

**Parental experiences of raising a child with medium chain
Acyl-CoA Dehydrogenase deficiency**

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Parental Experiences of Raising a Child With Medium Chain Acyl-CoA Dehydrogenase Deficiency

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Abstract

Newborn screening enabling early diagnosis of medium chain acyl-CoA dehydrogenase deficiency (MCADD) has dramatically improved health outcomes in children with MCADD. Achieving those outcomes depends on effective management by parents. Understanding parental management strategies and associated anxieties and concerns is needed to inform provision of appropriate care and support. Semistructured interviews were conducted with a purposive sample of parents of children aged 2 to 12 years. Thematic analysis identified two main themes. Managing dietary intake examined how parents managed day-to-day dietary intake to ensure adequate intake and protection of safe fasting intervals. Managing and preventing illness events explored parental experiences of managing illness events and their approach to preventing these events. Management strategies were characterized by caution and vigilance and influenced by a lack of confidence in others to manage the condition. The study identifies the need for increased awareness of the condition, particularly in relation to emergency treatment.

Keywords

medium chain acyl-CoA dehydrogenase deficiency (MCADD), inherited metabolic disorder, genetic condition, child health, dietary management, lived experience, family life

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Background

Medium chain acyl-CoA dehydrogenase deficiency (MCADD) is the most common inherited disorder of fatty acid oxidation. The incidence is highest in populations of Northern Europe and it affects between one in 9,000 and one in 10,000 newborns in the United Kingdom (Grosse, Houry, Greene, Crider, & Pollitt, 2006; Oerton et al., 2010). The condition is due to a genetic mutation causing a deficiency of medium chain acyl-CoA dehydrogenase, an enzyme required to metabolize medium chain fatty acids. The lack of this enzyme results in improper metabolism of medium chain fatty acids and their accumulation in the blood. The metabolic crisis presents clinically as hypoglycemia and lethargy and without rapid treatment it can result in liver damage, brain damage, and death (Leonard & Dezateux, 2009).

Early detection of MCADD changes the natural history of this condition. In the absence of screening, the condition most commonly presents symptomatically and in 20% to 25% cases results in premature death or disability (Grosse et al., 2006). However, screening in the first few days of life

enables early detection and institution of preventive treatment with resultant good health outcomes in the screened population (Oerton et al., 2010; Joy, Black, Rocca, Haas, & Wilcken, 2009; Lindner et al., 2011; Wilcken et al., 2009). Early detection has also significantly reduced the likelihood of adverse events including death (Nennstiel-Ratzel et al., 2005; Wilcken et al., 2009). In 2009, MCADD was added to the newborn screening (NBS) program in England and Northern Ireland, 2011 in Scotland, and 2012 in Wales (Jameson & Walter, 2015). Over the past decade, MCADD screening has been embedded in similar programs across the world (Therrell et al., 2015)

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MCADD is managed by diet, the main requirement being to ensure that food intake is sufficient to meet metabolic demands and the prevention of prolonged periods of fasting. The permissible maximum length of fasting intervals when well increases with a child's age: 4 hours for the first 4 months, 8 hours for the second 4 months and 10 hours from 8 to 12 months of age. At 1 year, children can go up to 12 hours without food and this "safe" fasting interval must be maintained for the rest of their lives (British Inherited Metabolic Diseases Group, 2016). Specific management is required during periods of illness with the introduction of an emergency dietary/feeding regime involving feeding every 2 to 3 hours with glucose polymer feeds (called SOS in England). This highlights the cornerstone of treatment during illnesses, which is the reduction of fasting times to 2 to 3 hours.

The number of families raising a child with MCADD will have increased as a result of the NBS program enabling prevention of premature death. However, the social effects of the condition on the family have received very limited attention in the literature. A wide ranging ethnographic study examining the NBS program in the United States offered valuable insights into early parental responses to the disease: the shock, devastation, and fear they experience at diagnosis; the medical guidance and advice they receive around managing the condition during infancy; and the ways in which that management is negotiated between doctors and parents (Buchbinder & Timmermans, 2012; Timmermans & Buchbinder, 2013). A small study of parents ($n = 11$) with MCADD children under 4 years found that parenting stress levels were significantly lower than the normative levels for the U.S. population and attributed this to the personal and professional support they had received (Torkelson & Trahms, 2010). These findings contrast with those from a larger study that explored parents' perspective on child development, expectations for the future, and burdens on family life for a range of metabolic conditions (Gramer et al., 2014). Gramer et al. surveyed parents of 187 children aged from 6 months to 12 years ($M = 3.5$ years) with metabolic conditions of which 25% had MCADD. Despite good outcomes for the children identified through the NBS, a considerable proportion of parents (75%) reported that the disorder and treatment placed a substantial burden on their family. A total of 56% rated the burden as mild or medium and 19% rated it as severe or very severe. There were higher odds of severe or very severe burden for the family for diagnoses with a risk of metabolic crisis as compared with those without such a risk. However, there was no explanation of what constituted that burden of care.

There is a need to understand how parents manage MCADD, the challenges they identify in caring for their child and the ways in which they respond to those challenges. There is also a need to understand how those challenges change over time as the child grows older to inform care delivery for these families. This article arose from a larger study examining family planning decisions among parents of children with rare metabolic conditions. A substantial proportion of those

recruited to the study were parents of children with MCADD, whose accounts offered important insights into parents' lived experience of having a child with MCADD.

Method

A descriptive qualitative study design was adopted to gain understandings of the parental experience of raising a child with a metabolic disorder. A purposive sampling approach was used with parents recruited to the study from two hospital sites within one Regional National Health Service (NHS) Specialised Metabolic Disorder Service in England. Those eligible to participate were identified by the metabolic consultant and given verbal and written information about the study at a routine clinic appointment. Parents interested to participate were contacted by the research team and recruited to the study, either as an individual or as a couple. Written informed consent was obtained prior to data collection.

Data collection involved a single semi-structured interview which was conducted by one member of the project team at the participants' homes, at a time and place convenient for them. The interview schedule comprised a small number of key questions to enable in-depth discussions that adequately captured individual experiences and perceptions (Cresswell, 2008). Interviews were digitally recorded and transcribed verbatim. They lasted from 30 to 100 minutes.

A thematic approach to data analysis was used (Braun & Clarke, 2006). This involved familiarization with the entire dataset, open inductive coding of all interviews and development of a thematic structure with overarching main themes and their subthemes. To ensure rigor of the process, all members of the research team contributed to the analytic processes. Interviews were independently coded by two researchers (HP and KM) in the initial stages to agree coding descriptors which were then used across the dataset. The thematic structure was developed through an iterative ongoing analysis involving testing and refinement of candidate themes to ensure that similarities and differences were articulated and concepts were comprehensively captured.

Ethical Approval and Research Governance Permission

We were granted ethical approval by the Local Research Ethics Committee (15/YH/0035) and Research Governance approval by the local NHS Foundation Trusts.

Findings

An Overview of Study Participants

Ten families contributed to this part of the study. We interviewed eight mothers, one father, and one couple. The median age of the children ($n = 10$) with MCADD was 3 years (range 2–12 years). Eight were boys and two were

girls. The number of children in the families ranged from one to four. Pseudonyms have been used throughout in the reporting of findings.

Themes

Two major themes emerged from the study. “Managing dietary intake” contained four subthemes: “infant feeding,” “feeding a child,” “involving others in the care,” and “concerns for the future.” “Managing and preventing illness events” contained two subthemes: “accessing emergency hospital treatment” and “taking a preventive approach.” Some of the quotes we have used for reporting the themes include colloquial terms—we have provided explanations in square brackets to aid understanding.

Managing dietary intake. This theme explored how parents managed day-to-day dietary intake to ensure that their child was eating enough food calories and that fasting was not extended beyond permissible intervals. Although, as Ben’s father identified, the condition is “intellectually easy to manage, feed and keep fed,” the reality of making that happen was challenging and produced a number of anxieties and uncertainties, which changed as the child got older.

Infant feeding. Parents had found it stressful to ensure that their baby received regular feeds and an adequate energy intake, particularly during the night. Ian’s mother explained that as her son got older “I had to set an alarm clock to wake him up to give him a feed because he would have slept through from quite a young age.” Similarly, Craig’s mother described the difficulties of feeding a baby who was reluctant to eat: “You had to make sure you fed him every four hours, and he didn’t like food when he was a baby so you’d be force feeding him to get the food. It was very stressful.” Advice from well-meaning family members added to the stress of the situation as Ellie’s mother explained:

They would say: “oh if they’re not hungry don’t feed them . . .” and we are trying to explain to them that actually we can’t do that. Like they would see you trying to give her milk after three hours or something and saying, “well she’s clearly not hungry.” It frustrates me.

Anxieties around adequate energy intake, particularly in those breastfeeding, created a tendency to overcompensate with the feeding regime, as Ellie’s mother acknowledged:

The most difficult thing was trying to understand how much is enough milk . . . I was having to wake her up every single night, you know, and obviously at first two or three times, four times a night . . . Probably waking her up more than I needed to because I wasn’t sure how much she’d had.

Parents had found it difficult to make the transition to longer intervals between feeds, particularly extending them to

12 hours. They described themselves as being fearful, hesitant about making the adjustment, and had to be encouraged to do so by health professionals. Parents’ tendency was to delay, in some cases, continuing night feeds for several months longer than recommended. When they did finally take the plunge and allowed their child to sleep through the night, it caused huge anxiety, as Ellie’s mother explained:

It’s like the first time you drop the dream feed or the night time feed, you know, the first time you let her go twelve hours, it was horrendous . . . I think she was probably about 18 months, so she was well over the one year mark that they say. In the first couple of nights I was going in every two or three hours and checking she was all right and that, you know. But, and then you go in and wake her up in the morning and you’re like is she all right?

Feeding a child. As the child got older, some of the early anxieties resolved. Parents became more confident and the management of the dietary intake became embedded into daily routines. Harry’s mother described how “when he was like six months old I was a nervous wreck, [but] I’m used to it now,” and Ellie’s mother similarly explained “but I think from how scary it was, now it is just everyday life to us.”

However, there were challenges associated with ensuring adequate food intake in a child who was assuming an increasing degree of agential control. For 5-year-old Craig who “seems to like his food even if he’s got a cold” and 3-year-old Freddy who “tells me [his mother] when he’s hungry for dinner, I’m not like force feeding him at twelve o’clock,” this presented relatively few problems. It was much more problematic for children such as 4-year-old Glyn who resisted eating because “he doesn’t feel that he wants to eat.” Parents were acutely aware that refusing to eat and missing meals is not an option for an MCADD child because, as Dan’s father explained, “It’s not food because you’re hungry; it’s food to keep you alive.” They reported persistent concerns about how much was enough and commonly reverted back to baby food or supplementary bottle feeds to ensure adequate intake:

Because I try and say how do you know what’s enough and what’s not enough, sort of thing . . . surely if you have a couple of mouthfuls that’s not going to be enough. So that’s why I have to keep trying these different things, as well as back to the baby food. But at least he’s eating isn’t he, he’s having something. (Ben’s mother)

They also identified anxieties around managing increased metabolic demands because of the uncertainties associated with determining energy requirements. When Freddy “started running about and it were hot weather,” his mother’s reaction was “oh god he can’t he’s using so much energy up.” Ian’s mother found that when her son became more anxious, “he does need extra snacks then because that must take up a lot of energy I imagine” explaining that she was “always almost second guessing everything all the while.”

Parents adopted a highly risk-averse approach to dietary management, relying heavily on high-calorie snacks and drinks to ensure that energy levels were topped up and safe fasting intervals were protected. Ellie's mother described "having a constant supply of snacks readily to hand, so she can nibble on something as soon as she wakes up in the morning." Ian's mother explained that "if we're out and about I always make sure I've got some snacks, always, always" and Dan's father explained that "Coke is his standard drink, and that's us being a little bit belts and braces [extra cautious]." However, the health consequences were a matter of ongoing concern for many parents who grappled with the implications for the general nutrition and dental health of their child as Freddy's mother indicated:

I'm constantly worrying about his diet and should I be giving him too much sweets . . . it's like, it's hard enough knowing the right things to give him anyway.

Involving others in the care. Parents were acutely aware of the burden of responsibility associated with caring for their MCADD child, which influenced whether and how they involved others in care. They were particularly cautious about involving others in night-time care because of the importance of adhering to the safe fasting interval. Harry had never spent a night with his grandparents because his mother "couldn't put that responsibility onto somebody" and none of the older children had yet stayed over with friends.

Parents had to be confident that anyone involved in caring for their child understood the condition, the importance of regular food intake, and the potential consequences of illness events. Almost none had ever heard of the condition and when parents tried to explain it, they commonly faced a range of reactions from "people who look at you in horror" or "a sense of panic and fear." Overall, they had to deal with a lack of understanding and in some cases disbelief about "that pretended disease" or the diagnosis with the suggestion from Ben's grandfather that "I think they've got it wrong."

Concerns for the future. All parents, including those of very young children identified the additional concerns associated with transition to adulthood and independent living, particularly the threat that adolescent behaviors, sleeping patterns, and alcohol usage posed to safe fasting intervals:

Obviously when you drink you tend to forget don't you, or fall asleep or have a lie in [sleep for longer]. (Craig's mother)

If you read the history of MCADD . . . , if a child's not dead frankly by the age of three the next risky time is 18 when they go away to university, when they . . . fly the nest [leave home], when they start doing silly things like drinking and others, where they're spending all night going to concerts and clubbing and all the rest of it. (Dan's father)

Parents were already anticipating the challenges they expected to face as their child got older and identifying strategies for survival. Although most felt that their child would manage the situation because "she'll have lived with it for so long" (Ellie's mother), a wider social understanding of the condition and having strategies including "a special friend that makes sure he eats or wakes him up" (Craig's mother) was seen as crucial for survival.

Managing and preventing illness events. Managing illness events explored parental experiences of managing illness events and their approach to preventing these events.

Accessing emergency hospital treatment. Most of the children in the study had needed emergency hospital treatment for illness events, after having failed home dietary/oral emergency treatment, in the first years of life. For example, Freddy "was really poorly with chicken pox, and he wasn't eating or keeping anything down," "when Jenny was 13 months old she got Rotavirus," and Ian was "admitted once [when] he was very young and he had a sore throat and he was being very sick."

Parents' experiences of accessing emergency hospital treatment varied considerably. Ellie's mother has had a very positive experience at the hospital: "they took us really seriously and saw us as quickly as they could." More commonly, parents reported negative and highly stressful experiences of having to forcibly advocate with health professionals to ensure that their child received care with the degree of urgency that the condition required:

I phoned the ambulance service and I said my baby's unwell. And they asked me what was wrong, and it was like "well that [diarrhea and vomiting] is not something." I said no, he's got MCADD, and they didn't know what I was talking about. And I had to get a taxi and I was like right, we need to get there as quick as possible. (Ian's mother)

So first of all we had a paramedic who tested the blood glucose and said "oh no he's fine." I said no he's not, this is more serious than that. "What's he got?" MCADD. "Oh I don't know, is that like diabetes? Well his blood glucose is fine." No you're missing the point, can we go to hospital please? (Dan's father)

And at first in A&E [accident and emergency department], I was trying to explain like what was wrong, he's got MCADD he needs a glucose drip up, it was like, I felt like they were fobbing me off a bit [not taking my concerns seriously] like I didn't know what I was talking about. And then [the metabolic consultant came] and he was just "right you need to do this now." (Freddy's mother)

Parents had been prepared to deal with services that were not familiar with the condition. They all carried a priority card that indicated their need for rapid access to care and an MCADD information pack (hospital emergency regimen) "to guide the medical teams on how to treat him." Metabolic

specialists had emphasized the need to “stamp your authority, and if you have to you scream and shout at them” (Harry’s mother). However, the onus of responsibility this placed on them was daunting for some:

And in the paperwork this sentence that says “always listen to the parents because they know more than you do.” And it’s quite daunting . . . But I think for me you worry, being given this paperwork it makes you worry even more, because it’s kind of like your child’s health is more in the hands of fate, if those doctors follow what is said on that. (Harry’s mother)

Many of the parents had “been bitten” by their earlier experiences with hospitals, as Ben’s dad described it, and were highly ambivalent about hospitalization. As their child grew older and they became more confident with MCADD, parents felt increasingly able to manage illness events at home. Ready access to support from specialist services enabled them to do so. This was clearly beneficial for the child but emotionally and physically exhausting for the parents:

I don’t know how we did it, but we managed at home and I was up, it was every hour-and-a-half we were syringing another 50 ml of SOS [the high-calorie drink treatment] into her . . . and I was on the phone to the dieticians and the consultants pretty much every day, twice a day. And although it was horrible we saved her the trauma of going in and having the needles and the IV line, so. We probably had it for about three nights, so up every single night, but we just needed to do it and it worked and she was fine. (Ellie’s mother)

Taking a preventive approach. Parents described taking a range of preventive measures to reduce the likelihood of serious episodes that might necessitate emergency treatment. They were acutely aware of the importance of not becoming complacent about the possibility of illness events as their child grew older and the continued need for prompt action. At the first sign of illness, their priority was to maintain energy intake over and above any other dietary considerations. Harry’s mother explained that:

When he’s unwell everything just goes out the window and it’s just like you know what, if you don’t want to eat your breakfast you can have two chocolate bars instead. I don’t mind as long as you’re getting something in you and we’re keeping you safe.

At the first indication of diarrhea and vomiting, parents went into “full mode immediately” to ensure small and frequent intakes of SOS, the high-calorie drink. Rapid action was essential because every illness was seen to carry the threat of metabolic crisis. As Jenny’s mother explained:

You panic, you think, she can’t eat, she can’t keep anything down and you don’t know where that cliff is . . . she seems OK but they tell us that they can suddenly drop off a cliff and suddenly get really bad.

As their child entered and moved through the educational system, parents were reliant on the actions of teaching staff to keep their child safe. Parents acknowledged how daunting it was for teachers to be in a situation where, as Harry’s mother explained, “You’ve got this child, if anything looks wrong get him to A&E. But there’s nothing you can do on.”

In almost all cases, the nurseries and schools had no prior knowledge or experience of MCADD and so parents had to work with teachers to ensure they had adequate understanding of their child’s condition. In some cases, they had received assistance from health professionals, particularly specialist nurses and dieticians, who acted as an ongoing source of information and visited schools and nurseries to deliver educational sessions. This assistance was highly valued, particularly the health professional’s ability to convey information in a balanced and nonemotive way as Ellie’s mother explained:

The metabolic nurse came into the playgroup, spoke to them . . . The way I had explained it compared to the way the nurse explained it, she did it so much better. I think she emphasized when they need to act and how quickly they need to act and, but also kept it quite simple and understandable which I probably ranted a bit.

The default position for parents and teachers was to err on the side of caution and keep the child at home at the first indication of illness to enable close surveillance and prompt intervention. Ellie’s mother described how “if she’s ever sick at school, then they’ve phoned us up straightaway . . . to pick her up” and Ian’s mother explained that although “he’s got a snack box at school and obviously the SOS sachets . . . if he’s got a bit of a cold or whatever, school will probably send him home if they’re a bit worried about him.” In some cases, schools and parents had adopted a wider preventive approach to limit exposure and thereby reduce the likelihood of illness events:

They [the school] rang me half nine in the morning, “you should be aware there’s four children away with tummy upsets, do you want Dan in or out?” And so he came out . . . Prophylactically removing him from school to make sure he doesn’t pick up any bugs . . . that’s worked reasonably well, they’ve been very good like that. (Dan’s father)

Discussion

This is the first study that examines in detail the experience of a group of parents with an MCADD child. The challenges that these parents identified concerned two key aspects of managing the condition: first, those related to day-to-day dietary management including protection of the safe fasting interval, and second, those relating to illness events. Collectively, they indicate the likely basis for family burden reported by Gramer et al. (2014) and while our findings would suggest that the level of burden reported in that study

is likely to diminish for parents of a child with MCADD as their child grows older, they also identify the ongoing concerns for parents posed by the potential threat of metabolic crisis.

Parents described a range of condition management strategies and a set of concerns and anxieties which changed as the child grew older. The first year of life, with the demands of frequent feeding and ongoing anxieties about the quantity of food their child was taking, was particularly difficult for parents. Parental anxieties about the vulnerability of newborns and fears for their survival are acknowledged among the parents of healthy children (Lupton, 2011), and are particularly associated with specific conditions including prematurity and chronic childhood illness where they contribute adversely to social adjustment (Anthony, Gil, & Schanberg, 2003; Perrin, West, & Culley, 1989). For the parents of MCADD children, these anxieties about infant vulnerability were ever-present because of the immediacy of the connection between food and survival, and particularly apparent at the transition points over the first year when feeding intervals were extended. Although from a medical perspective, this staged increase in length of fasting represents safe practice grounded in robust scientific evidence, to the parents it represents periods of danger when they were being asked to simply increase the amount of time between feeds and trust that their baby will survive.

Parents used a range of measures designed to protect the safe fasting interval: extended use of and ready reversion to infant feeding patterns in young children, biscuits beside the bed ready to be nibbled as soon as the child awoke, and supper before bed in the older children. The day-to-day dietary management regimes that parents imposed were commonly more stringent than those in the national guidelines and collectively reflect a risk-averse approach, which parents readily acknowledged. Timmermans and Buchbinder (2013) reported similar approaches to feeding regimes in their case study of Kari and her mother, which they understand in terms of a parent who sees her child continuously at risk of metabolic crisis and requiring constant vigilance. Our findings resonate with this analysis. Feeding management decisions were designed not only to protect the fasting interval from the vagaries of individual eating and sleeping patterns but also to protect parents from their own persistent fears of finding that their child had slipped into a coma overnight.

Parents adopted a similar risk-averse approach to illness events. The medical perspective understands MCADD as a medical vulnerability that matters most when a child is fasting due to illness (Timmermans & Buchbinder, 2013), and the emergency regimen is the way in which that vulnerability is managed. Although this might seem relatively straightforward, the situation is more complex and problematic from a parental perspective. They had to manage considerable uncertainties about that period of vulnerability: where, when, and how an illness event begins and at what rate it might be progressing and taking the child toward what Jenny's mother

described as "the edge of the cliff." They did so by keeping their child away from school when others were ill to reduce exposure to infection, and removing their child from school at the first indication that they might be ill, to enable increased parental surveillance. These approaches reflect the parents' anxieties about the capability of others to act appropriately around illness events. The delays and difficulties in accessing treatment were particularly distressing for parents and the onus of responsibility that they experienced when faced with staff who appeared to have no knowledge of the condition was daunting. This indicates the need for greater awareness of the condition among emergency care providers.

Parents speculated on their concerns about the safety of their child in adolescence and early adulthood, concerns which were largely related to alcohol intake. Similar concerns have been identified by parents and clinicians (Schatz & Ensenauer, 2010; Torkelson & Trahms, 2010) and are legitimized by clinical evidence that indicates adult presentations of previously undiagnosed MCADD are commonly precipitated by fasting and alcohol consumption (Lang, 2009). The number of children living in England with MCADD will increase year on year as a result of NBS and strategies will be required to help keep them safe. The use of medic alert bracelets from adolescence onward to help facilitate prompt emergency care has been suggested (Jameson & Walter, 2015), but the effectiveness of these rely on the actions of others and indicate the need for a greater level of awareness of the condition both within social groups and at a societal level.

Poor adherence to medication among adolescents which is associated with increasing independence and agential control is a recognized problem in managing chronic diseases such as asthma (Peterson-Sweeney, McMullen, Yoos, & Kitzman, 2003). These problems are unlikely to be as great for those with MCADD because a drug regime is not part of the management. However, adolescence may be associated with problems of adhering to dietary management requirements which impact on long-term health and well-being as well as the ever-present possibility of a metabolic crisis. Consequently, we might anticipate significant difficulties for the child and their family during adolescence. This indicates the need for ongoing work to understand the issues they face and the support they require to enable these children to transition safely and successfully into adulthood.

Strengths and Limitations

There are a number of strengths and limitations to this study. Recruitment was from two hospital sites within one regional specialist metabolic unit which enabled us to include a substantial number of parents. This is a particular challenge for rare conditions and increases the relevance of these findings to other areas where NBS for MCADD is established. However, it is important to acknowledge that the families in this study received specialist input into their condition which might not

be available to those who do not have access to a metabolic specialist center, either in England or elsewhere in the world. The extent to which these parents' experiences reflect those of parents who have not received ongoing specialist input is unknown. The rarity of the condition limited the sample size. While we are confident that the themes capture the realm of experience, additional interviews would have expanded the degree of variability within that experience.

The study included parents with children of different ages including those who were among the first to be diagnosed through NBS in England. This enabled us to provide a longitudinal perspective of the ways in which parental experiences change over time. However, it is important to recognize that the experiences of those diagnosed in the early days of the program might differ to some extent from those diagnosed more recently as the number of children with MCADD increase and the experience of managing them develops.

Conclusion

The management requirements of MCADD present parents with substantial challenges and are associated with concerns and anxieties that change in nature as their child grows older. Early detection of MCADD through the NBS program enables good health outcomes, but achieving these is dependent on effective management by parents initially and then by the child as they grow older. To ensure those outcomes, parents adopt management strategies which are characterized by a high level of caution and vigilance which is influenced by the rarity of the condition, the lack of awareness in the general population, and a lack of confidence in others to manage the condition. There is a need for increased awareness of the condition, particularly in relation to emergency treatment, to support parents in managing the condition and ensure the continued good health outcomes in the increasing numbers living with MCADD diagnosed at birth as they grow older.

Authors' Note

The views and opinions expressed are those of the author(s), and not necessarily those of the NHS, the National Institute for Health Research (NIHR), or the Department of Health (DoH).

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References

- Anthony, K. K., Gil, K. M., & Schanberg, L. E. (2003). Brief report: Parental perceptions of child vulnerability in children with chronic illness. *Journal of Pediatric Psychology, 28*, 185–190.
- Braun, V., & Clarke, V. (2006). Using thematic analysis in psychology. *Qualitative Research in Psychology, 3*, 77–101.
- British Inherited Metabolic Diseases Group. (2016). *MCADD dietetic management guidelines*. Retrieved from <http://www.bimdg.org.uk/site/guidelines-enbs.asp?t=6>
- Buchbinder, M., & Timmermans, S. (2012). Newborn screening for metabolic disorders: Parental perceptions of the initial communication of results. *Clinical Pediatrics, 51*, 739–744.
- Cresswell, J. (2008). *Research design: Qualitative, quantitative, and mixed methods approaches* (3rd ed.). Thousand Oaks, CA: Sage.
- Gramer, G., Haege, G., Glahn, E. M., Hoffmann, G. F., Lindner, M., & Burgard, P. (2014). Living with an inborn error of metabolism detected by newborn screening—Parents' perspectives on child development and impact on family life. *Journal of Inherited Metabolic Disease, 37*, 189–195.
- Grosse, S. D., Khoury, M. J., Greene, C. L., Crider, K. S., & Pollitt, R. J. (2006). The epidemiology of medium chain acyl-CoA dehydrogenase deficiency: An update. *Genetics in Medicine, 8*, 205–212.
- Jameson, E., & Walter, J. H. (2015). Medium-chain acyl-CoA dehydrogenase deficiency. *Paediatrics and Child Health, 25*, 145–148.
- Joy, P., Black, C., Rocca, A., Haas, M., & Wilcken, B. (2009). Neuropsychological functioning in children with medium chain acyl coenzyme a dehydrogenase deficiency (MCADD): The impact of early diagnosis and screening on outcome. *Child Neuropsychology: A Journal on Normal and Abnormal Development in Childhood and Adolescence, 15*, 8–20. doi:10.1080/09297040701864570
- Lang, T. F. (2009). Adult presentations of medium-chain acyl-CoA dehydrogenase deficiency (MCADD). *Journal of Inherited Metabolic Disease, 32*, 675–683.
- Leonard, J. V., & Dezateux, C. (2009). Newborn screening for medium chain acyl-CoA dehydrogenase deficiency. *Archives of Disease in Childhood, 94*, 235–238. doi:10.1136/adc.2007.134957
- Lindner, M., Gramer, G., Haege, G., Fang-Hoffmann, J., Schwab, K. O., Tacke, U., . . . Leichenring, M. (2011). Efficacy and outcome of expanded newborn screening for metabolic diseases—Report of 10 years from South-West Germany. *Orphanet Journal of Rare Diseases, 6*(1), 44.
- Lupton, D. A. (2011). “The best thing for the baby”: Mothers' concepts and experiences related to promoting their infants' health and development. *Health, Risk & Society, 13*, 637–651.
- Nennstiel-Ratzel, U., Arenz, S., Maier, E. M., Knerr, I., Baumkötter, J., Röschinger, W., . . . von Kries, R. (2005). Reduced incidence of severe metabolic crisis or death in children with medium chain acyl-CoA dehydrogenase deficiency homozygous for c. 985A> G identified by neonatal screening. *Molecular Genetics and Metabolism, 85*, 157–159.

- Oerton, J., Khalid, J. M., Besley, G., Dalton, R. N., Downing, M., Green, A., . . . Dezateux, C. (2010). Newborn screening for medium chain acyl-CoA dehydrogenase deficiency in England: Prevalence, predictive value and test validity based on 1.5 million screened babies. *Journal of Medical Screening*, *18*, 173–181. doi:10.1258/jms.2011.011086
- Perrin, E. C., West, P. D., & Culley, B. S. (1989). Is my child normal yet? Correlates of vulnerability. *Pediatrics*, *83*, 355–363.
- Peterson-Sweeney, K., McMullen, A., Yoos, H. L., & Kitzman, H. (2003). Parental perceptions of their child's asthma: Management and medication use. *Journal of Pediatric Health Care*, *17*, 118–125.
- Schatz, U. A., & Ensenuer, R. (2010). The clinical manifestation of MCAD deficiency: Challenges towards adulthood in the screened population. *Journal of Inherited Metabolic Disease*, *33*, 513–520.
- Therrell, B. L., Padilla, C. D., Loeber, J. G., Kneisser, I., Saadallah, A., Borrajo, G. J., . . . Adams, J. (2015). Current status of newborn screening worldwide: 2015. *Seminars in Perinatology*, *39*, 171–187.
- Timmermans, S., & Buchbinder, M. (2013). *Saving babies? The consequences of newborn screening*. Chicago: The University of Chicago Press.
- Torkelson, R., & Trahms, C. M. (2010). Assessment of parenting stress related to treatment and management of a child with

medium chain acyl-CoA dehydrogenase deficiency identified by newborn screening. *ICAN: Infant, Child, & Adolescent Nutrition*, *2*, 181–186.

- Wilcken, B., Haas, M., Joy, P., Wiley, V., Bowling, F., Carpenter, K., . . . Boneh, A. (2009). Expanded newborn screening: Outcome in screened and unscreened patients at age 6 years. *Pediatrics*, *124*, e241–248. doi:10.1542/peds.2008-0586

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