

Anna Chowaniec-Rylke*

University of Warsaw

FAT-FREE, HIGH IN CALORIES. MANAGEMENT OF DIETS IN THE CASE OF A RARE METABOLIC DISORDER

LCHADD is a rare metabolic disorder with a worldwide prevalence estimated at 1/250,000. However in Poland birth prevalence is predicted to be 1/120,000 and as high as 1/20,000 in the Pomeranian district. The bodies of LCHADD patients cannot produce the enzymes which are key in the process of fatty acid oxidation. That means that life-threatening episodes occur only when the body is “fasting” – the calorie intake is lower than the body’s needs. There are no known cures or medications for LCHADD, but with proper treatment patients can improve and survive into adulthood. The treatment involves a strict diet, high in calories but fat-free. In this article I will sketch the dietary choices made by patients’ parents. Those findings will be established based on anthropological fieldwork conducted for over a year among those families. My intention is to show how decisions made by caregivers regarding an ill child’s diet can be interpreted in the broader context of consumer society, agency and a social model of disability.

Keywords: medical anthropology, rare disease, diet, agency, disability

INTRODUCTION

It is a common knowledge that eating too much fat can be harmful for our health. Most of us should avoid excessive amounts of fat in our diet – especially saturated fatty acids, which are proven to contribute to many diseases like obesity and atherosclerosis (Achremowicz and Szary-Sworst 2005: 24). On the other hand healthy fats omega-3 for example – are needed for our bodies to work properly. However, in case of LCHADD¹, a rare metabolic disorder, long-chain fatty acids should be completely removed from the patient’s diet. The problem is that those kinds of fats are the most common in our food. So what happens when the most easily accessible foods can be potentially destructive to the health of LCHADD patients? In this article I will try to sketch out the dietary choices made by the parents of LCHADD

* Corresponding author: Anna Chowaniec-Rylke, Uniwersytet Warszawski, Wydział Historyczny, Instytut Etnologii i Antropologii Kulturowej, ul. Żurawia 4, 00-503 Warszawa; e-mail: anna.chowaniec90@gmail.com.

¹ In this article I will be using an abbreviation of the full name of the disease: Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency.

children. First, I will briefly describe medical anthropology (from the perspective of which I will lead the discussion following) and methods of my fieldwork; then I will proceed to explain exactly what LCHADD is. I will then show how patients' caregivers deal with very demanding dietary requirements. In the last part I show how patients' families work to make their lives as close to "normal" as possible by gaining control over dietary restrictions, and how self-made food can influence social perception of disease and disability. My intention is to show how decisions made by parents regarding their ill children's diets can be interpreted in the broader context of consumer society, agency and disability.

MEDICAL ANTHROPOLOGY

Medical anthropology is a sub-discipline of cultural anthropology that studies concepts of health and disease, healthcare systems and bicultural adaptations (McElroy 1996). It examines the ways in which culture and society are organized around and are influenced by health-related issues. Early studies of what we now call biomedicine (or Western medicine) were primarily conducted by sociologists during the 1950s and 1960s (Gaines and Davis-Floyd 2004: 95). Rare diseases became a subject of anthropological research², but to my knowledge there are no anthropological studies (in the field of medical anthropology) about links between food and rare metabolic diseases. The anthropological study of food and eating has a longer history, beginning in the nineteenth century with Garrick Mallery and William Robertson Smith (Mintz and Du Bois 2002: 99). There have also been studies about food, diseases and society (for example, Native Americans' embodiment of modernity, Wiedman 2012; food beliefs and practices among British Bangladeshis with diabetes, Chowdhury, Helman et al. 2000; and Mexican ethnicity in diabetes research, Montoya 2007).

FIELDWORK³

My research, unlike most "classic" ethnography, is not happening in one geographical place. Fieldwork in medical anthropology is rarely linked to a specific place⁴; it is rather a process of participating in the discourse, which consists of meeting the subjects of your research (patients and their families, doctors, dieticians, workers and volunteers in patient organizations), as well as keeping track of media coverage, internet forums and Facebook groups, and

² There are examples of anthropological studies concerning mucopolysaccharidosis and the Turkish migration background in Germany (Dilger, Leissner et al. 2013) or the role of an online community for people with a rare disease in the case of primary biliary cirrhosis (Lasker, Sogolow et al. 2005).

³ The article is based on data collected during two years of anthropological fieldwork conducted within the framework of the project "Socio-cultural dimensions of rare diseases on the example of LCHAD deficiency. Comparative study of Poland and Finland" financed by the Polish National Science Centre (2015/17/B/HS3/00107).

⁴ However there is a possibility of conducting medical anthropology research in specific places such as hospitals, as shown by van der Geest and Finkler (2004).

attending events such as conferences and workshops. During my research I have met a total of 18⁵ Polish patients with LCHADD and their families. I also followed a Facebook group which brings together people with LCHADD and their parents, and attended the 14th and 15th editions of the International Conference on Rare Diseases, the National Conference for PKU and Rare Diseases Patients (both in 2016) and the official Rare Diseases Day celebrations in 2017 and 2018. Owing to new diagnostic possibilities (discussed below) LCHADD patients are almost exclusively children, teenagers and young adults (in the group of participants, the children were mostly in kindergarten [8 children], or of elementary or secondary school age [4 children]. In 2011 the two oldest people with LCHADD in Poland were 23 years old (Sykut-Cegielska, Gradowska et al. 2011: 186). Thus during my research I spoke mostly (but not only) with caregivers – usually mothers of LCHADD patients. In my opinion this approach was the most beneficial for the research – children that young cannot make medical decisions (the responsibility is always on parents). Also, it is the parents' concern to adhere to the dietary requirements. That is why I interviewed them rather than their children⁶. The interviews gave me an opportunity to discuss the child's biography – LCHADD can be seen as an episodic disease, following a trajectory of relatively good health and its rapid deterioration. Children look and behave like their peers, but all of them experienced prolonged hospitalization. Without the parents' narratives about the past my observations and conclusions would be incomplete. I often spend all day with a family⁷, which allows me to observe how food is prepared and when, how, what and with whom the children were eating. I choose participatory observation rather than interviews, and in this text I will rely mostly on those observations and my field notes. The method of looking at the phenomena on a microscale allows the researcher to focus attention on relationships and dependencies that would be unnoticeable from a bird's eye perspective. At the same time, the anthropologist is not trying to discover the laws that govern social life, but rather certain trends that are closely related to specific historical and social contexts. The research field of an anthropologist should therefore be specific problems (Halemba 2011).

LCHAD DEFICIENCY

LCHAD deficiency (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) is a rare metabolic disorder⁸. The worldwide birth prevalence is estimated at 1/250,000⁹. However,

⁵ There are no data concerning the total number of people with LCHADD in Poland. I estimate (based on publications as well as the number of Facebook group users) that there are about 100 patients.

⁶ Ethnographical research among children is possible, but it is a much more ethically demanding process. However, there is a discipline that studies children in their social and cultural context (childhood studies). Although it is a very young discipline, there are Polish studies on childhood (Interdisciplinary Childhood Studies Team of Warsaw University).

⁷ The length of my meetings with parents varied, all depending on the parents will to participate. I usually spent a few hours before and after noon, but in some cases families allowed me to stay with them for longer (3–14 days).

⁸ According to European Commission on Public Health a rare disease is a condition with 1 : 2000 prevalence (<https://www.ima.org.il/filesupload/imag/0/200/100025.pdf>).

⁹ http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=5.

around the Baltic Sea the frequency is higher: birth prevalence is estimated at 1/120,000 in Poland and 1/20,000 in the Pomeranian district¹⁰. LCHADD is an hereditary, autosomal recessive disorder, which means that if both parents are carrying a copy of a defective gene there is always a 25% chance that the disease will affect the child¹¹. In 2010, 59 patients were diagnosed with LCHADD in Poland (Piekutowska-Abramczuk, Olsen et al. 2010: 374).

The LCHADD patients' bodies cannot produce enzymes which are key in the process of fatty acids oxidation (Sykut-Cegielska 2006). This process plays a very important role in supplying energy to the organs. The organism draws energy from glucose, the main fuel of the body. When the glucose supplies run out, the mitochondrial fatty acid β -oxidation process starts (Sykut-Cegielska 2006). This process is essential when the body uses more energy than is produced from carbohydrates (Tyni and Pihko 1999). When the glucose supply runs out, symptoms (some life-threatening) may occur (including cardiomyopathy, cardiac arrest, coma, ketotic hypoglycaemia, and retinopathy, Sykut-Cegielska 2006). Such episodes (called "metabolic decompensation" by doctors and parents) may happen when the body is "fasting". It does not necessarily mean that the patient is hungry – early stages of infection, vaccination or even physical activity can lead to depletion of calories needed. An attack of the disease can be fatal, and for a long time it was. In the past, before the disease was well-researched, it may have led to sudden deaths in infancy (bodies of new-borns and small children quickly consume glucose, Sykut-Cegielska 2006). Until recently the mortality rate was 36% among Polish patients (Sykut-Cegielska 2006: 56) and it is estimated that approximately 40% of children with LCHADD die in the early days or years (Autti-Ramo et al. 2005: 1130). LCHADD was first diagnosed in Poland in the early 1990s but diagnosis was possible only once the child's health was rapidly deteriorating (Sykut-Cegielska 2006). The situation changed in 2000 when The Institute of Mother and Child began a pilot new-born screening program¹². This led to earlier diagnosis, so children do not have to experience a long hospitalization.

There are no known cures or medications for LCHADD. However, with proper treatment patients can improve and survive into adulthood. Treatment involves a strict diet that should be followed for the rest of the patients' life, and it is the only known way to keep the patient relatively healthy. The two main dietary requirements are: patients must avoid "fasting" – meals should be high-calorie, with lots of slow digestible carbohydrates (rice, barley or even corn starch); and the diet should be free of long-chain fats, because the organism cannot produce energy from this source. Medium-chain triglycerides are added artificially in form of oil, powder or margarine. In cases of rapid metabolic decompensation, the child is often taken to hospital¹³, where doctors administer intravenous glucose infusion in hopes of improving the child's state (Haas and Burgard 2016). This is the only known way of helping LCHADD patients in such an episode.

¹⁰ Ibidem.

¹¹ <http://chorobyrzadkie.blogspot.com/2012/12/deficyt-lchad-rzadka-mutacja-genetyczna.html>.

¹² <http://przesiew.imid.med.pl/choroby.html>.

¹³ Some parents found other ways, other than taking child to hospital, for example glucose infusion is administered by a befriended nurse or by hospice workers at patients home. Those parents were avoiding taking their children to hospital, since the likelihood of infection increases, and one visit to a hospital can easily turn into a long stay.

Although there is no certainty regarding the amount of different fatty acids in one's diet (Sykut-Cegielska 2006: 57), my interlocutors followed two main rules: plenty of calories and no fat. Keeping with the diet and occasionally occurring health problems were the two central themes in their experience of living with LCHADD or caring for an LCHADD patient. It also influenced every family member (to varying degrees) and the household as a whole. However, it must be noted that the group of people that I met is not homogeneous by any means. They share many experiences and opinions, and they face the same choices, but that does not mean they represent some kind of common front. Their decisions regarding medical procedures, their children's educations, and family economy (social welfare) differed. Despite that, regarding diet all of them share common ground. Below I discuss some of the themes that recurred in almost all of my interviews and in the Internet discourse.

EVERYDAY LIFE WITH LCHADD

Probably the biggest everyday struggle for parents is to keep track of what and when the child should eat. The doctors' order is in most cases to feed the child every three–four hours (when they are infants). Most of the children I met were at some time in their life fed by nasogastric tube or PEG¹⁴ (either in the first few weeks after diagnosis, during hospitalization, or because of additional health problems that prevented the child from eating orally). Parents, however, were not very fond of this solution¹⁵ and tried to avoid it whenever possible. I think that suggests that eating “normally” – through the mouth – was incredibly important for the parents (for the reasons I discuss below). Feeding through a tube or PEG is probably safer in some ways: parents do not have to deal with the child's appetite (or lack thereof) or with making different kinds of food (because when feeding this way, the most common foods are porridge and a special formula). Despite the convenience and safety of these methods, parents choose to struggle with the diet by watching over what their child eats, preparing meals, and doing everything else that is required. When asked why they made the decision not to feed by tube, in most cases they said something along the lines of ‘making the child's life more close to being “normal”’, which meant eating like their peers and family. In the long run, this rough path that they chose helps their children by making them more independent in terms of education, hobbies, etc. Teaching them to eat was, is, and will remain a challenge: these kids do not feel real hunger for most of the time (since they are almost constantly eating). Preparing the required meals pushed some parents (mostly mothers) to resign from work¹⁶.

¹⁴ Percutaneous endoscopic gastrostomy is an endoscopic medical procedure in which a tube is passed into a patient's stomach through the abdominal wall.

¹⁵ It also applies to other medical procedures involving the interference in the body, such as PEG (percutaneous endoscopic gastrostomy) or a central venous catheter. Both of those procedures were, if possible, avoided by the parents.

¹⁶ According to Polish law, the parents of a sick or disabled child are entitled to an allowance. There are two types of monthly financial support: higher for parents who have resigned from work to take care of their child (1406 PLN or 335 EUR) or lower for families with a disabled child (153 PLN or 36 EUR). The exact amount of the allowance is determined by a medical commission. In order to receive a higher benefit, the committee

The main struggle with an LCHADD diet is to eliminate all fats. Although as mentioned above there is no consensus on the permissible amount of fatty acids, parents avoided all kinds of fats. The only oil or fat that their children ate were MCT oils and margarine¹⁷, added to food artificially. That means that sick family members cannot eat the same meals as the rest of the family. I asked mothers – who in majority were responsible for cooking – how they managed with all kinds of different diets in one household. It was obvious that the children required separate meals, but for the sake of convenience many of these families ate the same meals (sometimes prepared in a different way). During my visits I often ate dinner with the family. The meals contained, for example, fat-free soup that all the family ate, potatoes and vegetables (for the whole family), a chicken fillet for the child and pork chops for other family members, zucchini pancakes (fried in MCT oil for the child), and dumplings with strawberry sauce (for all family members). The children ate separate meals, but mothers, in most cases, tried to make the same kinds of food for the whole family. Although this meant that mothers were preparing a few different meals, they were willing to put the effort into making meals that could be eaten at the same time at the same table. Later I will return to these examples and I use them to show how, in my opinion, parents tried to gain a sense of agency over dietary requirements, and how it links to a social model of disability.

FACEBOOK GROUP

One major source help for the parents is a Facebook group that I came across. It not only allows parents and LCHADD patients to create a network and connect (which is probably the most important point of making the forum¹⁸), but it is also a platform to share new recipes and fat-free products. The most substantial part of the forum content is made up of articles about new recipes, links to cookbooks, photos of desserts made by the parents, and questions regarding products and new ideas. Some parents are more internet-active than others, but all interlocutors admitted to have looked there for inspiration. The most recently published recipes included birthday cakes, cupcakes with fruit, potato and pumpkin pancakes, and chicken fillet with vegetables. For the Fat Thursday celebration some mothers shared their recipes for donuts and *faworki*, which are traditional Polish Fat Thursday cakes (some of them were steamed, yeast-based cakes, and some were fried according to the original recipe, but

must recognize that the child requires constant care and is not capable of self-care. The decision on entitlement to the allowance is issued on a temporary basis, up to 16 years of age. The parents' encounters with the committees are problematic. LCHADD is not always considered a disability; it all depends on the medical expertise. But since most doctors in the adjudicating medical committee are not familiar with the disease those decisions regarding levels of disability may differ. That means not all parents have the opportunity to resign from work and live on social benefits.

¹⁷ Those products are imported to Poland (as a bearer prescription medicinal product).

¹⁸ For the families of children suffering from rare diseases and the patients themselves, the Internet becomes the only available platform to establish contact with each other, as the rare nature of the diseases makes it very hard for them to meet in person. This is not only because the diseases themselves are rare, but also because patients are spread all over the globe, and sometimes the severe course of the diseases makes it impossible for the patients and their parents to travel and meet other people affected by the same condition.

in MCT, not vegetable oil); and for Christmas and Easter celebrations: pate, lean sausages and cookies. Parents also share new products that they have found – for example the cocoa powder substitute carob, vegetable pate, chestnut spread, and bottled egg whites¹⁹. Some parents also share pictures and information about kitchen appliances, such as Thermomix (a multipurpose appliance) or a kitchen scale that counts calories.

NEW PRODUCTS, NEW POSSIBILITIES

All of that would not be possible if those products were not easily accessible. A very important point to make when discussing LCHADD patients' diets is that the accessibility of the products plays a crucial role in making children's lives more "normal", as it allows them to dine with their families and be guests at birthday parties or celebrations. The large selection of fat-free products on the market allows parents to prepare meals that resemble what the rest of the family eats. In the past, it was not that easy. Now, because of economic changes and the development of the consumer society²⁰, those kinds of products are available. 0% fat milk, fat-free cottage cheese, super thin slices of cheese and poultry sausages are products that look and taste like "original" products. By buying these kinds of food, parents can bring their child's dietary requirements closer those of their peers and family. Another significant improvement caused by changes in the market are snacks high in calories. Jellies, straws to make instant chocolate milk, candies, and lollipops are just some of the snacks that parents bought for their children. Although not particularly healthy for any other child, these sweets are high in calories, which ensures high levels of glucose in the child's system. They can also be taken for a walk or to school, when the child's body burns more energy. Some parents have special drawers full of candy, for the child to help themselves at will. Because the children cannot eat chocolate, nuts or pre-packed cakes, the variety of fat-free sweets was the parents' "secret weapon" on the front of keeping their children's calorie intake high without using any fats.

GAINING AGENCY, CHALLENGING DISABILITY

Those observations lead me to my final observation. The route that the parents of children with LCHADD are taking is by no means easy. Preparing home-made meals safe for their children was an everyday struggle. They were not, however, only "making meals": all of the given examples show that it is their way of gaining control²¹ over the diet, in a battle to

¹⁹ Only egg whites are fat-free, LCHADD patients cannot eat the yolk.

²⁰ Consumer society is a society in which people often buy new goods, especially goods that they do not need, and in which a high value is placed on owning many things (Cambridge Dictionary). This led to the greater selection of the products on the market.

²¹ I understand agency, after Martin Hewson, as the experience of acting, doing things, making things happen, exerting the power and controlling things (Mills, Durepos et al. 2010).

make their children's lives more "normal". Not only did they try to make the children meals as close to their own food as possible. New products available on the market let them do so. The Facebook group is full of recipes that resemble something well-known, like poultry pate, cheesecakes and more, all of which were LCHADD-friendly. Also, the examples given above show that the whole family in most cases eats the same meal, only with slight variations.

There are times when an increase in recipe posts can be seen. Holidays and other celebrations – such as Christmas, Fat Thursday, and children's birthdays – are important in the families' lives, but because of dietary restrictions most of the traditional (or traditionally prepared) meals are forbidden. Parents use all of the available resources to make the celebrations close to "normal" – so they can spend this time with family and friends, eating at the same table. They ask other parents for ideas, share their own recipes, and buy new fat-free products. In doing so they are trying to overcome the hardship of the disease.

There is also one more example of how new possibilities help parents of LCHADD children. Those novelties have allowed children to live lives similar to their peers. Because of the available products, children can eat brunch at school – if they have a gym class, they should eat some sweets, and they are obligated to eat whatever their parents have prepared. What they have prepared, however, is probably the most important part. When I interviewed parents about that, a clear image appeared. If the child is in kindergarten, in most cases parents prepare meals for them. They not only make them at home: they try to cook something close to what the rest of the children are eating that day, or what children of that age typically eat. Also with packed lunches for school, children were given sandwiches, fruit, and sweets, just like other kids. That does not mean that the relations were smooth among parents, teachers, and headmasters. Schools were often prejudiced as to having a sick child under their guardianship. That led some of the parents to choose inclusive schools (and in two cases schools for children with special needs), hoping that it would be easier to establish the best care²². It was still, however, an extremely stressful time for the parents, as they could no longer watch over their children. The child's diet, although very restrictive because of the life-threatening metabolic disorder, was similar to their peers, which was important for the parents. By trying to make food that resembled that eaten by the kids in school and kindergarten, the effort was to not secondarily "disable" their sick child. According to the social model of disability, it is the social, not medical, barriers that make impaired persons disabled (Barnes and Mercer 2008; Crow 1996; Shakespeare 2006). In this model, disability is the disadvantage or restriction of activity caused by social organisations which take little or no account of people who have impairments, and thus exclude them from participating in the mainstream of social activities (Shakespeare 2006: 198), such as dining with friends and family. Being fed by tube certainly socially disables patients, by withdrawing them from a very important social activity – eating together – which is one of the most important family bonding activities (Haukanes 2007). Food is probably the only area in which parents can gain some sense of agency over the disease. They cannot always help their child with medical conditions, and they can try to make their life as "normal" as possible. Preparing

²² Parents were using different techniques to ensure their children will eat at school, but most commonly they asked teachers to remind the child that they need to eat their lunch. Unfortunately it did not always worked.

food that looks and tastes like the food that other family members and friends eat allows those kids to lead fairly uninterrupted social lives. By choosing to pursue a diet instead of feeding through a tube, parents create an opportunity for their children to be like other kids. Preparing food which meets dietary requirements but is also very typical and traditional is not an easy task. By looking for recipes and buying new products it is, however, possible. All of that not only shows the parents' determination in modifying an extremely restrictive diet to overcome social barriers created by it, but it also brings their children closer to the parents' main goal – independence and social inclusion. Although families had different stories, their children were individual people with their own struggles, and this pursuit to not make their child socially disabled was common for all of them.

CONCLUSION

The main purpose of this article was to show how diet may be interpreted and implemented by the caregivers of a child with a rare metabolic disease. New possibilities – like the internet and the variety of fat-free products and sweets – have allowed the parents to gain agency in the case of LCHADD. Although the LCHADD patients' diet is restrictive and hard to maintain (all of the meals must be home-made), parents found ways to adapt it to their and their children's lifestyles. Efforts made in preparing meals had two purposes: to keep the child healthy, and to make their lives as close to those of their peers as possible. Parents' main intentions are to make their children independent and not to let them be in any way socially disabled by their diet. By interpreting the diet so that it meets the needs of an LCHADD child, they work to make meals look and taste similar to those eaten at the same table (either at home or in school). That encourages their children to take part in social activities, such as dining with other people, and thus ensures they will not be seen by their peers (or other members of extended family) as “more disabled” than they are (in their own eyes). Of course adjustments to the diet will not completely answer the question of the definition of disability in cases of rare metabolic disorders, but since it is one of the crucial elements in the parental experience, it cannot be overlooked. Preparing food and managing meal times becomes the core of the lives of parents of LCHADD children. The laborious path they choose to ensure their children are not secondarily disabled suggests that they consider just how important it is that contemporary society can easily exclude someone, even if the difference lies only in the diet.

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BEZTŁUSZCZOWE I WYSOKOKALORYCZNE.
PRZESTRZEGANIE DIETY W RZADKIEJ CHOROBY METABOLICZNEJ

LCHADD jest rzadką chorobą metaboliczną, która na świecie występuje z częstotliwością 1/250 000 żywych urodzeń. W Polsce liczba ta jest dwa razy wyższa, a na Pomorzu sięga nawet 1/20 000. Ciała osób z LCHADD nie wytwarzają enzymów koniecznych w procesie oksydacji kwasów tłuszczowych. Oznacza to, że zagrażające życiu dekompensacje metaboliczne pojawiają się, kiedy organizm znajduje się w stanie „przedłużonego głodu”, co oznacza, że ciało zużywa więcej kalorii, niż przyjęło z jedzeniem. Na LCHADD nie ma leku, ale dzięki odpowiedniemu postępowaniu chorzy na LCHADD mają szansę na dożycie dorosłości. Najważniejszym elementem „leczenia” LCHADD jest restrykcyjna dieta: wysokokaloryczna, ale beztłuszczowa. W artykule omówiłam wybory dotyczące diety dokonywane przez rodziców dzieci z LCHADD, wśród których prowadziłam ponad roczne badania etnograficzne. Celem artykułu będzie przedstawienie i zinterpretowanie decyzji dietetycznych, podejmowanych przez opiekunów chorych dzieci, w szerszym kontekście konsumeryzmu, poczucia sprawczości oraz społecznego modelu niepełnosprawności.

Słowa kluczowe: antropologia medyczna, rzadka choroba, dieta, agencja, niepełnosprawność