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RESEARCH ARTICLES

"You Become Pretty Much a Healthcare Worker"

The Parenting of a Child with Inherited Metabolic Disease and its Metaphors

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Abstract

Parents of a child diagnosed with a rare inherited metabolic disease (IMD) are required to actively participate in their child's treatment, by managing the risk of metabolic crisis and ensuring that they adhere to a stringent diet. In this Research Article, I discuss the specific roles, tasks, and knowledge that Polish and Swedish parents of children with IMDs have developed. I also pay critical attention to the notions and metaphors that parents, clinicians, and social scientists use to describe these. The prevailing metaphors of professionalisation used to describe parental roles contribute to the acknowledgment of these caregivers' unique expertise. However, I argue, they also overly stress an individualistic perspective and obscure the relationality of care, collaboratively achieved between parents, patients, relatives, and healthcare providers.

Keywords

Inherited metabolic diseases, Rare diseases, Illness narrative, Family, Caregiving.

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Introduction

> [The thought that] entered my head is that it lies with us whether he is going to handle it [his treatment]. It is we who will see if he gets the food he should get, the right food … Everything seems to hang … to rest on our shoulders. That he will feel well … [to put it] simply (Ingrid[1], mother of Leonhard, age 10, with VLCADD, Sweden).

> [The nurse said] ‘if something strange happens you come to the emergency room’ ... And so I asked her, ‘What could be strange?’, and she said ‘well, if she gets a fever of 37.5 degrees [C] for example’, uh and I think that ... when she said that I understood how narrow the margins were … (Natasha, mother of Agnete, age 11, with LCHADD, Sweden).

When a newborn child is diagnosed with an inherited metabolic disease (IMD), the whole family embarks on an anxious journey during which they learn to cope with a rare disease that has an intricate aetiology, is very demanding in terms of daily treatment, and affects probably no more than a few dozen children in a given country. For years to come families will have to keep their child on a stringent diet, based on the elimination of common foodstuffs, the use of special medical foods, and a strict feeding schedule. They will also need to monitor their child’s condition in order to make a timely decision on hospitalisation if they present signs of exacerbation (known as ‘metabolic decompensation’), which can be triggered by various stressors, from prolonged fasting and physical exercise to vaccinations and common infections. And they will be constantly reminded that the slightest mistake on their part—sleeping through a night feeding or bringing the sick child to the hospital a few hours too late—can lead to a health crisis ending in death or irreversible disabilities. Families will also need to learn how to navigate the healthcare system, deal with a broad range of medical specialists, place orders in specialised pharmacies (which sell medical foods), fill out paperwork for health authorities (who will approve—or not—the supply and reimbursement of those foods), and unravel the intricacies of disability welfare systems. As a result, they will systematically take the disease into account when making almost every decision, from the most important—where to work and live, what school to send their children to—to the most mundane—how to spend their holidays, what to offer guests at a birthday party, or how to prepare their child for a sleepover at a friend’s house.

In this Research Article, I focus on the knowledge practices, skills, and roles that Polish and Swedish parents of children with IMDs develop, acquire, and share in the process of learning to live with their child’s metabolic disorder. I will show that

1 All personal names and toponyms have been altered.

the parents perform numerous roles and tasks and develop a multifarious ‘parent-patient’ knowledge within a vast network comprising clinical professionals and a biosocial community (Rabinow 1996) of peer-parents. I will also analyse the concepts and metaphors that parents and doctors (as well as anthropologists) use to describe the roles, tasks, and knowledge of caregivers of children living with chronic conditions and disabilities. I will argue that the prevailing metaphors of professionalisation used to describe parental roles, while contributing to the acknowledgement of these caretakers’ unique expertise, also overly stress an individualistic perspective and obscure the relationality of care, collaboratively achieved between patients, relatives, and healthcare providers.

A study among people living with inherited metabolic diseases

This article draws on an ethnographic study conducted among Polish and Swedish families, clinical staff, and persons otherwise engaged with rare metabolic disorders. Particularly, this research focuses on families living with, and professionals treating and caring for, a variety of fatty acid oxidation disorders (or FAODs), including those known by the acronyms MCADD, LCHADD and VLCADD, as well as organic acidemias (including glutaric, propionic, isovaleric, and methylmalonic acidemia), and maple syrup urine disease (MSUD)[2]. These diseases pertain to the biomedical category of inborn errors of metabolism, which encompass a vast group of rare, genetic, chronic, often disabling, and possibly lethal conditions (1583 disorders have been identified at the time of writing this article)[3].

Each of these disorders consists of an enzymatic deficiency caused by a genetic mutation that disturbs the process with which the body metabolises fat, in the case of fatty acid oxidation disorders, or amino acids, in the case of organic acidemias and MSUD (maple syrup urine disease). This translates into two kinds of issues for the person affected and their body. Firstly, the deficiency of enzymes causes an excess of unprocessed chemical compounds which are often toxic and may lead to the permanent damage of internal organs, including the brain. Secondly, there is a shortage of the products of disrupted reactions (mostly energy), which may result in, among others, the risk of a life-threatening crisis, commonly referred

2 The common names of fatty acid oxidation disorders are acronyms referring to deficiencies of specific enzymes. For instance, LCHADD stands for ‘Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency’. The common names of acidemias refer to acids whose urine concentrations are elevated in affected patients. Maple syrup urine disease (or MSUD) receives its name for the characteristic smell of the urine of those affected. When I do not specify the name of the disease in the text, I do it in order to protect the privacy of my research participants.

3 See www.iembase.org for more details. In the European Union, a disease is considered rare if it affects no more than one person in 2,000, and in the US, when it affects fewer than 200,000 people. Although the category of rare diseases has emerged in public discourse and health policy to highlight common problems faced by patients, their caregivers and healthcare providers, it remains an extremely heterogeneous group of diseases, with many subcategories presenting unique configurations of challenges due to their aetiology and treatment options.

to in biomedicine (and among patients and their relatives) as ‘metabolic decompensation’ (Saudubray, Baumgartner, and Walter 2016).

As of yet, no treatment available can cure these diseases. The symptomatic treatment available mostly relies on: a dietary regimen (involving frequent feeding to avoid prolonged fasting, elimination of several common foodstuffs, and the use of special medical foods or supplements), activity management (avoiding excessive exertion), and monitoring of the child’s condition in order to make a timely decision on hospitalisation. A constant element in the patients’ and caregivers’ experience is an alternation of periods of stability and periods of exacerbation (due to infections and other health stresses). The treatment also involves regular check-ups in the metabolic ward or outpatient clinic. Sometimes, biomedical feeding technologies are used such as the NG-tube (i.e., nasogastric intubation, a flexible tube that is introduced through the nose into the stomach), or PEG (i.e., percutaneous endoscopic gastrostomy, in which a feeding tube is placed through the abdominal wall into the stomach). The use of PEG depends on the treatment models used in various countries. In Sweden (and Finland), PEG is a fairly standard procedure for cases of LCHADD and VLCADD, whereas in Poland it is viewed as an intervention of last resort (Rajtar 2017). In some organic acidemias and in MSUD, a liver and/or kidney transplant may be performed. All disorders covered by the study (and a number of other IMDs) are included in newborn screening panels in Poland and Sweden (since 2014 and 2010, respectively). This means that the majority of people affected with these disorders in these countries are diagnosed within the first weeks of life, which spares them of a ‘diagnostic odyssey’—a common problem for people living with other rare diseases—and allows for the early introduction of adequate treatment. In many cases this may radically limit the impact of the disease. Nevertheless, despite all the health benefits of early diagnosis, living with these disorders inevitably involves the management of several metabolic risks.

My ethnography is part of the collective research project ‘*An Anthropology of Rare Diseases. A Study of the Baltic Sea Region*’, which focuses on families living with fatty acid oxidation disorders, organic acidemias, and maple syrup urine disease (MSUD), in Poland, Sweden, and Finland.[4] Within the context of this project, I conducted ethnographic fieldwork in Poland and Sweden. The data were gathered through interviews, participant observation (of in-person and online workshops for families and professionals), and content analysis of Facebook groups and blogs. Between December 2018 and January 2022, I interviewed 76 people. These included patients and families (parents of children or dependent adults) and

4 The project focused on these countries because some of those diseases seem more prevalent in the Baltic Sea region (see Rajtar 2019, 94).

professionals (i.e., metabolic paediatricians, dietitians, geneticists, psychologists, biomedical scientists, and activists). A total of 29 patients and/or families and 16 professionals participated in these interviews in Poland, and 7 patients/families and 24 professionals in Sweden. Interviews held during and after the outbreak of the COVID-19 pandemic in March 2020 were conducted remotely. The families were contacted mostly through social media (Facebook groups) and through referrals from parents who had already taken part in the study (using the snowball sampling technique). A few parents were contacted during an anthropology conference on rare diseases that took place in Poznań, Poland, and through the intermediary of healthcare staff. Professional participants were contacted mostly through their institutional or organisational emails.

During my fieldwork, I sought to meet patients and families living in diverse health, social, and cultural situations. Nevertheless, the final composition of my research participants unevenly reflects the diversity of possible situations. I met many more Polish families than Swedish ones, but many more Swedish professionals than Polish ones; significantly more families living with disorders of metabolism of fats (fatty acid oxidation disorders) than those living with disorders of metabolism of proteins (organic acidemias and MSUD); far more parents than patients (I interviewed only two adult patients); many more families with small children and toddlers than families with older children and teenagers;[5] and, among the parents, many more mothers than fathers.[6] Despite these limitations in scope, I hope this research will contribute to a better understanding of the practicalities of living with inborn metabolic diseases.[7]

Patients’ and caretakers’ roles, tasks, and knowledge

During the last few decades, the experience of living with chronic diseases or disabilities has been a focus of sustained interest within medical anthropology and related disciplines (see, e.g., Heurtin-Roberts and Becker 1993; Manderson and Smith-Morris 2010; Ginsburg and Rapp 2020). This scholarship includes vital research on parenting and caring for disabled and chronically ill children (Sobo 2010; Landsman 2009; Mattingly, Grøn, and Meinert 2011; Rapp and Ginsburg 2011; Trnka 2017) which has produced a vast array of concepts to describe

5 I intended to interview also the affected children. Unfortunately, when the outbreak of the COVID-19 pandemic forced me to continue my research remotely, I opted to forgo interviewing the children. I did not feel confident to build a relationship of trust and to rely on computer-mediated communication in assessing the child’s well-being during the interview.

6 Mothers are the main carers of children with disabilities and chronic illnesses (Landsman 2009). While this seems to be the case for parents of children with IMDs too, during my research I also met fathers who were very involved in the care of their children.

7 Although healthcare systems, organisation of treatment and care, as well as parents’ attitudes and experiences related to metabolic diseases may differ significantly in Poland and Sweden, this article is not intended as a comparative analysis. Most of the facts presented are common to both ethnographic contexts. Where this is not the case, it is explicitly stated in the text. I develop a contrasted analysis elsewhere (Rogalski forthcoming).

patients’ and caregivers’ roles, skills, and knowledge. These concepts circulate in academic literature representing different disciplines and approaches, but are also present in public discourses and are often drawn from the patients’ and caregivers’ own notions of their experiences. On the one hand, in the social science literature, parents’ and caregivers’ roles and tasks are often described as a kind of ‘work’. A prominent example is Sobo’s study (2010) on how the parents of children with special healthcare needs master care-related roles and responsibilities. She identifies, for instance, an ‘instrumental work of care coordination’ that involves ‘knowledge management’ (2010, 220, 225). She also describes the expressive, symbolic and emotional ‘culture and social work’ of parents and caregivers, which consists on reframing the child’s condition as something positive (for instance, labelling the child as exceptional or special), or raising awareness about disability (idem, 224–6). Another oft cited and related notion is that of ‘chronic homework’, which was introduced by Mattingly, Grøn, and Meinert (2011), who define it as ‘the work patients and families are expected to carry out in their home contexts as part of the treatment of chronic conditions’ (idem, 347). On the other hand, scholars often underline and characterise the specialised knowledge that parents develop while caring for their children with chronic diseases and disabilities. There is extensive literature that highlights the unique expertise that arises from living with a chronic illness or disability (see, e.g., Akrich 2010; Pols 2013; Callon and Rabeharisoa 2003). In this respect, a vast number of authors use the notion of ‘experiential knowledge’, that is, a truth ‘learned from personal experience with a phenomenon rather than … acquired by discursive reasoning, observation, or reflection on information provided by others’ (Borkman 1976, 447; see also Blume 2017; Baillergeau and Duyvendak 2016).

Through an overview of roles, tasks, and knowledge of parents of children with IMDs, I will show that they develop unique expertise, combining biomedical and lay knowledge. Thus, they do not merely apply treatment principles taught to them in the clinic (thereby challenging the notion of ‘homework’ (cf. Mattingly, Grøn, and Meinert 2010) carried out at the dictates of the teacher-clinician). I will also show that the parents themselves often describe their roles using the ‘professional metaphor’ and identify various kinds of work involved in caring for their children, as well as stressing particular expertise that they gain in the process. At the same time, I will argue that although the ‘professional metaphor’ used to describe parental roles as well as the emphasis on unique parental expertise capture certain aspects of parenting a child with a metabolic disease well and help legitimise parents’ roles and knowledge, this may put excessive emphasis on parents as individuals. Following Trnka (2017), who in her critical analysis of the rhetoric of self-management and responsibility associates this kind of rhetoric with a neoliberal ideal of autonomy, I will argue that focusing on parents as professionals of care risks obscuring the ‘inherent interrelationality of healing’ (idem, 7) and the

fact that care is always collaboratively produced by patients, caretakers, and healthcare providers.

Parents involved in treatment: Between the home and the clinic

Learning about the diagnosis of metabolic disorders requires the parents to assimilate a rich set of representations concerning fundamental aspects of corporeality and personhood. That knowledge is ‘difficult to take in all at once’, as Emilie, mother to Stine, an adult woman with LCHADD (in Sweden), put it in our interview. If during the first months following the diagnosis, metabolic paediatricians regularly summon the diagnosed child and their parents to the hospital (at the beginning, every fortnight), it is not only in order to check how the child responds to dietary treatment (‘how she stabilises’), but also to make sure that the parents grasp fundamental notions about aetiology and rules of treatment. Throughout those educational hospital stays, the parents are acculturated (Sobo 2010) into a biomedical conception of their child’s body and the most basic relations linking them with their material and social environment. While they see their child weighed, measured, counted from different angles and converted into a body functioning first and foremost in terms of energy consumption (cf. Yates-Doerr 2017), the parents are exposed to the discourse of biochemistry, metabolism, and nutrition. This discourse is made accessible through ‘industrial’ metaphors, descriptions of the body as factory (Landecker 2013) or metaphors referring to logistics or communication, where chemical compounds move from one biochemical reaction to another. The parents are also taught about Mendelian inheritance and genetics, and learn that they both carry a mutated gene which they have transmitted to the child. As a result, the diagnosis introduces a notion of genetic risk into their kinship networks (Novas and Rose 2000) and reshapes the fabric of kinship (Rapp and Ginsburg 2011).

Since the treatment of inherited metabolic diseases consists of diet, activity management, and special attention to the health condition, the parents play a crucial role in it. As they learn to perform the treatment, they acquire and develop knowledge that does not fit within conventional categories of ‘lay’ and ‘certified’ or ‘biomedical’ and ‘experiential.’ From the very beginning, there is an understanding among the parents that they are acquiring a particular experiential knowledge that the doctors do not have. The doctors themselves highlight the value of parental knowledge, since they often orient new parents to parental Facebook groups (cf. Herbst 2016) or arrange meetings between them (see Rogalski 2022). They also involve more experienced parents in the training of medical students. For instance, Patrycja, mother of Gaia (age 3, Poland), with isovaleric aciduria, says proudly that she always allows visits from medical students at the metabolic ward—otherwise,

‘who will treat those children later?’ She instructs the trainee doctors on ‘what [a metabolic disease] looks like in everyday life, not only in the bookish life. Books are important. You can read about things in them, but the daily life of such a child and her family, that is a completely different story’. Hence, the parents develop particular expertise, seen by both doctors and parents as legitimate and necessary in complementing ‘bookish’ biomedical knowledge.

But there is more to parental knowledge than just a lived experience of the disease. The parents also acquire biomedical knowledge and clinical expertise. At some metabolic wards, they are taught about the metabolic diseases together with medical students. For instance, Dr Grabowska (head of a Polish metabolic team) arranges teaching sessions where metabolic specialists explain metabolic diseases to both the parents and medical students. She also explicitly compares the education of parents on the ward to formal medical training—when parents of newly diagnosed children ask her when their children will be allowed to go home, she jokingly replies: ‘When you pass the exam on this disease with me’ (see more in Rogalski 2022).

Apart from the theoretical knowledge about the disease, parents acquire practical clinical nursing skills, such as, for instance, putting a NG tube in a baby—some of these parents even teach these skills to parents of newly diagnosed children. Also, through observing doctors and nurses perform various tasks (inserting a cannula, regulating a glucose drip, etc.) the parents gain a sort of passive knowledge about biomedical routines. They are neither explicitly instructed how those tasks should be performed, nor are they expected by the healthcare staff to be knowledgeable about them. The value of such knowledge is often foregrounded by the parents in narratives that recount antagonistic encounters with healthcare staff:

> [My daughter Stine] was probably two or three years old, and we came with her [to the hospital] […] we had that paper [from the metabolic ward], and on that paper, there was a short description of her diagnosis and what was needed to be done in case of emergency, and that it had to be done without delay, it had to be done now. And there was this doctor […] who did not seem to understand what he read … He just read, and he read, and he read, and he read, time and time, and time again, and at last he said: ‘so, it says here that she should have a glucose drip’, [and I said:] ‘yes, so put it in’. (Laughs). […] And then the doctor calculated how many drops she should have, and then I came back [later], and I looked at the drop counter and I saw that it was not right. So, I talked to one of the nurses who seemed good to talk to, and the nurse looked at the drop counter and she also said that it looked weird. And then I said, ‘bring in a doctor who knows something’. (Laughs). So then one of the regular doctors that we had a lot of contact with before came in, and he too

> looked at the drop counter and said: ‘this is wrong.’ [And I said:] ‘Yes, thanks, I know it’s wrong’. (Laughs) (Emilie, mother to Stine, adult woman with LCHADD, Sweden).

Emilie’s stressful experience, recounted in vivid detail many years after it happened (her daughter was turning eighteen when I met her), is typical of encounters with non-specialist healthcare services. The physician who admitted her daughter clearly had no prior experience treating patients at risk of metabolic decompensation, and even the formal, written note from the metabolic clinic was not enough to assure the child proper treatment. The mother had to find ways to alert the healthcare staff—find an approachable nurse and get another doctor to see her daughter. Emilie summarised the challenges and skills necessary in dealings with healthcare actors further in our interview:

> You must learn how to talk to doctors, how to talk to nurses, to question things, to ask ‘do you really know what you are doing?’ And, to pay attention, well, if they put a cannula, a needle, and you think ‘this needle is not long enough,’ you really need to keep track of that […]. Regardless of the time of day.

Another area where some parents become significantly involved in the clinical aspects of treatment concerns the child’s diet, and the activities related to monitoring it. The treatment of metabolic diseases requires parents to keep track of their child’s diet: parents should avoid giving their child certain foodstuffs, use special medical products and various substitutes, count calories and certain nutrients, etc. In general, though, the doctors and dietitians do not expect them to invest too much time and effort in this task. Typically, after the initial period when parents are asked to take notes on every meal, the doctors will only ask them to provide a detailed description of what the child ate in the days leading up to the follow-up appointment. Nevertheless, some parents, for various reasons, maintain extremely detailed food-records, and—as one case illustrates—this effort can have a clinical relevance.

Olle, father of Felicia (toddler diagnosed with glutaric acidemia type 1, GA1, Sweden) has been closely monitoring her diet, ever since she started on solid foods as a baby. He uses an elaborate set of Excel spreadsheets, which he has programmed himself, and a meticulously compiled database of foodstuffs coded in terms of calories and protein intake (with separate values for lysine, which is the amino acid that people with GA1 cannot efficiently metabolise). For Olle, keeping a record of a meal involves copying a product from the database, pasting it into a spreadsheet tab corresponding to the current day and recording the amount that the child had eaten in grams (calculated with an electronic kitchen scale). The spreadsheet is then set up to calculate the calories, protein, and lysine, and then add them up in a different cell. Then, another spreadsheet returns average values

and visualises the tendencies on four charts. Interestingly enough, Felicia’s doctors and dietitians—who at first viewed Olle’s detailed record-keeping as something that helped him cope emotionally with his daughter’s disease—with time became interested in his data. Today they rely on it for very fine-grained adjustments of lysine intake. The last time we spoke, Olle said that they were thinking about augmenting her lysine daily intake from 1100 to 1200 milligrams—an adjustment that would have been impossible to envisage without Olle’s meticulous record-keeping. Olle’s story shows how being an ‘Excel parent’—as Swedish doctors and dietitians call such overscrupulous parents—in the end may prove useful and allow for the implementation of a more tailored dietary treatment.

Dealing with emergencies

It should be clear now that parental roles go far beyond the mere practical application of treatment regimens, and that parents gain genuine biomedical knowledge and clinical expertise, which come to play a crucial role in the quality of treatment their children receive. But the parents’ encroachments into the clinical domain may give rise to tensions between them and healthcare professionals, as I will develop in this section. This becomes evident in the ways that parents deal with their children’s health emergencies. I am going to discuss this issue in detail as it sheds an important light on parental roles and their metaphoric expressions.

Living with IMDs often entails risk of sudden deterioration (termed ‘metabolic decompensation’), when the affected person requires urgent hospitalisation. From a clinical perspective, providing an emergency treatment to a ‘decompensated’ child is not difficult—it consists mainly in rapid administration of a glucose drip (though much more concentrated than in common health crises). Sometimes, though, if a hospital has no previous experience with metabolic diseases and is therefore reluctant to admit the child, then the parents must try any means at their disposal to obtain the treatment that their child needs. In such cases, they may produce written information from the metabolic clinic, which is as much a source of information for the emergency physician as it is a document attesting to the parent’s competence and authority. As a last resort, they can call their metabolic paediatrician and give the phone to the on-call physician. But any disagreements with hospital staff take time and the child’s condition may deteriorate extremely fast.

When reflecting on these instances of confrontations with healthcare staff, the parents often objectify their knowledge and role—that is, they give expression to their experience in a form that can be understood by others—primarily by employing the metaphor of professionalism. For instance, Emilie (quoted in the previous section) summarised her experiences by saying: ‘you become pretty

much a healthcare worker’ (Swedish: *man blir ganska mycket vårdpersonal*). The quasi-adoption of professional biomedical roles by parents has been reported in the medical anthropological literature on parents and caregivers of children with chronic or disabling diseases. To cite but two examples, a mother interviewed by Trnka and McLauchlan, reporting that she was self-medicating her daughter (living with asthma) with antibiotics, described herself as ‘half a doctor’ (Trnka, McLauchlan 2012, 13); while one of Landsman’s research participants, a mother to a girl with a chronic disability, reported that when she argued with hospital staff over medical decisions they had made, ‘one doctor said to me, “you’re not a nurse”. I said, “I am *now*”’ (Landsman 2009, 101, emphasis in original). What is interesting in the case of caring for children with IMDs is that the metabolic paediatricians themselves will often describe the parental role in terms of biomedical professionalism, as I will show below.

In fact, metabolic clinicians are aware of the difficulties that the parents meet in situations of emergency when confronting non-specialist healthcare services. Apart from investing considerable effort in educating general paediatricians and other specialists about metabolic diseases[8], they also go to great lengths to prepare the parents for encounters with doctors, nurses, and paramedics. Such efforts often involve describing parental roles in terms of biomedical professionalism. A prominent example is a metaphor used by Dr Piotrowski (Poland), as recalled by Kamil, father of Ada (age 2, with MCADD, Poland):

> [The doctor] convinces us that […] we are part of the lifesaving team (Polish: *zespół ratowania życia*). This means that if there are a doctor and paramedics present, then we are included, we are not bystanders who drive after the ambulance. No, we need to be inside the ambulance, in the hospital ward, and we have to instruct, because an internist or a paramedic, they don’t know these diseases. […] [He says] ‘Under no circumstances should you stand aside. Be aware that you are part of this team, and you should instruct [the doctors]. And the doctor is supposed to listen. You don’t need to tell them how to do specific lab tests […]. No, you are there to say that such an [intravenous] infusion is needed. If you learn the formula of how much glucose is needed per weight, that is great, and the doctor must do it and that’s all’.

Encouraging the parents to actively participate in their child’s emergency medical care, Dr Piotrowski portrayed them as part of a ‘life-saving team’ (along with physicians and paramedics), and conferred on them a directive role in the team, based on their biomedical knowledge of the metabolic disease. Although the notion of a ‘life-saving team’ does not refer to any official unit of emergency healthcare, it

8 They visit paediatric wards or organise workshops for specific groups of healthcare professionals, for instance paediatric neurologists.

does, however, echo the notion of ‘emergency medical teams’ (*zespoły ratownictwa medycznego*), which are units of the Polish emergency medical services. In a similar vein, during an online workshop for parents of children with metabolic disorders, Dr Piotrowski described parents as being part of the healthcare personnel. Concluding his presentation of the treatment regimen, he said:

> ‘You are of great help. Because in fact, the team treating the patient with a [metabolic] disorder is not only a doctor, dietitian, psychologist, nurses, the entire laboratory, and all those people in biochemistry, but most of all, [it includes] you, the parents. You as parents are an integral part of that team’.

These are good examples of how healthcare professionals may themselves legitimise parental roles and knowledge as part of formal, systemic, biomedical care. They suggest a strong alignment between parents and doctors of metabolic diseases when it comes to confrontations with non-specialist healthcare staff. However, the metabolic specialists do not always approve the parents’ incursion into the clinical/biomedical domain. This happens, for example, when parents—in an effort to decide whether their child needs to be admitted to hospital or can be treated safely at home—order specific laboratory tests that are normally available to use by doctors. In fact, despite being aware of the various symptoms to look out for, the decision to take the child to hospital is always a matter of worrying uncertainty for parents. The seemingly simple rule of treatment—‘if anything strange happens, come to the hospital’—proves difficult to apply in practice (see more below). Moreover, the burden of the decision rests entirely on the parents. On the one hand, if the parents go to the metabolic ward, the doctors, regardless of the outcome of a cursory examination of the child’s condition, will most probably admit them for a few days. On the other hand, if the parents go to a local hospital, where the staff is not familiar with the child’s metabolic condition, they are expected to insist and have their child admitted. As a result, for the parents, taking their child to the hospital is tantamount to having them admitted.

For as much as parents are worried for their children’s health, none of them want to spend a week in the hospital if it is not necessary. Faced with this predicament, the Polish parents found a solution that is illuminating when it comes to defining parental roles in terms of the opposition of ‘lay’ versus ‘clinical’. When in serious doubt as to whether their child should be hospitalised, many parents will go to the hospital or a regular diagnostic lab and order a test that measures creatine phosphokinase (abbreviated as CK or CPK) levels in the blood. Here, it is revealing

that these parents order it in a 'wild' manner (cf. Callon and Rabeharisoa 2003)[9]: independently, without a doctor's referral, and for their own purposes. For many of them, the CK test is a regular item within their care decision flowcharts, as the one described by Małgorzata, mother of Krzyś (age 1.5, with LCHADD, Poland):

> If Krzyś has a runny nose but is eating well, then we know that he can handle it; when a cough starts or when there is something more, an infection, then, for instance, on the next day or two days later, we check if the CK is not rising, but if, for instance, there is fever or, God forbid, vomiting, then we go directly [to the ER].

In general, doctors seem to approve of the fact that parents do CK tests on their own. Anna, mother to Leon (age 11, with a fatty acid oxidation disorder, Poland), when asked if her doctor was aware that she performed the CK test on her son, said: 'I have talked it through with him and he trusts me. He knows that I won't do anything that could make my child's condition worsen, and he knows that I am doing these tests'.

The point at which the 'wild' creatine kinase testing clashes with doctors' recommendations, however, is when parents, rather than relying on the standard lab definition of elevated levels of CK, set their own cut-off levels based on what they think is appropriate for their child. These values are discussed within Facebook groups founded by parents of children with metabolic diseases. During our interview, Dr Grabowska (Poland) praised the 'wild' CK testing in principle ('Very good, this is what they are taught for', she said) but sharply criticised parents who challenge the official thresholds. She evoked a notion of responsibility ('Who then takes responsibility for this child?') and referred to each child's clinical singularity ('Each child is different, each child reacts differently, each has a different growth dynamic of CK concentrations'). Interestingly, on the last point, she implicitly agreed that CK levels can differ for individual children. Thus, she did not question the threshold adjustment itself, only the parents' competence to do it. This is a compelling example of how a parent-patient community may appropriate a particular biomedical technology and how 'experience-based expertise' can become blurred with the 'certified expertise' of scientists and clinicians (see Akrich 2010; Pols 2013, 77). It also shows that self-management and responsibility may be differently understood by parents and physicians. 'Wild' CK testing is certainly a sign of parental responsibility and proactive management of a child's illness, but

9 In their research on a French patient organisation, Callon and Rabeharisoa (2003) coined the expression of 'research in the wild' to characterise a particular mode of collaboration between scientific and non-scientific communities. In contrast, I describe CK testing on parents' order as 'wild' because it is done independently from clinical advice.

doctors expect parents to limit themselves to performing the test and claim exclusivity in interpreting the results (cf. Trnka 2017).

The professional parent at home

As I have shown above, the role of parents in the treatment of their children living with IMDs involves acquiring considerable biomedical knowledge and developing genuine clinical expertise. But even the most mundane tasks involved in parenting a child with an IMD constitute an important area of parental expertise, in which the role of the parent cannot be reduced to a mere top-down application of treatment rules taught at the clinic. Here too, as I will show, the parents tend to objectify their competencies (that is, to make them more tangible for themselves and for others), by drawing on the metaphor of professionalism. First, I will show in more detail how the parents apply and elaborate on the rule laid down by the clinic on how they should pay attention to symptoms of exacerbation, and then I will provide an overview of the practices involved in observing the diet of children with IMDs and adjusting their treatment to their everyday life.

At the clinic, the parents are told to pay attention to multiple kinds of signals that may indicate an upcoming metabolic decompensation. Some are self-evident signs of illness, such as fever, vomiting, lethargy, lack of appetite, diarrhoea, and so on. But while in a healthy child those symptoms justify hospitalisation only if they persist for a few days, the child with an IMD should be taken to the hospital as soon as they appear. ‘The margins are narrow’, as a Swedish mother to a girl with LCHADD said in an interview. Then, there are also symptoms specific to each disorder. For instance, in children with LCHADD or VLCADD, leg pain is an index of rhabdomyolysis—a dangerous condition in which the body breaks down muscles to obtain energy—whereas in disorders of amino-acid metabolism parents pay special attention to problems with motor coordination. These symptoms are abstracted from the clinical experience or are identified by the parents who, over time, find their own indexes of their child’s condition. Parents may be sensitive to olfactory cues of metabolic imbalance. For instance, in the maple syrup urine disease (MSUD), the very name of the condition is motivated by a characteristic urine odour, which the Polish parents also compare to Maggi, a popular seasoning sauce. In isovaleric acidemia, the metabolic imbalance manifests itself through the smell of sweat, which is likened to the odour of ‘sweaty feet’, or cheese (see e.g., Saudubray, Baumgartner, and Walter 2016, 8; Gick n.d., 6) or ‘sour milk’ (according to Stanisław, an adult living with isovaleric acidemia, Poland). The parents of children with MCADD report a fishy-smelling sweat, according to posts on a (Polish) Facebook group. Apart from the smell, the parents are sensitive to scores of other symptoms. The Polish parents of children with acidemias or MSUD determine that their child is ‘disturbed’ (*zaburzone*)—as they

term the metabolic imbalance—when they notice stumbling or changes in mood. For parents of children with LCHADD or VLCADD, the fact that their baby ‘pours herself’ (Polish: *leje się*)—i.e., is so weak that her body seems to pour between the parent’s hands—is a clear signal that they need to take them to the hospital. (Dr Norén, from Sweden, with whom I spoke about this expression described the clinical condition as ‘loss of muscle tone’.) The parents also develop a sort of embodied perception and simply ‘see in their child’s eyes’ (Polish: *widzę po oczach*) that they are unwell. As Anna, mother to Leon, age 11, with a fatty acid oxidation disorder (in Poland) said:

> ‘If you have a small baby and he has a fever, you can’t tell if he’s sleeping or maybe he is already falling into a coma, because the blood sugar drops. So, I pricked him all the time and checked his blood sugar. Now, I [simply] look at him and I know if his values are high or not’.

This semiotic vigilance displayed by parents is a deeply reflexive activity, for the relationships between the symptoms and a child’s health condition are seldom one-to-one. The leg pain in children with LCHADD or VLCADD need not be an index of rhabdomyolysis because a child’s legs may hurt for other reasons, for example, after physical activity or because of a growth spurt (so-called ‘growing pain’). Also, some symptoms are not always self-evident—the parents may be in doubt about their own senses, particularly when it comes to identifying odours. The picture is further complicated as some children may not present clear symptoms even when the lab tests indicate a metabolic imbalance.

In order to describe this heightened attention to the child’s condition, Agnieszka (mother to Marta, age 10, with LCHADD, Poland) uses the term ‘monitoring’ (Polish: *monitorowanie*). Another expression she uses is ‘doing an interview’ (Polish: *robię wywiad*, which also translates as ‘taking a medical history’). She uses this term when she talks about determining whether worrying symptoms in her daughter are signs of metabolic imbalance:

> Let’s say that she [my daughter] returns from school and says ‘my back hurts’. Then immediately my stomach tightens. Ok, her back hurts because she had a PE lesson, did flips, maybe it’s muscle soreness, that’s possible, or she starts having muscle aches, and then you start to think. [So] first, I do an interview: ‘do your legs also hurt, or is it only your back? maybe your backpack was too heavy?’ And I wonder if I should go and take the CK [creatine phosphokinase] test, or if I can wait a bit longer.

The difficulties and uncertainties related to the rules of treatment and care are not limited to this watchfulness over the child’s condition. Even the simplest rules related to the diet—although presented and explained in educational booklets

prepared by metabolic centres or published by manufacturers of special medical foods—prove difficult when it comes to applying them in everyday life or aligning them with other ‘matters of importance’ (Pols 2013, 87; see also Risør and Lillevoll, 2021), such as school education, professional life, sports, outdoor activities, celebrations, or holiday trips. In fact, preparation of everyday meals is an area of constant parental innovation. Although the diet regimens in the treatment of IMDs involve the use of special medical foodstuffs (such as MCT-oil, amino acid-based formulas, etc.), for the most part they consist of regular meals (controlled in terms of nutrients and scheduled in order to avoid prolonged fasting). The need to follow strict schedules and nutritional charts makes mealtimes particularly stressful and IMD Facebook support groups are replete with posts of desperate parents asking for advice about children who refuse to eat. In the case of a healthy child who is a picky eater, for example, a parent can indulge them, allow them to skip a meal, or resort to some proven foods that the child eats happily. In the case of a child with a metabolic disorder, however, dietary compliance is a matter of life and death. The parents of these children thus systematically explore new foods, invent new dishes, or modify existing recipes to meet the requirements of IMD diets. Such tinkering—as the parents themselves often describe their efforts (Polish: *kombinować*, Swedish: *att mixtra*)—may seem of little significance and elicit little prestige (in contrast, for instance, to special medical foods developed scientifically and industrially). Nevertheless, the efforts of these parents represent a particular epistemological orientation, one that is aimed at achieving desired goals with the limited material at their disposal (bringing to mind the classic Lévi-Straussian (1962) notion of ‘*bricolage’*). The parental bricolage often goes hand-in-hand with a kind of substitutive ontology. For instance, *mizeria*, a Polish cucumber salad normally made from thinly sliced cucumbers mixed with dairy cream and dill, is not suitable for children with LCHADD because of its high-fat content. But parents may substitute 0% yoghurt for cream and obtain an ‘LCHADD-version’ of *mizeria*. A good part of the foodstuffs they use (prescription medical foods or regular market products) are thought of as ‘substitutes’ (Polish: *zamiennik*, *zastępnik*; Swedish: *ersättning*) for common foodstuffs or meals (for instance, special formulas with a specific fat or amino-acid composition are substitutes for common infant formulas, and MCT-margarine is a substitute for butter). Such substitutes are often labelled as ‘versions’ qualified through reference to their nutritional characteristics (e.g., low-fat or low-protein), the eliminated ingredient, or the disease / patient target group. This kind of labelling carried out by parents and families yields trophonyms (i.e., terms designating food) such as ‘cheese-free cheesecake’ (Polish: *sernik bez sera*, note the expression’s oxymoronic character), ‘LCHADD chicken nuggets’ (in Swedish), or ‘pancakes for lchadders’ (Polish: *naleśniki dla lchadków*—where *lchadek,* plural genitive: *lchadków*, literally ‘little LCHADD’, designates a child affected with LCHADD disease).

The parents also constantly strive to make various activities possible for a child with an IMD, for instance, airplane travel or outdoor sports. When planning vacations, parents note the location of the nearest hospitals (elaborating a sort of mental emergency map), devise and prepare high-energy snacks and drinks that enable the child to stay outdoors and do sports for a long time, adjust timetables to schedule more frequent breaks, or manipulate feeding tubes for easier feeding outdoors. For instance, the parents of Agnete (age 11, with LCHADD, Sweden) have found a way of feeding her when they go skiing in the Alps: they move her feeding tube up her body so that it rests close to the collar of her ski suit, making it easier to inject her with a high energy mix (of their own recipe, based on MCT-oil, a quick-acting sugar and corn starch), which they do every hour or 30 minutes. This makes it possible for the family to stay outside and ski all day long.

It is important to note, however, that parental bricolage is also a political effort to deal with the disease. As Agnete’s mother, Natasha, puts it: ‘We have always tried to find ways around that disease, so it does not define [our daughter]’. For parents, then, looking for practical solutions to everyday problems is thus related to the very definition of the child in terms of notions of illness and normality and their identity as a ‘sick’ or ‘well’ child. It is thus a site of micro-activism—i.e., daily, meticulous, and grass-root acts—for social inclusion (Dokumaci 2020).

Finally, the care of a child with IMD also entails bureaucratic and social skills. These may be necessary to ensure the child a safe stock of special medical foods (which in Poland is particularly bureaucratised and unpredictable), or enforce their entitlement to social support (disability status with full care allowance, in Poland, or targeted benefits and allowances, in Sweden). Referring to her struggles with the social welfare system, Emilie said: ‘There are other things too, not only LCHADD. You also gain knowledge about how health care works, you gain knowledge about how government works (laughs), how legal processes work ... yeah’. Moreover, since successful care for a child with IMD requires the involvement of communities, parents also develop important networking skills*:* they educate relatives and broader communities about the disease and its treatment. When the child starts kindergarten or school, for example, parents explain the disease and its treatment to directors, teachers, kitchen staff, parents of other kids, etc. Here too, the parents objectify their tasks through metaphors of professionalisation. For instance, Agnieszka uses the metaphor of ‘bookkeeping’ (Polish: *prowadzić księgowość*) to describe keeping track of special foodstuffs and doing the paperwork needed to order them, and talks about ‘good cooperation’ with specialty pharmacies, employing the language of business relationships (Polish: *dobrze się współpracuje*).

The power of words and the relationality of healing

Parents and doctors systematically use metaphors of professionalisation, drawing from worlds of legitimate, established, and socially-valued activities. We can see this when Dr Piotrowski refers to the parents as a ‘life-saving team’, when Dr Grabowska mentions a ‘final exam’ for parents after teaching them alongside medical students, or when Agnieszka’s uses the word ‘bookkeeping’ to refer to the paper-work involved in care provision for their child. Other metaphors allude to scientific activity, such as when parents say that they ‘experiment’ with some solutions to problems encountered while caring for their child. Their terms also take on the language of supervisory, technocratic and bureaucratic work, for example, when they refer to solving childcare problems as ‘finding solutions’ (Polish: *znajdowanie rozwiązań*) or organising parental care as ‘setting up a system’ (Swedish: *sätta ett system*). When Agnieszka speaks of a ‘good cooperation’ with a pharmacy, she is alluding to the language of business and networking. At first, one may see here a conceptual effort to legitimate parental knowledge and expertise, by showing its extension and similarity to socially sanctified roles and practices. But I argue—following Rodinelly Medeiros’s (2019) work on alternative farming practices in northeast Brazil —that such metaphoric notions, rather than being mere descriptions detached from practice, are in fact ‘lexical-practical tools’ (idem, 23) playing a crucial role in the everyday life of families. I understand these as concepts that have a real impact on the lives of the people who use them and help them to transform the world around them. That concepts have an active role in the treatment and care is evident, for instance, when a doctor convinces parents that they are part of ‘the life-saving team’. The use of such depictions may be a powerful and decisive tool, encouraging parents to insist—when the time comes—on their expertise, to check if physicians’ decisions correspond to the regimen of metabolic crisis management and, if needed, to question them and effectively demand proper treatment. The parental metaphors may also have a practical leverage, even in performing more mundane tasks. For instance, the notion of ‘bookkeeping’, used to refer to keeping track of special foodstuffs, allows parents to objectify any activities related to the ordering of those products. These include monitoring supplies, anticipating when they could run out, remembering to order them well in advance, keeping track of the dates of check-up visits to ensure the doctor fills in a special form in a timely fashion, as well as checking if all other necessary forms are duly filled in, for instance. Furthermore, it also allows the parents to activate skills and qualities commonly associated with bookkeeping—such as regularity, accuracy, attention to detail, respect for forms, valorisation of the paperwork, etc. By using metaphors of professionalism, they not only justify the effort and time that they have invested in those activities but are also able—perhaps—to perform them more effectively. In this respect, such notions have not only a practical but also a political dimension.

At the same time, these notions, foregrounding parental expertise, liken parents to professional figures—a specialised clinician, paramedic, meticulous bookkeeper, responsible and successful manager. This drawing of parallels between the parental experience of IMDs and the idea of ‘work’ in general tends to import a relationship of detachment or alienation between the professionalised subject (the caregiver) and the object of their professionalism (the child). For instance, in the case of Agnieszka’s quote above, the professional metaphors she employs (‘monitoring’, ‘doing an interview’) certainly capture effectively the prolonged and systematic character of the task of attending closely to a child’s health condition. Through this language, she communicates the supervisory relationship she has over the object of this observation (her child), as well as the specific knowledge and capacity of rational reasoning and other competences required, to conduct such observation and analyse its outcomes. At the same time these notions—while they foreground the parent’s expertise and routine—obscure the emotional burden (‘my stomach tightens’) of making the right decision, and the fact that getting the child to the hospital on time is not a bureaucratic problem but a matter of life and death.

Moreover, the metaphors of professionalisation tend to overemphasise the individualistic dimension of parenting and obscure the relationality of healing. First of all, they tend to silence the role of the child in the treatment. Yet—as it is evident in the narratives of parents in my research—over the years, children become more and more actively involved in managing their disease: they learn to distinguish foods which are allowed for them, recognise symptoms and share them with their parents, ask for snacks themselves, manage social knowledge about their illness, etc. Second, in addition to clinicians, parents, and the children themselves, scores of other people make an appearance in parental narratives, acting as important agents in treatment and care of a child with a metabolic disorder. Many parents rely on grandparents (for instance, one Polish mother’s mother is a nurse and was instrumental in helping her daughter understand the disease as well as took an active role in her grandchild’s care). Others mention babysitters, or friends who provide respite care, long-time personal assistants (in Sweden), and so on. The presence of these persons in the narratives of these families contrasts the individualistic character of the predominant terms and concepts used to describe parental roles and tasks. In fact, while the discourse concerning the professionalisation of the parental roles tends to foreground individual responsibility for treatment and care, it is counterbalanced (especially in Poland) by another, which stresses relationality of healing and draws on the idiom of kinship and community. For instance, the Polish doctors I interviewed refer to the biosocial community of parents as a ‘clinical family’ (Polish: *rodzina kliniczna*), while the families tend to build strong relationships with their physician (who may be referred to as ‘auntie’ or ‘our Mr Doctor’ by the family). Equally, participants in

Facebook groups for parents of children with IMDs create an image of the group as a ‘girls’ community based on relationships of camaraderie (the group brings together mostly mothers, as the care of children with IMDs is strongly gendered). All these examples mark the limits of the discourse of professionalisation of parental roles.

Conclusion

In this Research Article, I have demonstrated the diversity of roles, tasks, and kinds of knowledge that the parents of children with rare metabolic diseases learn to perform, acquire, develop, and share with their peers. They range from practical, and embodied expertise, related to efforts of accommodating treatment regimes with values of everyday life, to biomedical and clinical competence, making these parents important actors in the treatment of their children. I have also shown that parental efforts cannot be thought of merely as a direct application of treatment principles that they have been taught in the clinic, which challenges the appropriateness of describing the parental care of a child with IMD as mere ‘homework’ (cf. Mattingly, Grøn, and Meinert 2011). In sum, in the treatment of children with inherited metabolic diseases, the distinction between patients’ and caregivers’ ‘experience-based expertise’ and the ‘certified expertise’ of scientists and clinicians becomes blurred (see Akrich 2010; Pols 2013, 77).

I have also highlighted the importance of concepts used to describe parental tasks, roles and knowledge. I have evinced a pervasive discourse of parental professionalism, employing references to biomedical practice (parents as ‘healthcare staff’), the organisation of medical services (parents as part of the ‘life-saving team’), scientific practice (parent who ‘experiments’), surveillance work (parent who ‘monitors’), capitalist division of labour (parent who does ‘bookkeeping’), and business relationships (parents who ‘cooperate’ with pharmacies). I have argued that those concepts are not mere descriptions. By opening some paths of reflection, and closing others, they have a real influence on the worlds of families living with rare metabolic disorders. At the same time, I have argued that while treating the parenting of a child with IMD as a profession certainly leads to the empowerment of parents and an acknowledgement of their efforts, it can also exaggerate the individualistic aspect of caregiving and obscure its relational character. The metaphors of professionalism may also alienate parents from their children, framing the children as ‘objects’ of depersonalised (however competent) knowledge.

From a more general perspective, the proliferation of metaphoric descriptions of patient and parent-patient roles among clinicians, patients, families, and social scientists seems to indicate that those roles are still ill-accommodated in common

representations of what treatment and care is or should be. This is despite many decades of patient activism, the advanced move towards the responsibilisation of patients and families, and a profound reorientation of healthcare towards chronic conditions. This article shows that this area of reflection is still important, and I argue that medical ethnographers and anthropologists should accompany parents (and doctors) in forging new concepts to objectify and justify the ways and practices of their everyday ‘metabolic living’ (Solomon 2016, 12).

Authorship statement

This article is authored by Filip Rogalski based on research conducted by him. The research was planned in collaboration with Małgorzata Rajtar, PhD, the PI of the project.

Ethics statement

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