helps a great deal to . . . manage my condition.''
	''*I look at it as a positive thing . . . I am happy where I am and I think that I am here for a reason. At the age I am I am pretty healthy for the condition I have, so I tend to look at the positive aspects rather than the negatives . . . I just make the most of it, there is no reason why I should not lead a normal life now.''*

and friends. Eight participants highlighted that they had adapted to life with cystinosis despite the difficulties they had encountered. Four participants described individual strategies which had helped to cope with physical symptoms, and 10 participants described methods to overcome the side effects of cysteamine. Positive aspects of living with cystinosis were highlighted by three participants, including beneficial health behaviors (not drinking alcohol to excess or smoking), positive personality attributes (determination and resilience) and an enhanced appreciation of life.

Discussion

This study highlights the physical and psychological impact of cystinosis in adults. Most common complaints related to the physical effects of cystinosis and the side-effects of cysteamine, both of which impacted on relationships, autonomy and social life. Although the HADS scores revealed considerable variation, the mean levels of anxiety and depression were relatively high when compared with published data from other patient groups. For example, the scores were considerably higher than those found for patients with type 1 diabetes[10] and for general practice and general medical patients,[11] but below that of psychiatric outpatients. Those interviewed reported that both the physical consequences of cystinosis and side-effects of cysteamine exerted a major negative impact across many aspects of physical and social functioning. Many participants mentioned traumatic social experiences directly linked to physical signs or disabilities, including bullying at work and school. Similar results were found in a survey of quality of life (QoL) in children and young adults with cystinosis, where the psychosocial aspects of QoL were significantly impaired compared to healthy controls.[12]

All participants acknowledged that treatment with cysteamine could slow disease progression but many were unable to take the medication as prescribed. For some patients, a perceived higher QoL, albeit potentially reduced in years, was preferable to a longer life but with continued side-effects. Additionally, some patients were conscious of the fact that they had survived years past their projected life-span. It is well known that this situation can evoke fatalism where patients consider themselves to have ''no future to lose''.[13] Under these conditions, poor adherence to medication is unsurprising.

Most participants felt that their autonomy was compromised by cystinosis. Impairments in functioning and side-effects of medication were often linked to failures to meet expected adult mile-stones, such as leaving home or experiencing close relationships. Again, this phenomenon is not uncommon in patients with chronic illnesses during childhood.[14]

Parents of children with cystinosis often have a significant influence on the development of autonomy. It is well known that the effect of parental relationship on psychosocial outcomes of children with chronic diseases can be very variable ranging from parental overprotection[15,16] and parental perceptions of child vulnerability[17] to distress.[18] Nonetheless, some participants reported a positive experience of parental care and an appropriate balance between dependence and autonomy.

While our findings suggest reduced QoL in adults with cystinosis, some participants actually reported higher QoL than expected from the severity of their condition. This may be related to their adaptive coping mechanisms and attitudes. Some patients were able to generate beneficial effects of cystinosis including personal growth and an increased implicit appreciation for life and healthy life-style choices. This phenomenon of ''benefit-finding'' is not unusual across life-threatening disease.[19]

This is the first study to explore the illness experiences of adult cystinosis patients and provides information those specialists and patients may use to facilitate appropriate support and treatment. This is particularly important since cystinosis remains an orphan disease and not all nephrologists have experience looking after patients with cystinosis. Secondly, cystinosis affects multiple organ systems which mean that patients often attend several different specialist clinics and may see medical staff that is less familiar with the complexity of cystinosis.

Despite these strengths, our study has some limitations. Firstly, not all patients participated in both studies. The first part of the study was done anonymously, and as a result we were unable to link the results of the HADS with the subsequent information gained through interviews. Also, we were not able to assess any associations between the HADS scores and comorbid conditions. Secondly, patients experienced two different methods of interview (face-to-face or telephone interviews) and two different interviewers. Finally, while the same script was used, patients partaking in telephone interviews had less personal contact with the interviewers, and may have experienced different interviewer styles.

In conclusion, living with cystinosis has significant effects on patients' well-being, social functioning and QoL. Most complaints related to the physical impact of cystinosis and the side-effects of cysteamine, both of which impacted on relationships, autonomy and social life but also led to successful coping strategies. Future research should investigate psychological correlates of resilience, autonomy and distress, to gain insight into ways to better support people with cystinosis.

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Declaration of interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

References

1. Gahl WA, Thoene JG, Schneider JA. Cystinosis. *N Engl J Med.* 2002;347:111–121.
2. Anikster Y, Shotelersuk V, Gahl WA. CTNS mutations in patients with cystinosis. *Hum Mutat.* 1999;14:454–458.
3. Kalatzis V, Antignac C. Cystinosis: From gene to disease. *Nephrol Dial Transplant.* 2002;17:1883–1886.
4. Gahl WA, Balog JZ, Kleta R. Nephropathic cystinosis in adults: Natural history and effects of oral cysteamine therapy. *Ann Intern Med.* 2007;147:242–250.

5. Brodin-Sartorius A, Tete MJ, Niauded P, et al. Cysteamine therapy delays the progression of nephropathic cystinosis in late adolescents and adults. *Kidney Int.* 2012;81:179–189.
6. Gahl WA, Reed GF, Thoene JG, et al. Cysteamine therapy for children with nephropathic cystinosis. *N Engl J Med.* 1987;316: 971–977.
7. Cherqui S. Cysteamine therapy: A treatment for cystinosis, not a cure. *Kidney Int.* 2012;8:127–129.
8. Zigmond AS, Snaith RP. The hospital anxiety and depression scale. *Acta Psychiatr Scand.* 1983;67:361–370.
9. Ritchie J, Spencer L. Qualitative data analysis for applied policy research. In: Bryman A, Burgess R, eds. *Analysing qualitative data*. London, UK: Routledge; 1993:173–194.
10. Shaban MC, Fosbury D, Kerr D, Cavan DA. The prevalence of anxiety and depression in adults with type 1 diabetes. *Diabet Med.* 2006;23:1381–1384.
11. Spinhoven PH, Ormel J, Sloekers PPA, Kempen GIJM, Peckens AEM, van Hemert AM. A validation study of the Hospital Anxiety and Depression Scale [HADS] in different groups of Dutch subjects. *Psychol Med.* 1997;27:363–370.
12. Ulmer FF, Landolt MA, Ha Vinh R, et al. Intellectual and motor performance, quality of life and psychosocial adjustment in children with cystinosis. *Pediatr Nephrol.* 2009;24: 1371–1378.
13. Gibson B, Zitzelsberger H, McKeever P. 'Futureless persons': Shifting expectancies and the vicissitudes of progressive illness. *Socio Health Illn.* 2009;31:554–568.
14. Friedman D, Holmbeck GN, DeLucia C, Jandasek B, Zebracki K. Trajectories of autonomy development across the adolescent transition in children with spina bifida. *Rehabil Psychol.* 2009;54: 16–27.
15. Drotar D. Relating parent and family functioning to the psychological adjustment of children with chronic health conditions: What have we learned? What do we need to know? *J Pediatr Psychol.* 1997;22:149–165.
16. Wells RD, Schwebel AI. Chronically ill children and their mothers: Predictors of resilience/vulnerability to hospital and surgical stress. *J Dev Behav Pediatr.* 1987;8:83–89.
17. Anthony KK, Gil KM, Schanberg LE. Brief report: Parental perceptions of child vulnerability in children with chronic illness. *J Pediatr Psychol.* 2003;28:185–190.
18. Thompson RJ, Gustafson KE, George LK, Spock A. Change over a 12 month period in the psychological adjustment of children and adolescents with cystic fibrosis. *J Pediatr Psychol.* 1994;19: 189–203.
19. De Ridder D, Geenen R, Kuijer R, Van Middendorp H. Psychological adjustment to chronic disease. *Lancet.* 2008;372: 246–1242.