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Living with Gitelman disease: an insight into patients' daily experiences

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Abstract

Background. Gitelman disease presents with musculoskeletal complaints and fatigue. Surprisingly, there is no clear-cut correlation between biochemical abnormalities and symptoms.

Methods. Starting from the hypothesis that the way patients comprehend their illness within their sociocultural frameworks reflects on their way of adapting to it, this study investigated how adult patients experience the disease in everyday life. We conducted a qualitative analysis based on interviews with 12 patients. Interviews were audio recorded, fully transcribed and analyzed using the constant comparative method described by Strauss and Corbin.

Results. A typology of the experiences emerged from the data and was tested on each transcript with an explicit search for disconfirming cases. Patients fell into four main groups: (i) those considering Gitelman disease a disabling illness, (ii) those considering it a normalized illness, (iii) those considering it a different normality and (iv) those considering it an episodic disability. Each pattern of experience was characterized by particular (i) ways of interpreting symptoms (ii) ways of managing Gitelman disease in everyday life, (iii) general lifestyles and (iv) risks for the patient's psychosocial life.

Conclusions. These findings suggest that health care providers should take advantage of considering patients' own perception of the disease in order to adjust the care and advice provided.

Keywords: Gitelman disease; hereditary disease; hypokalaemia; quality of life

Introduction

Mutations in the gene encoding the sodium chloride cotransporter in the distal convoluted tubule cause Gitelman disease. This disorder is characterized by hypokalaemia, alkalosis, hypomagnesaemia, hypocalciuria and normal blood pressure [1, 2].

It is traditionally assumed that on supplementation with potassium and magnesium, Gitelman disease is characterized by a relatively mild course and that affected subjects are asymptomatic [1, 2]. However, the two studies that specifically addressed the quality of life of Gitelman patients provide discrepant results. A study conducted among children shows that the illness does not adversely interfere with subjects' mood or social relationships [3]. This is in contrast with the results of an inquiry among affected adults, indicating an increased prevalence of reported symptoms [4].

It is tempting to assume that in Gitelman disease, quality of life is influenced by the patient's age and gender, by mutation type, by regulatory or modifier genes, by intake of salts and by correction by other tubule segments [1, 2, 4, 5]. In addition to the factors mentioned, the interpretive sociological paradigm provides an original and interesting key for understanding, as it suggests that the way patients comprehend their illness within their sociocultural frameworks reflects on their way of adapting to it [6].

Based on this hypothesis, we undertook a qualitative study to explore how young adult patients subjectively experience their illness in daily life. Specifically, this paper pursues four objectives: to illustrate how participants describe and give meaning to symptoms, to highlight the strategies they use to manage their illness, to illustrate the impact of illness on their daily life and identity and to describe the psychosocial risks correlated with Gitelman disease experience.

Materials and methods

The study population consisted of 12 unrelated Italian Gitelman patients (seven female and five male subjects, aged 20–37 years). The diagnosis had been made ≥5 years before the present study and was based on the following criteria [7]: normal blood pressure, hypokalaemia and hypomagnesaemia of renal origin, increased urinary chloride excretion and reduced urinary calcium excretion. Biallelic mutations in the gene encoding the thiazide-sensitive sodium chloride cotransporter had been identified in 11 patients. The manifestation of the disease, established using

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the criteria of Riveira-Munoz *et al.* [5], was severe in 4 of the 12 patients.

We conducted a qualitative study based on in-depth interviews with the aforementioned 12 patients, according to our standard procedure [8–10], based on the Grounded Theory approach [11]. Grounded Theory is a research method that works in a reverse way from traditional research. Rather than beginning with a hypothesis, the first stage is data collection. From these data, the key elements are marked with a series of codes. These are grouped into similar concepts. From these concepts, categories are formed, which are the basis for the creation of a theory [11].

Participants were current or former patients of one of the authors (A.B.) and were living in northern Italy. We limited the recruitment to this geographical area in order to collect data in a homogeneous sociocultural context. Variability of respondents' experience was ensured by the diversification of the sample in terms of gender, age and education. Approval for the whole study was granted by the governing Ethical Committee and informed consent was obtained.

Interviews were conducted between October 2009 and May 2010.

After attending the laboratory for the measurement of plasma potassium and total magnesium levels [7], the participants were asked to describe their experience in the order and manner desired. However, to make sure all the points were covered, we prepared an interview grid with the main topics to be treated: illness trajectory, difficulties, coping strategies, resources, patient–physician relationship, representation of symptoms and medications, daily activities, social networks, social and personal identity. Interviews lasted about 1 h, were audio recorded and transcribed. During transcription, pseudonyms were given to participants.

Analysis was facilitated by the use of the software Atlas.ti [12]. Consistent with the Grounded Theory approach, data collection and analysis were parallel processes and were performed by one of the authors (M.C. Z.). However, the researchers engaged in regular discussions on emerging patterns to ensure analytical rigor.

A four-folder typology of Gitelman disease experience in daily life was progressively developed and tested on each transcript with an explicit search for disconfirming cases. Processes underlying the different types of experience were also identified. At the end, an overall tendency to saturation was observed, i.e. information from the last interviews tended to be redundant, confirming thus the emergent theoretical model. This paper focuses on part of this model by describing the four-folder typology of Gitelman disease experience.

Results

Analysis reveals four ways of experiencing Gitelman disease in daily life: as a disabling illness, as a normalized illness, as a different form of normality and as an episodic disability.

Each pattern of experience is characterized by a specific way of interpreting symptoms, i.e. of perceiving frequency, intensity, and distress of symptoms, and of giving meaning to them. Based on symptoms interpretation, patients develop peculiar ways of managing their condition in everyday life. They also develop peculiar lifestyles, i.e. stable patterns of behaviour that reflect their personal philosophy of life. Finally, each pattern of Gitelman disease experience is characterized by some correlated risks for the patients' psychosocial life. Table 1 summarizes these patterns and the corresponding clinical and laboratory data.

In the following, we illustrate each pattern of Gitelman disease experience through the description of a specific case.

Alfred: Gitelman disease as a disabling illness

Symptoms interpretation: persistent invasive troubles. The main symptom Alfred perceives is invasive exhaustion. He is unable to realize projects important to him (such as going out with friends) or to assume his daily responsibilities (such as attending school or work regularly). Alfred is focused on his illness and tends to interpret any sign of fatigue as a consequence of the disease. Health is, for him, the ideal condition, i.e. the absence of any nuisance. Assigning a pathological origin to his tiredness, Alfred feels powerless and anxious about his future.

Illness management: over-/undercompliance. In the hope to improve his fatigue, Alfred overcomplies with medical advice. He often takes more supplements than prescribed and follows general medical suggestions as absolute rules. However, being unsatisfied with the results of his efforts, Alfred tends to become discouraged. Thus, sometimes he 'transgresses' his rules to see whether something changes: 'I have never done physical exercise because the doctor discouraged me. But now I'm flying in his face because I go to the gym. I want to see how I feel. I think that he does not agree, because this makes me sweat, but ... '

Lifestyle: disrupted life. Alfred describes his life as characterized by important restrictions: 'I feel constrained by the illness. I dislike my life, because sometimes I feel that I have energy to burn, but I stay quiet: if then I get sick?' Beside restrictions, Alfred considers that he has many obligations: medication taking, medical consultation and diet. Thus, Alfred considers Gitelman disease as disrupting his life and everyday routine, at home, at work and in leisure time. Gitelman disease also influences his self-perception and ideas about the future. Alfred has strong feelings of impotence and defeat and tends to assume a self-pitying attitude: 'I go on, what else can I do? My father tells me: "console yourself, you are not the only one". But why the hell should I care? (...) If I had nothing, I would be happier'. Ultimately, Alfred does not accept the disease and is unable to integrate it into his life.

Risk: chaotic activism. Confronted with symptoms, he is neither able to manage nor able to accept; Alfred's main wish is to find a definitive medical solution. Over the years, he has consulted several health professionals, and he has tried complementary/alternative medicine. His risk is to accumulate attempts for resolving the situation and to be systematically frustrated, alternating this way feelings of hope and despair.

Bertha: Gitelman disease as a normalized illness

Symptoms interpretation: well-controlled troubles. Bertha does not experience strong symptoms. She only reports a general sense of lack of energy and, more rarely, pins and needles in her hands. Despite this lack of symptoms, Bertha has a strong sick identity. 'I don't think I'm like others who have nothing. A healthy person does not have to do all these things'. The disease is seen as a medical condition because of medical treatments. Bertha

Table 1. Patterns of disease experience and clinical and laboratory data in 12 unrelated Italian patients affected with Gitelman disease (seven female and five male subjects, aged 20–37 years)

	Disabling illness, Alfred	Normalized illness, Bertha	Different normality, Clark	Episodic disability, Debora
Symptoms interpretation	Persistent invasive troubles	Well-controlled troubles	Insignificant troubles	Episodic significant troubles
Illness management	Over-/undercompliance	Absolute compliance	Self-management	Collaborative management
General lifestyle	Disrupted life	Slowed-down life	Ordinary life	Vigilant life
Correlated risks	Chaotic activism	Unquestioning passivity	Illness banalization	Medical dependency
N	2	1	7	2
Female gender	0	1	4	2
Age (years)				
Current	23, 24	25	20, 22, 24, 28, 31, 33, 36	28, 37
At diagnosis	13, 17	10	5, 6, 9, 12, 15, 15, 17	6, 7
Disease severe[a] (*N*)	0	1	1	2
Mutations in the gene encoding the sodium chloride cotransporter (Nucleotide level, protein effect)	c.2899A>G, p.Arg967Gly homozygous; c.2542G>A, p.Asp848Asn/ c.506-1G>A, splice site	c.557G>A, p.Gly186Asp/ c.625C>T, p.Arg209Trp	c.184G>A, p.Asp62Asn homozygous; c.557G>A, p.Gly186Asp/c.1742T>A, p.Met581Lys homozygous; c.2295de1, p.Phe765LeuX10; c.1046C>T, p.Pro349Leu/c.1432A>G, p.Lys478Glu; c.2191del, p. Gly731GlyX3/c.1315G>A, p. Gly439Ser; c.1742T>A, p.Met581Lys homozygous; no mutations detected	c.1625T>C, p.Leu542Pro; c.1388G>A, p.Gly463Glu/c.1844C>T, p.Ser615Leu
Current plasma level				
Potassium (mmol/L)	2.6; 2.7	2.9	2.5, 2.5, 2.6, 2.7, 2.8, 2.9, 3.0	2.7; 3.1
Magnesium (mmol/L)	0.61; 0.69	0.52	0.41; 0.43; 0.44; 0.50; 0.52; 0.60; 0.65	0.50; 0.52
Current drug Management	Potassium[b] (*N* = 2), amiloride (*N* = 2), magnesium (*N* = 1)	Potassium[b] and magnesium	Potassium[b] (*N* = 7), spironolactone (*N* = 1)	Potassium[b] (*N* = 2), amiloride (*N* = 1), magnesium (*N* = 1)

[a]Criteria suggested by Riveira-Munoz *et al.* [5].
[b]As potassium chloride.

considers them both the essential condition for her normal functioning and the marker of her illness.

Illness management: absolute compliance. Bertha considers that her role in managing illness is to thoroughly comply with her doctor's instructions. She is extremely diligent: she regularly goes to the specialist, she limits her sweating and she neither forgets to take supplements nor changes the amount of medication absorbed. Ultimately, Bertha does not take any initiative: her compliant attitudes and behaviours aim at maintaining the status quo, avoiding any possible danger.

Lifestyle: slowed-down life. Bertha is satisfied with her life. She considers restrictions and obligations related to her illness as mild constraints. Therefore, she describes her lifestyle as perfectly normal. Despite her perception of normality, however, Bertha's efforts to cope with her illness influence her life rhythm: this is slowed down to adapt to the management rules she follows. For example, Bertha accepted a job which she was over-qualified for as a saleswoman as it is close to home and causes no stress.

Risk: unquestioning passivity. Bertha executes medical recommendations without trying to understand them. For example, she has avoided any physical activity since childhood because the doctors had told her to be careful: 'The doctors told me: "if you take medications and then you do physical exercise, that makes you sweat, it would be better to avoid it (...)." They told me: "do it until when you feel tired, then stop." So I told myself: it's better if I don't do anything. Instead of exercising and discovering that tests aren't good and then I have to take more medication, I prefer doing nothing at all'. Her way of experiencing and managing illness, characterized by lack of reflexivity, risks leading her to a passive attitude in terms of self-management.

Clark: Gitelman disease as a different normality

Symptoms interpretation: insignificant troubles. Clark reports occasional episodes of sleepiness and cramps. Nonetheless, he interprets these troubles as insignificant incidents. He considers that any human being is faced with some health problems. Thus, his perception of health is wide enough to include these mild nuisances. 'Clearly one would prefer to be fit as a fiddle. But as there are so many things that can go amiss in our body, this is the best to have'. Clark also considers his symptoms as the normal consequence of his activities. 'When I say that I am tired, my mother immediately asks me: "have you taken the pills?" But what the hell has this to do with the pills? I am tired because I do many things!'

Illness management: self-management. Clark manages his illness mainly autonomously. The medical specialist played a central role when the situation was unstable, but nowadays, Clark takes his blood test periodically in a public laboratory and interprets the results alone. He consults the specialist only in case of significant changes in his biochemical values or in preparation for particular events (trips, surgical intervention). He has obtained several prescriptions from his general practitioner to build up a supply of medications. Additionally, Clark makes day-to-day decisions concerning the treatment, based on how he feels, on his life constraints and personal values. For example, sometimes he does not take medication because he does not want to draw people's attention. He also develops some personal strategies to cope with symptoms (i.e. eating chocolate). If necessary, he is able to accept symptoms without dramatizing. He is convinced that he has learned to evaluate his body and therefore he is able to take some reasonable risks.

Lifestyle: ordinary life. Clark strongly defends his ordinary identity. He underlines that his life not only is normal but also hyperactive and overbooked. Clark pursues his ambitions without being constrained by his illness, leading an enriched life where there is no place for the sick identity. Additionally, his way of managing illness makes his contact with medical institutions rare, fortifying this way his identity as a healthy person living a normal life.

Risk: illness banalization. Clark tends to forget that he has an illness that needs to be kept under control. For example, he reports that he regularly forgets to take pills. 'I am so involved in the normality of life that I almost forget that I have it (...). The most difficult thing is to remember to swallow the pills'. Moreover, Clark is so self-confident that he admits not having consulted a doctor for years.

Debora: Gitelman disease as an episodic disability

Symptoms interpretation: sporadic significant troubles. Like Clark, Debora considers that she sometimes suffers from unimportant, occasional troubles (sleepiness, cramps). Differently from Clark, however, Debora has to face the memory of traumatic experiences related to Gitelman disease: on four occasions, she suffered from tetanic crises due to flu or hot weather, and she had to go to the hospital for mineral salt injections. During these attacks, Debora was confronted with several problems: physical (during the crisis, she was at risk of heart attack), sensorial (the attack was painful), aesthetic (she suffered from paralysis leaving her with deformed features), psychological (she felt extremely anxious), social (she found it difficult to explain her illness at the first-aid station because of its rarity), and functional (she could hardly move and speak). 'I couldn't drive: with the paraesthesia you are so rigid that you feel like a trunk! You really can't move, you can't even do a phone call! I thought: if nobody does anything, what will I do? (... .) My sister brought me straight to the hospital for injections. And every time I have to pass the same stupid procedure. Last time I got angry. "What are you doing?" [to the doctors]. "We are searching your illness in the computer" "And what about asking me? I just need a little bit of potassium!"'

Illness management: collaborative management. Debora is not especially worried about mild symptoms such as

sleepiness and cramps. She manages them by taking basic precautions, such as taking her medications regularly and avoiding activities which cause excessive sweating. On the contrary, she is extremely worried about tetanic crises. Her efforts are mainly oriented to preventing and limiting them. She monitors her body to recognize warning signs. She organizes her life to be ready to face eventual crisis: she informs her friends about what to do in case of crisis, she always carries a detailed description of her illness and treatment in her wallet, etc. This way of managing illness is not an individual activity: to realize it, Debora needs the collaboration of her family, of her friends and of her doctor.

Lifestyle: vigilant life. Debora describes her life as normal. Despite her perception of normality, however, her efforts to prevent and limit her crises involve a price to pay: the price of continuous vigilance. Gitelman disease does not concretely influence Debora's activities. However, it forces her to continuous self-observation and planning for the worst.

Risk: medical dependency. To manage her tetanic crises, Debora requires the collaboration of her family and of her doctor. Even if she does not ask for help, she needs to know that she can count on them. Otherwise, she feels lost and in danger. Thus, Debora tends to avoid unfamiliar contexts. For example, going on vacation is stressful, and changing doctor is inconceivable. Ultimately, the risk of her way of managing Gitelman disease is developing a kind of dependency, especially on her doctor: 'I am always in contact with him. I really don't know what I would do without him'.

Discussion

In Gitelman disease, the clinical manifestations include musculoskeletal complaints, which are traditionally due to hypokalaemia and hypomagnesaemia, and fatigue, which might be more common in patients with greater degrees of salt wasting. However, there is little or no correlation between, on the one side, the manifestations and, on the other side, either extra- or intracellular electrolyte levels or increased release of prostaglandins [1, 2]. Our data suggest that patients affected by Gitelman disease very heterogeneously experience this genetic disease in their daily life: as a disabling illness, as a normalized illness, as a different form of normality or as an episodic disability.

Surprisingly, we did not observe any correlation between severity of the clinical manifestations [5] and expressed quality of life. For instance, the manifestations were mild in patients who consider Gitelman disease a disabling illness (Table 1). Consistent with the interpretive approach, however, our study was not built up to make statistical correlations but to understand the previous experience of the individuals. Our results show that patients' personal interpretation of symptoms play a role in the diversification of the experiences.

While we are not able to state that the four patterns of experience identified are exactly the same in other diseases, several studies have observed that patients with a same chronic condition can experience their illness in very different ways, according to how they make sense of it [8, 13–16]. In chronic conditions, indeed, patients' interpretation of illness is particularly important. Chronic sufferers live in the so-called 'dual kingdoms of the well and the sick' [17]. Being sick, but asked to live 'as normally as possible' [18], they have to place themselves in between these two extremities.

In Gitelman disease, the uncertainty concerning the meaning of the illness is even more evident. Not only is this a chronic disease, but, although it is well defined at a biomolecular level, it appears particularly ambiguous from a phenomenological point of view: symptoms (cramps and fatigue) can be seen as pathological conditions or as usual experiences, while treatment (potassium and magnesium) can be considered as pharmacological medication or as ordinary dietary supplementation. Thus, since Gitelman disease contains elements of both illness and wellness, people have a particularly wide margin of interpretation of their health condition.

In our study, the majority of participants interpret Gitelman disease as a different kind of normality. Though the qualitative nature of the investigation does not allow for statistical generalization, it seems that most young adults are little affected by the illness.

This study has the advantage of being the first in-depth investigation of daily experience of Gitelman patients using validated qualitative research techniques [19]. It also provides a good example of the potential gulf between 'medical' and 'social' models of disease. However, it has limitations which are unavoidable with investigations on rare phenomena. The Grounded Theory approach [11] suggests that data collection and analysis continue until data saturation. The rarity of the disease, however, meant that we stopped after 12 interviews as new data could only have been collected in other geographical contexts. Signs of saturation were already evident, but some further data would have provided more details on the developed typology, and in particular, on the pattern 'normalized illness', which is constituted by only one person.

Despite this limitation, findings suggest interesting paths for future research and clinical practice. Concerning research, it will be useful to explore what influences the experiences of Gitelman disease. Our ongoing analysis suggests that pivotal are the kind of support provided by patients' family, and patients' personality traits. It will also be worth investigating the transferability of our four-folder typology to other chronic diseases. Concerning clinical practice, these results underline that health care providers would benefit from considering patients' own perception of the disease in order to adjust the care and advice provided. Patients who experience Gitelman disease as a disabling illness could be encouraged to distinguish normal from pathological tiredness; patients who experience the disease as normalized illness should be trained to evaluate their situation day-by-day and to diversify their strategies of management; patients who experience the disease as different normality should be warned about the importance of medical care; finally, patients who experience the disease as an episodic disability could be helped in planning strategies to manage tetanic crises.

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Conflict of interest statement. None declared.

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Metabolic syndrome and the risk of calcium stones

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Abstract

Background. The metabolic syndrome (MS) is associated with increased prevalence of kidney stones, yet the specific stone type remains largely unknown. This study was conducted to assess whether risk factors associated with calcium nephrolithiasis increase with individual characteristics of the MS.

Methods. A retrospective analysis was performed in 109 non-stone-forming subjects and 128 recurrent calcium stone formers from Dallas, Texas. A separate analysis was performed in 140 recurrent calcium stone formers from Bern, Switzerland. Demographic, anthropometric, serum and urinary profiles were measured.

Results. In non-stone formers from Dallas, urinary calcium (3.6 ± 1.8 to 6.0 ± 2.9 mmol/day, $P = 0.0003$ for trend, zero to four features) increased with increasing features of the MS. This change was attendant with a significant rise in supersaturation index (SI) of calcium oxalate

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