Thoughts into Action

What is quality of life as defined by people living with inherited metabolic disorders?

Contents

If this report brings up anything you'd like to talk about, you can call Metabolic Support UK at 0845 241 2173 or email us at contact@metabolicsupportuk.org

Foreword: Elin Haf Davies, Chair of MSUK

Metabolic Support UK (MSUK) have been supporting people living with Inherited Metabolic Disorders (IMD), and their communities, since 1981.

We are an organisation that strives to be led by the needs of our community, supported by evidence and data. Since the COVID-19 pandemic, our digital adoption has enabled an increased reach to people living with an IMD.
Despite being physically apart, it feels that we are now closer as a community, with a new sense of unity and togetherness. Thoughts into Action is a manifestation of that trust and collaboration; asking the people we represent what matters to them and then acting on it.

This unique research project sets out to determine what quality of life is as defined by people who live with an IMD. It puts our community at the heart of our work; terms such as patient centricity and patient engagement are now not only mainstream but becoming mandatory.

MSUK's 2030 strategy establishes individual support, building communities, empowerment and advocacy for the IMD community in an inter-linked approach. This requires open, transparent collaboration and excellence at every juncture.
Our future success will be determined by having a true understanding of the factors that our community feel impacts their everyday quality of life. This understanding provides us with the knowledge to question how effectively current systems help them to achieve a good quality of life. We want this work to deliver meaningful change that goes beyond conversations and written recommendations.

I would like to thank the people who took the time to speak to us and to share their stories. It is a privilege to listen and be able to take action.

CEO view: Kirsty Hoyle

In 2021, I moved from CEO of a disability rights organisation to a rare disease organisation and was immediately struck by the difference in the images, narratives and schemes of work that surround our rare community, despite the similarities of their experiences.

People living with a rare disease are bombarded with literature that talks of the conditions they live with as having intolerable burden or devastating impact.

Does this 'devastation' narrative genuinely support people living with a rare disease to live well? And do we, as a patient organisation, do enough to explore what living well with a rare disease looks like?

I believe our role as a patient organisation is to reflect the self-reported needs of our community and to develop services and a funding model that supports this. Thoughts into Action is an integral part of that process

For this research we have resisted the pressure to provide statistics that may tell us how many people identify with an issue - but not why or how the issue arises. Knowing that, for example, '7 out of 10 people with an IMD feel lonely', may be useful for campaigning but does not reflect the nature of the human experience.

As a rare disease community, we want to challenge the quantitative bias; an issue is not only valuable should a certain percentage of people be affected by it. Our grounded theory approach instead combines the stories we were told into valuable insight that will help us to support our community to live well with an IMD.

We hope you will read and respond to this story that shares the lives of our community as they live them: complex, interconnected and deeply personal.

Introduction

At MSUK we wanted to know what matters most to people living with an IMD and what factors they feel impact on their everyday quality of life. As a result, we decided to ask our community to tell us in their own words in a research project called Thoughts into Action.

Thoughts into Action is designed to centre our community's views genuinely and meaningfully within a landscape that currently relies heavily on the views of the services designed to support them. It is a 12-month research project that aims to answer one key question that will inform our work as an organisation going forward; What is quality of life as defined by the community of people living with an Inherited Metabolic Disorder (IMD)?

Quality of life (QOL) is a term used widely in the healthcare sector and is also quantified when used as a measure to make decisions on treatments and healthcare services. We want to know what our community believes it is – not what they are told it is.

Should we be guided by measurements of health-related quality of life (HRQoL) like EQ-5D, which is used to decide if a medicine will make enough of a difference to someone's health to justify the cost of it to our NHS? Do we use the benefits assessment process, which decides if someone is eligible for specific support? Should we be guided by those in the healthcare system delivering the care and services needed to support the IMD community? Or should we be guided by the person living with an IMD, who struggles to meet a friend for dinner without worrying that the restaurant will not have anything on the menu that is suitable for their needs? How much do we value the lived experience of a parent who knows there is something not quite right with their child but feel they are not being believed?

QOL is not simply impacted by healthcare, although when living with an IMD it is undoubtedly a significant contributor. A search of available data about living with an IMD returned very few reports about its impact on daily life experiences. This is concerning to MSUK. In the current environment, where licensed disease modifying therapies exist for only 5% of rare diseases, we must understand how the challenges of life impact on our community.

How does a person with an IMD live well when no treatments are available or even on the horizon for them? What does that look like and what do they need to achieve this?

Inherited metabolic disorders and rare disease

The definition of a rare disease varies depending where in the world you live. In the United Kingdom (UK), the European definition of 1 in 2,000 people has been adopted (1) to direct policies, services and support to this defined population. There are more than 6,000 different rare diseases recorded in the European Orphanet database (2) affecting as many as 1 in 17 people living in the UK (3). IMDs are often classed as rare, with figures suggesting that an IMD will be present in as few as 1 in 800–1 in 5,000 newborns across all disorders (4). IMDs are a large class of rare genetic diseases, with data suggesting there are over 500 different IMDs (5,6) They are the result of an enzyme or hormone deficiency affecting the metabolic pathway.

Like other rare diseases, it is common for people with an IMD to experience long delays in their ultimate diagnosis. This delay is contributed largely to the nature of the symptoms of IMDs which can be associated with more common conditions and may not be very specific (7). For a small number of IMDs, the condition can be identified through newborn screening in the UK (8). However, many individuals are not diagnosed until their adolescent years or later in adulthood, due to the fact that, in most cases, the initial symptoms of IMDs are unspecific and common to other more common diseases (7).

This delay may be exacerbated by the lack of specialist knowledge of rare diseases amongst healthcare professionals (9). In 2016, an NHS report indicated that there were just 15 consultants specialising in metabolic medicine across the whole of the UK (10). This is a worrying trend with more recent figures from research in 2023 highlighting the lack of specialist services and suggesting that only half of those living with an IMD in England and Wales have access to a specialist in their IMD (11). A diagnosis of an IMD can bring very difficult challenges; cognitive impairment, brain damage and life-threatening comas are some of the complications associated with the six IMDs included on the UK newborn screening panel (12). This is further confounded by the lack of disease modifying medicines or ‘cures’ available for the majority (13). At MSUK, we want to know what specifically can improve the lives of those living with a rare disease. Knowing this will allow us to do purposeful work on behalf of our IMD community. We are the only patient organisation offering support to all 20,000+ people living with one of over 500 IMDs in the UK.

The UK Rare Disease Framework

The UK Rare Disease Framework is a policy designed specifically to "deliver better health outcomes and improve the lives of those living with a rare disease". It has highlighted priority areas that each UK nation must consider within their implementation plan (14).

The policy landscape for rare diseases is dominated by this framework. Each of the priorities within the UK Rare Disease Framework (RDF) highlights the need to deliver improvements in the health outcomes for communities like ours. However, there is minimal focus on how their lives beyond healthcare can be improved. Similarly, the measurements commonly used to determine access to medicines and treatments are often grounded in health. Health-related quality of life measurement (HRQoL) is often a generic measurement, which has been shown in other progressive rare diseases to lack specificity, failing to capture many of the factors important to everyday life (15). These include factors reported as important to the IMD community including accessing support with education, employment and job opportunities, social care and financial assistance (16).

If the priorities of the RDF deliver against the policy intent of "improving health outcomes", this will indeed be a positive step forward for people living with an IMD and the wider rare disease community. If its intent is realised, people would see a more streamlined healthcare system, where they; receive an early diagnosis, engage with healthcare professionals who understand the difficulties of rare conditions, are managed by a multi-disciplinary team who adopt a coordinated approach to the delivery of their care. If they are very lucky their condition may be one of the 5% of rare diseases with an approved disease modifying therapy that they can access on the NHS.

However, it is not clear how it will improve the lives of those living with a rare disease if it fails to drive forward specific actions that address their wider needs. Will it have an impact on the additional support services that people from the IMD community rely on? How would any impact be measured outside of a healthcare setting?

Thoughts into Action: Research methodology

MSUK are interested in moving beyond statistic-based research that tells us how many people share a certain experience. Instead, we wish to understand why these people have that shared experience. For this reason, we chose to apply a qualitative approach to our research, using research conversations that were led by the participants.

A research method known as grounded theory (GT) was applied in the study. GT sets out to generate a theory. This theory is set in the data that has been collected and interpreted to understand the relationships and behaviours of specific groups of people. It aims to understand the main concern of a particular population and how they manage and deal with that concern. This method was applied to generate an understanding of the elements contributing to the daily lived experience of the IMD community, specifically within the framework of their perception of quality of life. It allows people to share what they are comfortable sharing, and what they feel is relevant.

Participants were recruited using a self-referral process in response to targeted emails and a social media campaign, supported by organisations from across the wider rare disease community. As part of the campaign, participants were asked to self-refer if they live with an IMD or if they care for someone living with an IMD whose communication needs they also support. This broad approach was used to limit any potentially biased assumptions about participants, while acknowledging that participants are unlikely to be representative of the full IMD community. Participants who indicated willingness to participate were asked to sign a consent form. Once consent forms were received, an invitation to participate in a research conversation was issued. The research conversations took place between April 2023 and June 2023 and were held virtually on the Zoom platform.

What is a Research Conversation?

Research conversations were conversations between one of MSUK's researchers and the participant. Research conversations followed a pre-determined introduction, followed by consent questions. All research conversations were recorded for analysis. Each research conversation started with the same conversation seed "I am very keen to learn about your everyday life and about what matters to you. I do not have a list of questions that we have to go through and there are no right or wrong answers.". Subsequently, conversations were led by the participant. Additional conversation seeds were pre-determined in case conversations stalled. At the end of the research conversation, the recorder was switched off and participants were thanked for their time and effort and asked about their experience. Safe-guarding measures were in place in case patients were distressed or disclosed concerning information.

All conversations were transcribed and de-identified. Each transcript was read and coded by the grounded theory researcher. A 'code' or label summarising a topic was assigned to each key topic discussed by the participants and recorded using the Quirkos(TM) analytic software. Codes developed as more conversations were analysed and were grouped to form concepts. Subsequently, broad groups of concepts formed categories, which were used as the basis for the theory that was generated. In line with GT methods, the different activities including the conversations and analyses were conducted "sequentially, simultaneously, serendipitously and scheduled" (17).

The final theory is based on data analysed and coded from 57 participants, representing over 30 different IMDs who accessed services and support across all four nations of the UK.

Participant breakdown

Parent or carer of person living with IMD: **31**
Child under 16 years of age living with IMD: **2**
Person living with IMD (16 years of age or over): **24**

Limitations of the research

We recognise there are a number of limitations associated with our recruitment method for the research. These include the self-referral recruitment method for the research, which may have excluded certain groups; the use of the virtual platform, Zoom, to conduct the research conversations; our decision to not collect data on protected characteristics, and the inclusion of participants from the four UK nations.

Ethical approval

The research received a positive ethics opinion from the University of Portsmouth Ethics reference #: 23/ETHICS/001 on the 31st of March 2023.

Taking directed action in different contexts of belief: A grounded theory

"I think that's where people need to understand in a little bit more depth, that it's not just the medical side of things that people with these disorders [are affected by], it's their day-to-day lives that are being affected. It's the social side as well." (TiA Participant, 2023)

It may seem obvious that people in everyday life will take actions to avoid harm. In fact, Hippocrates' famous quote of "First do No Harm" has been a guiding principle in medical ethics for centuries. However, imagine living your everyday life assessing social engagements, hospital visits, commuting to work, feeding your family and many more of the activities that most of us take for granted for their risk of harm because others fail to understand or believe the risks you face. This is the reality the IMD community live with.

The theory derived through our research is based on the shared and important concern of people living with an IMD: the fear of falling through a gap and coming to psychological or physical harm as a result of this fall. All people living with an IMD have a carefully woven 'net' of structures that nurture, protect, develop, and sustain them.

People with an IMD are not unique in having a 'net'. We are all supported in life by people, systems and structures that allow us to live. We go to school or work, have friendships, receive healthcare and we access both financial and other support from the state at various points in our lives. These structures are what enable people to live a fulfilling life and they form a metaphorical 'net' supporting people living with an IMD, as identified within our research.

Each person living with an IMD has a personal net, which consists of several sections. Nets are woven carefully from the moment symptoms of an IMD start to occur. Creating and maintaining a 'net' can be emotionally and physically exhausting and is an ongoing process throughout the life of a person living with an IMD. If symptoms occur before adulthood, parents and/or carers take responsibility in creating and maintaining a 'net', which in most cases is slowly transitioned to the person living with an IMD, as they grow older.

In this report, we use the metaphor of a 'net' to explain the structures that nurture, protect, develop and sustain those living with an IMD.

To overcome challenges and to attempt to live as 'normal' a life as possible - with the strongest support 'net' possible - people living with IMDs or those who provide their care, will take very specific and directed actions. These actions will be dependent upon the individual's level of physical capacity, their own capacity to understand, and the understanding and belief of others within the structures of their 'net'.

These three aspects can be part of the person living with an IMD in various ways. As a result, the research theory derived in our study suggests that the IMD community can be defined as specific groups or 'typologies'; **Managing, Fitting In, Routing to Live, Being Held** and **Hidden People**. The progression and management of a person's IMD means that is possible for them to transition between groups at different stages of life, depending on how their IMD affects their physical capacity, their capacity to understand and the belief of others within their 'net'.

Typologies

People with higher levels of physical capacity and capacity to understand are more likely to be **'managing'** and are described as having a stronger 'net'. They are often in their late teens or are adults and will have had their diagnosis for a while. People who are managing are complying with dietary guidance and adhere to medication schedules. Generally, they are likely to be in a settled period and, with the exception of their IMD, are considered healthy. Managing their IMD allows them to take holidays, work and socialise. Nonetheless, they will often still have to plan.
For example, one participant told us: "I think that's where people need to understand in a little bit more depth, that it's not just the medical side of things that people with these disorders [are affected by], it's their day-to-day lives that are being affected. It's the social side as well." (TiA Participant, 2023)

When people have an established net that works well it improves the quality of their life. Our participants shared stories where they are **'managing'** and therefore do not need to **'route'** as much as others. However, it is our reflection that participants who shared positive aspects of their quality of life more readily were those who have higher physical capacity and capacity to understand. We also reflect on the impact of upbringing and familial support; children inherit the net their parents establish.

One participant, an 11-year-old boy and his mum, shared their experience of a strong net supporting a good quality of life: "We've got a good team at [the hospital]. And I've got the direct email for a nutritionist … we have instant access to [the hospital]. We would phone them up and explain the situation and then we go straight into the children's ward where they prepare him for a drip pretty quickly. We have quite a positive outlook on life''. (TiA Participant, 2023)

Another participant explicitly recognised the importance of their upbringing and outlook of their parents: "I'm very privileged to have parents that kind of approached it that way...I've got no doubt they were worried sick at times [but] they've kind of pushed the boundaries so that I could go on and have a life [in which] I'm not constricted by my IMD. Although I deal with things safely, and make sure I'm not taking stupid risks that I don't need to take. But I'm aware that I'm really lucky to have had that." (TiA Participant, 2023).

Our research found that many children under the age of 18 were able to **'fit in'** due to the support and management their parents and carers provide on their behalf. Due to their age, they often have greater degrees of physical capacity, while having lower degrees of capacity to understand. If they grow up with a strong 'net' in place they may learn to manage their own IMD. Nonetheless, this often does come at an emotional and physical cost, with their parent(s) or carer(s) planning and **'routeing'** to make life as easy as possible for them.

One parent told us: "Things are as normal as they can be, there's an awful lot going on in the background. But for [our daughter], her life is normal. Like, my life, my husband's life is not normal in the management of her. But for [my daughter], it's as normal as it can be for a child her age. Until she's unwell." (TiA Participant, 2023)

People living with an IMD often end up in a position where they need to '**route to live'** due to a crisis or worsening of their (possibly undiagnosed) IMD. As a result of their IMD crisis or worsening, they have a lower physical capacity combined with a higher degree of capacity to understand **'route'** and plan to avoid gaps and to live. **'Routeing'** behaviour can involve adapting plans to conserve energy levels, planning a route to ensure access to disabled toilets or changing careers to support home working and medication regimens. Successful management of their IMD, effective treatment or an organ transplant may move these people from **'routeing'** to **'managing'.**

One participant shared: "So I was massively into horse riding. So I can't do that anymore. So again, it was like, those things stopped.…There's certain things I can do. I can go swimming; I can do stuff like that. But it's not the same. Not the same as galloping across the field." (TiA Participant, 2023)

Those **'being held'** are reliant on the support of others. Their parents or carers, healthcare, schools or social structures, such as social services, can be involved and have responsibility for keeping them safe from harm. Due to their reliance on others, these individuals will be particularly vulnerable to 'falling through their net' if their support system fails them.

They will experience life 'in the moment' without much thought or planning for their future.

One participant highlighted her past experience of 'being held': "I had a very hands-on mother, and I still do, who kind of took care of everything when I was younger. I think yeah, [I was] probably five when I really knew something's different, but it's taken me a long time to work out exactly what." (TiA participant, 2023)

For some people, their IMD may remain **'hidden'** and undiagnosed with minimal symptoms. They often **'manage'** their symptoms or **'fit in'**, not fully understanding the impact it has on them until they start to recognise their symptoms. Almost every conversation within our research featured a story of the long and difficult process of receiving a diagnosis and the lack of understanding and belief that participants faced from all corners of society about their symptoms and daily experiences. Once a diagnosis is confirmed, these previously **'hidden people'** may continue to **'manage'** or **'fit in'**, or may start **'routeing to live'**, as their diagnosis may have been the result of symptoms worsening.

Regardless of physical capacity and capacity to understand, the creation and management of a ‘net’ is dependent on many variables. In particular, the research has identified that having strong financial structures will help to create a net with stronger healthcare and social structures. It also found that knowledge of the different sources of information that help an individual to create, develop and restructure a ‘net’ is critical. Therefore, the security of an individual's ‘net’ and the actions they take when creating their ‘net’, will depend on their financial situation, their access to information and the understanding and belief of those who are essential to supporting their net.

A context of belief and disbelief

The context in which a ‘net’ is created or maintained has a high impact on the strength of the ‘net’. Our research found that the main contextual factors impacting the strength of the ‘net’ are belief and disbelief. Belief can be defined as an acceptance that something exists or is true, especially one without proof (18). On the other hand, disbelief is the inability or refusal to accept that something is true or real (19). For the IMD community, being believed means they can feel accepted and understood; they experience nurturing and supportive behaviour from others, and they feel heard. It makes the management of their IMD and any situations where ‘gaps’ arise within their ‘net’ much easier to manage.

One participant described the moment that led to her diagnosis, which helped her strengthen her ‘net’. This moment started in a context of disbelief and transitioned to a context of belief after the participant took directed action: "After many hospital appointments, I just broke down and said, ‘I should not be in the amount of pain I'm in for somebody who is of a younger age at that particular time’. And I just said, ‘I felt very un-listened to and very palmed off' by the particular doctor, I think at the time I said, ‘it just feels like you're reading another report from another doctor, you're not actually making your own decision based on what I'm presenting’. And they kind of took a step back and looked at me and said, ‘Okay, that's a very fair comment. We'll run some other tests.’ And if it hadn't been for that, I don't know if I'd have been diagnosed.” (TiA participant, 2023)

A context of belief or disbelief is directly linked to the level of understanding of others. Generally, in a context of belief, the people involved will have at least some level of understanding about the nature of the IMD, as well as what the current and future impact of the IMD has on the person living with the IMD. This results in actions which are nurturing and open, though potentially still uncertain. In contrast, in a context of disbelief little or no understanding of the nature of the IMD or the current and future impact of the IMD is present. This results in actions which are confrontational or sceptical, or a plain refusal to act.

What became clear through our research is that our IMD community are most vulnerable to the negative impact of disbelief and a lack of understanding when they are falling through a gap in their 'net'. They have experienced a variety of attitudes including ableism, intrusion, apathy and misunderstanding, which have resulted in behaviours including rejection, bullying, social smoothing and deprioritising.

Note-Medical Gaslighting:
Medical Gaslighting is a phrase being increasingly employed by those in the rare disease community to describe their experiences of being dismissed or disbelieved. Examples of this in practice were shared throughout our research.

Examples:
One participant described the moment that led to her diagnosis, which helped her strengthen her 'net'. This moment started in a context of disbelief and transitioned to a context of belief after the participant took directed action: "They've put it on my record that it's an eating disorder. So they automatically assume it's that when I've told them it isn't, it is this, but you give up telling them in the end. You get to a point where you just want to get better and get out of there. You just give up, there's no point arguing with them because you know they don't believe you anyway." (TiA Participant, 2023)

Another said: "I was pushing a buggy with my hands in agony. And doctors were saying, there's nothing wrong with you, because I was still using my hands. I kept saying, I don't really have a choice. I would be in bed with ice packs, hot water bottle, hands stretched out on two pillows. The doctor who did take it seriously rang me to apologise. I had mini fractures everywhere. They were in shock that that had been just so badly missed. It's very interesting seeing how, over the years, various specialists literally just look at what somebody else has written, and they don't listen to us at all as to what we present." (TiA Participant, 2023)

The types of attitudes described are unfortunately reflective of the experiences of many people who have reported their experiences of not being believed and even being accused of ‘faking’ their needs, particularly when their impairment may not be visible (20). In a recent research project, Challenging Disbelief and Disregard in the Lives of People Living with Chronic Illness, the issues and experiences of people who are living with chronic illness and energy limiting chronic illness was explored. The project identified similar experiences to those given in our research, with people describing disbelief, disregard and medical-gaslighting, which resulted in challenges accessing a diagnosis, treatment and social care, benefits and welfare support (21).

Throughout a person’s lifetime, they will experience many situations in which a context of disbelief will be present, some of which may have direct and irreversible consequences on their physical or psychological health, potentially resulting in death. The people tasked with providing the services and support that comprise an individual's ‘net’ need to believe them and support them, or, at the very least, not block their attempts to ‘strengthen their net’.

“We went to A&E about eight o'clock on the evening. And he spent all night in A&E. And they basically just said, ‘We've tested for everything, we've put him on antibiotics, he's not making improvement and we don't expect him to make it in the morning’. Basically we've got a wait for our baby to die. They said ‘his heart is just going down. He's gone into a coma and we don't know why’. At three o'clock in the morning, a student doctor comes in and said, ‘I know we've done all the tests [but] there's one more. So we can call out that we've literally done everything I'm going to do this test’ and it was a test for ammonia. And that one student doctor saved my son.” (TiA participant, 2023)

Living well with an IMD

Within this section of the report, we will explore the key areas, identified by the participants in our research, where gaps in the 'net' can impact on their everyday quality of life.

These six key areas are not conclusive. Instead, our focus reflects the areas identified in our research: **identity, employment and benefits, food, healthcare, transitions** and **mental health.**

We have also discussed access to treatments and clinical trials as a standalone issue that, whilst not identified as a 'gap', is an area of interest for our communities. We will explore the issues and opportunities in each area, using the experiences of our participants, and make recommendations to highlight where services and support can better enable our community to **'manage'** without the fear of falling. We hope to provide solutions that allow people living with an IMD to live well, now and in the future.

Identity: A lived experience

One participant described three issues that had impacted their confidence: lack of knowledge about their condition in medical services, being "wrapped up in cotton wool" by their family and how the "outside world viewed" them. They described how challenging these issues directly made positive change; when told by a healthcare professional (HCP) that they should not attend a music festival or drink alcohol they replied, "Is there any supporting evidence to say I can't do this?". When there was no evidence offered, they went ahead with their attendance, danced and had a drink at the festival and subsequently their HCP has been asking them to share how taking reasonable risks and having new experiences affects them in order to learn. They also believe that the resulting increase in personal confidence and more independence has had a direct impact on how others perceive them and said now that they "walk tall" other people are friendlier. For this participant widening and challenging the 'net' that had been designed for them has been the key to increased quality of life.

Identity: Rare disease and disability rights

“You don't have to look down at me. Like, I'm a human being. I'm still, you know, living and breathing. I'm just sick. Or you know, I have a medical issue. [That] doesn't make me any less of a human being” (TiA Participant, 2023)

There is a growing interest in the intersection between disability rights and the rights of those with rare diseases – and whether people living with a rare disease identify as disabled. Our research identified that there are different opinions in the IMD community as to whether living with a rare disease means also identifying as a disabled person or person with disabilities. This topic was explored during MSUK’s 2021 Rare Thinking Festival. The panellists discussed whether ‘considering oneself’ or ‘being labelled’ as disabled can contribute to being acknowledged for the need for support and the ability to connect with patient organisations and the wider rare disease community.

This reflects our grounded theory – if IMD communities have a positive and strong sense of identity and supportive networks, their ‘nets’ are more effective in achieving good quality of life. As evidenced in our research, many people are stymied in seeking adequate support by perceptions of ‘need’ and the onerous burden of bureaucracy that combine to delay or completely stop people applying for benefits or services that might support their quality of life.

Alongside individual identity, the views and understanding of people who form the structures of the ‘net’ are a key component to a good quality of life for people living with an IMD. One participant, who became unwell in their thirties, shared how other people’s attitudes stopped them from enjoying their passion of going to the cinema and the theatre. They shared “I find a lot of people look down on me. And they kind of feel sorry for me, and I don't want that”.

Another participant, who uses a mobility aid, was told, “you’re way too young for that”; one of many experiences that they feel “stops you from enjoying life”. It is not only the things that people say that can impact on how people feel about themselves; behaviours and actions have been reported as equally damaging. One participant explained, “it’s the way they look at you as well”.

In the UK, legislation in place to support rare disease communities is framed through 'disability' (22,23,24). Disability is one of the nine protected characteristics covered by the Equality Act (2010), which provides disabled people with measures to protect them from discrimination. Therefore, people living with IMDs who do not identify as having a disability or being disabled people may not access the full benefits associated with this protected characteristic. Rare disease specific policy does not cover this 'gap', the UK Rare Disease Framework does not consider social and financial needs within its priorities so ours is an underserved community.

Participants shared examples of how having positive societal interactions improves their outlook and quality of life, "It's really easy to fall in the trap of kind of excluding yourself and going 'oh I can't do that because I've got [an IMD].' But with a bit of forethought and planning you can find ways to still get enjoyment and get those experiences without necessarily putting yourself at risk. And that's something that's kind of, I suppose, came from being a kid. And being taught that actually, everybody's got something, everybody deals with things differently''.

Many adult participants living with an IMD live with their parents and have a carer or personal assistant to support their daily needs. Unfortunately, having a carer in attendance all the time led one participant in our research to feel "looked down upon" for their perceived lack of independence, which affected their confidence and quality of life. Another participant explained that they face prejudice assumptions about living with their parents, "I look after them money-wise, but you know, people just looked down on me."

This is not simply an issue faced by people living with IMDs or the wider rare community for that matter. The National Disability Strategy "makes a call to action" across society with plans to "build a national conversation about disability and a movement for change"(25). This strategy recognises the need to "transform the lives of all disabled people" . Changing societal views and 'levelling up' the opportunities for people with rare diseases - whether they consider themselves to be disabled or not - are crucial.

Without a change in attitude, people from the IMD community will continue to experience negative attitudes and discrimination in their everyday lives. The current UK Rare Disease Framework gives a nod to the fact that rare disease policy must align more closely with wider policy. If there is a strengthening of the protections of disability equality policies to include people more explicitly with a rare disease - this would be a good start.

Recommendations: Identity

The IMD community and the wider rare disease community may benefit from being more aware of the legislation that protects them and supports access to services, recognising that their own identity around disability may impact on this. Creating greater cross-sector engagement between organisations representing rare diseases, disability rights and long-term chronic conditions will strengthen the voice of communities who experience discrimination and disbelief from all sectors of society.

Note-Ableism:

Ableism emphasises discrimination in favour of non-disabled people (26). Although this language was not used by our participants, we want to encourage our community to be aware of language that can describe the things they experience to further their ability to advocate for their rights.

MSUK will:

- Design and deliver a 'Rare Disease & Disability Equality' Training Session in 2024 for the IMD and wider rare disease community
- Offer bespoke advocacy support to all IMD community members who suffer discrimination
- Launch our 'Living well with Rare Disease' campaign, focusing on everyday quality of life in partnership with other charities and organisations
- Amplify and promote the voices of 'living well' IMD influencers through our Youth Metabolic Advisory Council

We would like to see:

- People living with rare diseases to be considered as a specific group requiring consideration in Equality and Impact Assessments
- Specific reference to people living with rare diseases to be added to the Equality Act 2010 Equality and Human Rights Commission guidance
- Opportunities to engage and represent the IMD community in the research project Imagining Better Futures of Health and Social Care with and for People with Energy Limiting Chronic Illnesses
- Stakeholders representing mental health, long term conditions, disability rights and the wider rare disease community to advocate for an integrated approach to the development of policies

Guest contribution: David Rose

Accessing benefits and support: The impact of societal views and values around people living with a rare disease

I live with an ultra-rare disease, as well as several other disabilities. I am one of an estimated 20 people globally with my condition. It is hard living with a rare disease or other disability. Every day is a challenge in some way.

''I never know how exactly society perceives someone like me''

I have a real mix of invisible health aspects. As a result, I feel imposter syndrome all the time. I feel like people are judging me using a blue badge, the priority seats on public transport, or the special assistance at airports. Sometimes it feels like society still thinks you can only be disabled if you are a wheelchair user. Being ambulatory disabled like me can be very frustrating sometimes.

Imposter syndrome is felt even stronger when claiming disability benefits such as PIP, ESA, or Universal Credit. When I fill out the forms, it makes me feel like I am not as ill as I really am. The forms are not pleasant to complete and force me to be honest about how unwell I really am and what I really struggle with. It is rough! Society does not understand how much health can fluctuate and forms most definitely do not understand this. Plus, there is always the perceived scrounging image that comes with claiming benefits. Many forget that they probably claimed child benefit – that is a benefit too!

Separately, there is a huge information gap when it comes to applying for benefits. For some people, this is based on the many horror stories they have heard about disabled people being refused benefits that they should be entitled to. These stories put people off. In other cases, such as my parents, it comes down to being “in the know”. They never claimed child disability benefits for me. They never knew about getting a social worker. They never seemed to access some of the services available to them – because they are not always easy to find.

And even when you do receive benefits, which in some cases can be claimed alongside regular income from a job, it is often not that much money. Nonetheless, the benefit you do receive can be immensely helpful for the extra costs that come with being disabled. Especially in the current cost of living crisis.

Employment and benefits

‘’I don't live off the state. And even if I did, what's it to do with you?”

It is common for people living with an IMD to experience fatigue and pain, which can make it difficult to maintain a job without additional support. Our community shared many issues during our research, including examples of workplace bullying and employers attempting to terminate their employment.

In the UK it is against the law for employers to discriminate against disabled people; an employer must make ‘reasonable adjustments’ to avoid disabled people being put at a disadvantage compared to non-disabled people in the workplace (22) However, our participants described many situations where they either struggled to get these ‘reasonable adjustments’ or they were only granted on receipt of credentials confirming their IMD. These experiences are supported by the results of the MetabERN’s 2021 survey (7), which drew attention to the lack of flexible and suitable roles in the workplace where an IMD ‘resulted in a disability’.

One participant described feeling “*valued*” again in life on commencing a job within the healthcare sector, where flexible working was initially agreed with acknowledgment of the additional support, they would require performing their duties. Without a formal diagnosis, it was difficult to clearly identify the difficulties experienced and this led to stress and ill health.

When referred to the occupational health team, the lack of support led the participant to feel *“You've got no option, you've just got to give up. They weren't, they weren't prepared to change things for me, you know?”*

A lack of employment support sits on a background of financial pressures. The wider economic impact of living with an IMD can be witnessed through MSUK’s Cost of Living Report (March 2023) which found that living with, or caring for someone with, a rare disease like an IMD may affect the employment and income opportunities of family members (26). Over 50% of respondents have a household income significantly less than the median for the UK. Further to this, Scope states that life is already more expensive when you are disabled, with extra monthly costs of nearly £975 on average (27). This is exacerbated by a lack of flexible and suitable roles in the workplace, creating financial pressures in families who often require expensive but essential food supplements and dietary products (7).

Almost 82% of non-disabled people between the age of 16-64 were employed compared to just over 50% of disabled people in the same age group (28).

Disability Rights UK's disability employment charter has nine key asks of the Government, addressing the employment gaps, pay gaps, job satisfaction, and workplace wellbeing that have all been identified as issues facing disabled people (29).

Without income from employment, many of our participants described how they became more susceptible to the development of gaps in their 'net'.

Access to benefits and financial support are key issues for our IMD community. When asked about their experience accessing, or considering accessing, benefits in the UK system, respondents to MSUK's Cost of Living report said that, despite being eligible for benefits, many the process was "*lengthy*", "*stressful*" and "*startling*". Due to the length of these processes, and the high burden on those affected juggling work/caring/living with the condition, 35% of respondents did not claim benefits and those that did, did not receive their full entitlement. One respondent described ''*dreading*'' receiving their child's benefits renewal letter because it is ''*very in-depth, some of the questions are a bit strange and you've got to base the answers on your 'worst day' to be considered*'' (26).

Alongside the administrative burden and accessibility issues with accessing support, many participants are impacted by wider societal views on people who utilise the benefits system.

Describing the experience of applying for benefits, one participant told us:
"It is because of the type of questions that they asked. I'm not saying misleading, but they're very like 'can you dress yourself?'. Because it's such a borderline - or not quite this or not quite that - the question is sort of not quite appropriate. They didn't help you answer them. They look at that and really, they should look at the individual. It's not right to look at everybody the same, they should look at each individual case and deal with it accordingly". (TiA participant, 2023)

The benefits application process is lengthy and burdensome for everyone and particularly difficult to navigate as someone living or caring for someone with a rare disease. The process involves attempting to capture a vast spectrum of people and needs into standardised forms, as noted by one participant. They felt that *"[benefits assessors] don't fully understand IMDs at all, not at all"*. They found the forms to be

"geared around someone with a disability, as opposed to someone that has a disorder".

One participant discovered they were not eligible for benefits following the adult onset of their IMD, due to their previous financial position. Prior to their diagnosis they had a well-paying, physical job they loved, which allowed them to purchase a flat on mortgage. As their IMD progressed, impacting their physical health, the flat became unfeasible especially when combined with the drastic decrease in income from having to change roles. This meant they had to restructure their 'net' and move in with a partner. They were unable to access a service that should have formed part of their 'net' at a time when they were falling through a gap. Without any other options, this participant had to resort to crowdfunding to help restructure their 'net'.

Our participants feel that benefits assessment processes focus on needs related mainly to physical impairment and what qualifies as needing extra support, which completely ignores a whole section of difficulties faced by our community. It does not register the length of time, or the value of that time, to maintain food diaries, calculate the food for each meal, source the necessary ingredients, manage prescriptions and appointments, alongside learning, becoming an expert in your rare condition, and advocating for yourself or your child at medical appointments. This list of extra considerations that many take for granted, is not exhaustive. There are questions for the administration of medication overnight, but not for the administration of feeds and supplements overnight; a task that carries the exact same strains.

The structural and societal discrimination associated with, and the bureaucracy of, the UK benefits system, combined with whether the IMD community identify as disabled, combine to create gaps in the financial structures they rely on to support their 'net'.

Recommendations: Employment and benefits

Our research shows that access to support and funding and being able to do this without emotional burden or onerous administrative labour are key to good quality of life. Being valued as part of society regardless of economic or employment status is vital to the rare disease community. A better understanding of the social care sector

would benefit our communities; these complex administrative systems do not always work closely with the healthcare system and a high degree of personal motivation and knowledge can be required for success.

MSUK will:

- Highlight and promote our individual support service providing support with benefit claims, administrative forms, and employment discrimination
- Work with our Metabolic Advisory Council to co-produce resources that normalise discussions around money and financial support
- Promote a rights-based approach in our communications, reframing the narrative around benefits

We would like to see:

- Published guidance, co-produced with rare disease patients, for all Department of Work and Pensions personnel to alleviate burden of explaining, and increase belief for our communities
- A review of benefit claim forms to better reflect the needs and experiences of the rare disease community
- Employers explicitly state their support and protection for people living with rare diseases as part of their employment policies and procedures

Societal discrimination: A lived experience

One participant shared how words and actions of others made them feel "*ten times worse*". Whilst riding their mobility scooter, they almost collided with a stranger who shouted, "*get yourself a [expletive] job, trust one of you lot to almost hit me*". This interaction highlights an assumption that disabled people must be unemployed and, more importantly, that having paid employment is a marker of societal 'value'. In this case, the participant did work before the adult onset of their condition. When sharing this information, people have responded "*well at least you've done things*" which makes them feel as though others perceive their '*life to be over*'. ''*It's like they just view me as half, but because you're not capable to do something physical, which is terrible*."

Guest contribution: Stephen Kingdom, Disabled Children's Partnership

Looking through the wrong lens: The failure of social care for disabled children

"Social care" means different things to different people.

If you just follow the news, you could be forgiven for thinking it is just about older people; and even just getting them out of hospitals. But that ignores the crucial role that adult social care plays in supporting a whole range of people to live fulfilling, independent lives.

Similarly, if you asked a random person on the street what children's social care was, they would say it is about child abuse and children into care. The vital role that children's social care plays – or should be playing – to support disabled children and their families is rarely discussed.

Social care can enable disabled children, their siblings, and their parents to enjoy the same opportunities as other families. This social care can include, for example, support in the home; short breaks (respite) to enable disabled children to enjoy different activities whilst giving their parents a break from caring; and equipment or home adaptations. Sadly, however, this is not the reality that many parents face. Our Failed and Forgotten (30) report found that only one in seven families had the support they needed from social care.

Why is this the case? Almost inevitably, money is part of the issue. An economic analysis we commissioned found a pre-pandemic annual funding gap of a staggering £573 million (31). But it is also about how the system is set up, with assessment and support too often being seen through a child protection lens. This means that parents of disabled children find the system hostile and 'blaming' (32); and even where support is there, it is inappropriate for the family's circumstances or only provided at crisis point.

There are some positive signs on the horizon. Both the Independent Review of Children's Social Care (33) and the government's subsequent Long -Term Strategy for Children's Social Care (34) recognised the issues families face. And we were pleased that both took up our call for a Law Commission Review of the legal framework. However, without greater investment and greater priority, children and families will continue to struggle to get the support they deserve.

Read more about the Disabled Children's Partnership at: disabledchildrenspartnership.org.uk

Food for thought

One of the problems is sometimes with [my IMD], you don't feel truly human because you're so much different from everybody else. You go out. And then you see an ice cream van. And everybody has ice creams. And yeah, I can't have an ice cream." (TiA participant, 2023)

There is a popular narrative in society around food being medicine. For many within the IMD community food is actually medicine; it is a treatment essential in managing their condition. Despite the importance of food in supporting health needs, our community have highlighted issues in accessing the food that they need. These issues range from challenges in accessing appropriate menus when eating out to funding the highly specialised diets required to maintain their IMD. This sits on a background of a cost-of-living crisis and the impact an IMD can have on a family's income. Together these aspects directly impact the quality of life of our IMD community, causing them to fall through the net by being singled out in social situations and enduring financial hardship.

Despite the right to adequate food being recognised as a human right (35) access to appropriate food remains a challenge for many of our research participants. This subsequently led to participants '**routeing**' and '**fitting in**'.

When they found they were unable to '**route**' or '**fit in**' with their specialised diet, they often fell through gaps in their 'net'. The difficulties the IMD community face are compounded by the wider issues within the UK, with a recent report by Barnardo's suggesting that over half of families who have children under 18 in the UK have had to cut back on their food spending as a means of saving money. Recent years have seen significant increases in levels of malnutrition, hunger, and food bank usage against the backdrop of the cost-of-living crisis (36).

MSUK's 2023 *Cost of Living* report showed that the IMD community is not exempt from difficulties in accessing food. Over 85% of respondents stated that they had to maintain a specialised diet to manage their condition. Of these, more than 90% stated that they had been directly impacted by rising costs of groceries and food needed to maintain this diet. The report highlighted that these issues are not localised to those in their own home, but also affect those living in care:

"My daughter lives in a care home; we notice they use cheap sources of protein like cheese, bacon, sausages, and ham. She needs a low GI, cholesterol lowering, high protein diet, like lean meat, fish, pulses, fruit, and veg. Her seizures and behaviour are worse; we think she's suffered cognitive deterioration." Quote from MSUK Cost of Living Report (28)

Food for thought

These difficulties around food speak to a greater issue around who society is inviting in, who is deemed worthy of accommodation, change, and awareness.

An increase in time, planning, and routeing has been highlighted as a theme running through the whole 'net' for the IMD community. These demands on capacity can

mean dual-income households drop to single-income, whilst adults may face financial hardship through needing to reduce, or leave, employment.

These circumstances further compound the community's economic access to food. Some conditions are managed with low-protein diets, some with low-fat, some low-sugar etc. Each often relies on the popularity of "*fad*" or "*diet*" foods to meet their specialised dietary requirement. These foods, such as vegan options, are often higher in price. Relying on "*more expensive*" free-from options, "*assuming you can find [them] because they stop [them] quite regularly*", adds additional strain to finances and planning time. Factoring in how easily manufacturers can alter their recipes alongside this means those who rely on specialised diets to manage their condition may need to regularly check; labels; worrying about the potential loss of a favourite, or reliant food through changes that may seem inconsequential to others.

One participant shared their experience of the effort required to '**hold**' their child on a protein-restricted diet. Commercially available bread is not a viable option due to the protein content, so they rely on prescription bread; a provision intended to increase access to adequate food. However, to ensure access to bread, the family must remember to order at least two weeks before it is needed, accounting for returning the prescription and working with the singular manufacturing and delivery days per week.

Once delivered, the bread needs to be portioned and frozen "*because it goes off within two days*" and can only be ordered in a batch of four loaves. Despite thoroughly researching the IMD upon diagnosis, they were met with the unexpected cost of needing to purchase a second freezer to store this prescription food as "*bread alone, four loaves, is one and a half drawers*". This participant led with feeling "*very fortunate in [their] jobs and livelihood*" that they could afford to purchase an additional freezer and build a space in which to house it.

But what about people who are not that fortunate? Where is the accessibility and availability of adequate food for people who do not have the disposable income to meet these needs, or the time and energy to put towards the organisation required of these processes?

Another key aspect of good quality of life for our participants is the management of diet and the support to do so.

Dietitians play a major role in the management of many IMDs and for many patients are the main contact within the metabolic team, particularly at an early stage in diagnosis. Management itself is complex and requires precision and understanding to maintain safe levels of nutrients in the diet. Dietitians provide direct support including 'safe food lists' to assist in the development of simple recipes at home.

However, there are a lack of tools to assist people in other everyday tasks, such as calculating protein and nutrient content in supermarket or restaurant available foods.

One participant told us: "I'm trying to develop an app as well. […] Because I have, and some other people have, trouble working out sometimes how much protein is in certain foods. So, for example, they there's 2.2 grams of protein in 100 grams of McCain home fries. How much McCain home fries can I have for 4 grams of protein?" (TiA participant, 2023)

One participant shared how they '**held**' their child, *"she knew that some of the children might be having ice cream. But I obviously said you can't have an ice cream you can have an ice lolly. She was quite happy with that. As long as she's got something in place."* It requires confidence in teaching staff, their child's friends' parents, or family members to understand the IMDs requirements and the importance of following them.

There is a widespread misconception that certain IMDs are allergies and that they can be managed accordingly. As a result, eating out presents challenges, restaurants often label calories but not nutrition details on their food and staff are not trained to provide this.

One participant highlighted: "It's not a legal requirement in the UK to have nutritional information available for foods that are not pre-packaged. So if you find a place with them, you're doing well, and hope it's correct. There's a lot of places that don't even have menus available […] So you do not have any idea if she can eat anything there" (TiA participant, 2023)

Recommendations: Food for thought

When we look at the strict provisions in place to allow positive experiences for those with allergies, we must ask why this is not the same for rarer conditions with

alternative food requirements. This lack of provision and understanding, against a background of disbelief, means our IMD community must both do the work themselves in accessing information, or accept that they cannot join in.

MSUK will:

- Launch a 'Low Protein Challenge' – providing recipes, ideas and inspiration for our community
- Support our community in a social campaign designed to normalise their way of eating using social media and IMD diet specific imagery
- Provide clearer advice about accessing specialist dietary foods through our Individual Support work and our online advice hub.

We would like to see:

- The inclusion of medical dietary needs in assessment for Disabled Facilities Grants, administered through local councils
- Prescription food to be available free of charge beyond the age of 18 in England, akin to Wales, Scotland, and Northern Ireland services
- Mandatory protein content labelling in all public places serving food
- A clearer alert system to warn IMD communities when ingredients change on manufactured products

Note- Health warnings on food:
Health warnings on products are useful for people who are aware of their IMD and may be a helpful warning for those who are not. MSUK supports compulsory health warnings being added to protein shakes.

"Every time I went out to eat at a restaurant (we would ask for) plain pasta, don't put anything on it, just that. And then the restaurants come back and say, 'Oh, well, that sounds a bit boring. So we've put some oil on it, and some butter'. My dad would get really annoyed at them. I think I was quite aware that there was something different. And that there was all this going on." (TiA participant, 2023)

Healthcare

Gaining a formal diagnosis for a rare disease can be a long and painful process. For some people it can take up to nine years to secure a formal diagnosis (34), with

their journey fraught with misdiagnosis, fear, stress, anxiety, frustration and uncertainty and, ultimately, a need to become the expert in their own care (35).

Unfortunately, it is clear that all too often a diagnosis is only made on gaining access to a specialist who has knowledge and understanding about their particular condition (37,38,39). The participants in our research told us that they have experienced feeling "*broken and defeated*" before they had received a diagnosis, "*feeling desperate for information to understand the trajectory of their child's illness*" or receiving two pages printed from the internet as the only information and knowledge about their condition.

The importance of awareness, understanding and belief are the threads that links our theory together. How people live, fit in, route or are held are all impacted by the understanding, awareness and belief of those within the structures of their 'net'.

The European Commission made a commitment in 2008 to improve awareness and knowledge of rare diseases within Europe, identifying the need to improve the coding system to enable recognition of rare diseases within the service and create European networks to support the exchange of information (1).

These actions have undoubtedly gone some way to addressing the way in which specialist services record and share information about the morbidity and mortality of specific rare diseases across Europe. However, our participants have told us that this knowledge and awareness is not reaching the generalist healthcare professionals who they are reliant upon for their everyday care during and after their diagnosis.

One participant told us: "I went like, yeah, 'so I think I've got this [IMD]…... And this guy said '[This IMD] is so rare, of course, you don't have it'. Looked at the liver function tests and said, How much do you drink? I never drank an awful lot apart from one period of my life, about six months, when I was out with mates every night after qualifying but when I went to see him, I was saying, 'well, possibly half a bottle of cold lager on summer day'. 'So how much do you really drink?' asks this gentleman." (TiA participant, 2023)

MSUK are reassured that the UK Rare Disease Framework published in 2021 addresses this challenge directly as one of four priority areas (14). Each of the four Nations have produced action plans designed to address the issue, some with more tangible and measurable actions than others. MSUK are pleased to see the

announcement of the collaboration between Health Education England (HEE) and Medics4RareDiseases to create a rare disease resource hub to facilitate information sharing and training resources for healthcare professionals (14).

The current problem faced by the IMD community is twofold. Gaining access to specialists who understand their condition is difficult and appears to be due to limited numbers of IMD specialists available. This is then further confounded by the fact that those who provide the day-to-day care for people living with an IMD are general healthcare providers with little or no knowledge of the prognosis and clinical care required.

MSUK will address this by developing a full understanding of the IMD specialist support available in the UK, whilst supporting wider initiatives aimed at addressing the knowledge gaps in general medical care around rare diseases, seeking opportunities to improve education more specifically for IMDs.

The NHS and the dedicated healthcare professionals who serve our community are undoubtedly on their knees after the impact of the pandemic and the financial crisis that has followed, and we are sympathetic to the challenges of delivering education on rare diseases to an already stretched system. However, MSUK believes that our community need change now. It is not acceptable for our community to experience scrutiny and misunderstanding based on ignorance and lack of understanding.

A paramedic disclosed to us that they rely on 'Google' as their source of information when attending an emergency involving a person living with an IMD. We would like to see greater awareness of IMDs for all frontline healthcare professionals. One key issue for the IMD community is the timely undertaking of an ammonia test at times of crisis. This has been flagged both in our research and by members of the IMD community and healthcare professionals as a key concern.

Recommendations: Healthcare

As a patient organisation working with all stakeholders in this community, we understand that awareness and information about IMDs sits alongside the needs of

many other communities. We value our open and positive relationships with healthcare professionals and seek to collaboratively find ways to improve health outcomes, particularly around times of crisis and diagnosis.

MSUK will:

- Visit IMD clinics across the UK to promote our patient services and strengthen relationships with healthcare professionals
- Our IMD Information Hub, providing information on all IMDs will continue to be updated and expanded and publicly available
- Commission and deliver a full UK Service Review of the NHS Metabolic Services in 2024/5, providing a clear geographical picture of what is offered where, who is involved and what gaps there are
- Develop a resource for the IMD community to share with social/ healthcare organisations about the daily impact of living with an IMD
- MSUK will raise awareness of the Rare101 education module with our own IMD community and provide support materials they can use to raise awareness of this with the healthcare specialists who support their care
- MSUK will seek information on the plans to promote and evaluate the new rare disease education hub being rolled out as part of the 2023 England Rare Disease Action Plan and advocate that an impact assessment should be undertaken by the DoHSC Department of Health and Social Care to understand reach
- Launch an 'Ammonia Awareness' campaign with specific asks around ammonia testing awareness and training

We would like to see:

- Educational resources aimed at HCPs funded by, or partnering with, any UK public body, have mandatory evaluation processes reporting on engagement and impact evaluation as a condition of funding
- The adoption of Martha's Rule - a campaign asking for a rule giving patients the power to get an automatic second medical opinion about hospital care
- Ammonia Testing Guidelines (created by British Inherited Metabolic Disease Group) circulated and displayed with prominence in all emergency healthcare settings

Guest contribution: Dr Lucy McKay, CEO and Founder, Medics4RareDiseases

Practical rare disease training for healthcare professionals is essential

Medics4RareDiseases is a UK charity that provides and campaigns for education about rare disease for healthcare professionals. The ultimate aim is to reduce the time between first symptoms and receiving a final diagnosis and improve the healthcare experience for those living with rare and undiagnosed conditions.

Modern medical curriculums are packed full of information and, because of this, the stigmatising phrase, 'common things are common', is sadly universal rhetoric in medical training from the very beginning.

The M4RD Team are working hard to change this mindset and to empower medics with basic rare disease training, helping them to think outside the box, to think #DareToThinkRare. It would be impossible to redesign any healthcare curriculum to include over 7,000 rare diseases. Instead, what is needed is an increased awareness amongst healthcare professionals that that they should expect to care for patients living with rare conditions. Rare diseases are individually rare but people living with different rare conditions are common.

With clinicians on the Trustee Board, Staff Team, and alongside our Clinical Ambassadors, we understand the crushing challenges that NHS staff face on a daily basis. M4RD is here to support healthcare professionals and empower them with knowledge and tools to help care for and manage those patients with unusual and puzzling presentations, who take years to diagnose and are often lost navigating a confusing medical system of differing medical teams. In essence, to ensure people with rare conditions are looked after with clear management plans, treatment options, mental health support, opportunities for participation in studies, and the support of relevant patient advocacy groups.

M4RD has its own online training platform **M4RD Learn**. It provides a practical and flexible introduction to rare diseases for medics, free of charge. The patient experience is at the core of all of our training. M4RD is on the UK Rare Diseases Framework Forum and is working directly with the Welsh Implementation Group and NHS England, as well as with medical and foundation school, hospitals and primary care. In everything we do we use an unbiased approach and focus on common challenges so that rare disease is considered as a distinct discipline with its own unique profile of needs.

Read more about M4RD at: M4RD.org

Transitions

"You can't just pause having an IMD''

Participants told us that the risk of 'falling through the net' for people living with an IMD is heightened during times of transition, whether that be changes to employment, home life, education, or relationships. As one participant shared, *"You*

can't just pause having an IMD. And be like, 'Oh, well, sorry. I need to go and deal with this [transition] right now, so I can't have this.' It just continued on".

For many, growing up and working through transitions can bring a sense of independence. However, our participants reported extra considerations around healthcare transitions that place additional pressure during exciting events such as moving home or attending university. Where the understanding of the IMD decreases or the attitude of those providing the support changes, the risk to the person with an IMD of being 'dropped' increases.

Our research showed this shift in support and risk to be especially prominent when people living with an IMD start to reach 18 years of age and begin their healthcare transition from childhood to adult services. Until this point, young people have been 'held' by their parent(s) or carer(s), who have managed the issues arising from their IMD, creating a 'net' to support them.

Parents often liaise directly with professionals and develop a level of expertise to advocate for their child. They educate services and support staff on food and medicine needs, physical indicators of deterioration in wellbeing and the required actions to take. The transition to self-advocacy can be a rewarding experience for a young person, but it can also be daunting and require a level of emotional investment at a time when so many other parts of their 'net' require the same.

One participant described the anxiety of their child transitioning to adult care services despite being "*really prepared*": *"It's quite a trauma...because once you go to adult [services] some of the doctors won't let you in. You've been doing it for 18 years and then suddenly, you're not allowed in...it's not only a case of being in there on your own you don't have that same time. Appointments are much more condensed"* (TiA participant, 2023)

Transitions

A recent report published by the National Confidential Enquiry into Patient Outcome and Death (NCEPOD), concluded that clear pathways for children with long term, complex health conditions transitioning into adult services do not exist (40). The UK Rare Disease Framework gives the briefest of nods to the challenge of healthcare transition, with only the Welsh action plan producing tangible actions to improve this any tangible actions to improve this process (41).

Children often move from support under a specialist IMD service to management within a more generalist service without the specialist knowledge and understanding of their IMD (11), which can often be the result of a lack of specialists within the adult service (11, 42).

Different transitions were discussed by our participants: relationship breakdowns, new jobs, moving schools, aging, or leaving home. For each transition changes to the 'net' relate to increased risk of falling through other gaps.

"It's only now I'm starting to - kind of now everything else is a bit more settled - getting back on it with managing the condition. And seeing that as important again"

At these times, parents and carers may be excluded from appointments or hospital wards, leaving the young person feeling afraid and vulnerable if they have not received the appropriate help and support to prepare for these experiences (33).

Participants shared experiences of support being 'lost' as they moved into being classed as an adult; prescriptions for life-long medication and necessary prescription foods are no longer free.

One parent shared how the financial support they received to be able to travel with their child to her medical appointments was rescinded once their child became 18. These changes do not reflect the lived experience of the IMD community - despite the child reaching the age of 18 there were no changes to their support or financial needs. Yet they and their family must respond as though their circumstances have also changed because they have turned 18.

Transitions occur throughout a person's interactions with the healthcare system. Adults often experience regular change with one participant sharing: ***"Every six months...my consultants are changing; I got a text the other day saying she's moved to another country. So you're gonna have someone else. This doctor I've been seeing for five years, is gone. And a text is what I get."***

Recommendations: Transition

It is concerning to see that transitions are not considered an area of priority for policymakers, when it has been identified as a key time when people, and especially young people, with an IMD can 'fall through a gap in their net' and experience 'harm'.

It is also concerning to MSUK that there appears to be a disconnect between the delivery of specialist IMD services in children and adult services. As an organisation who supports the whole IMD community, we are compelled to understand how and where our community are engaging with specialist IMD services to ensure that we

can more effectively advocate for these where they may not exist. Our vision for transitions is that they no longer need specific attention, instead an individual's needs would be approached holistically, with a continuation of services that are not designated to be for "children" or ‘adults’.

MSUK will:

- Monitor and report the delivery against priority 3.1, which addresses transitions, within the Welsh action plan for rare diseases and its impact on the IMD community specifically
- Engage with policymakers from Scotland, Northern Ireland and England to understand why transitions are not a priority within their action plans and advocate that it should be
- Lead and deliver a UK IMD Service Review as outlined as part of the healthcare theme; this will have a specific focus on understanding and sharing the processes around transitions for UK metabolic patients"
- Engage actively in guidance and updates around transitions

What we would like to see:

- Continuation of services so each service currently only available for children has an adult counterpart with the long-term aim of combining services
- Published, tangible monitoring for any transitions guidance or action plans that measure against outcomes co-produced with a large patient community population
- The implementation of the ‘Ready, Steady, Go’ transitions framework for more young people living with an IMD

Guest contribution: Neil Fletcher, Roald Dahl Clinical Nurse Specialist for Teenagers and Young Adults, Barts Health NHS Trust

Healthcare transition pathways: What good should look like

For all children with a long-term condition, transition to adult care will at some point in their care process become relevant. Ideally, planning for a healthcare transition pathway is initiated by the age of 13 to ensure a smooth transition over the following three to four years. The transition should never be sudden or unexpected. Instead, it should be holistic and tailored to a young person’s individual needs, involving the multi-disciplinary teams (MDTs) involved in their care and accounting for condition-specific support and needs.

Throughout the process of planning and initiating the healthcare transition pathway, several aspects should be accounted for to help support a smooth transition. Similar to the paediatric services, patient-centricity is key. This means that the young person is more than their condition. Conversations around what matters to them should be encouraged, as part of which emotional and mental health and well-being is just as important a physical health. If they have a handheld or electronic health passport, the MDTs involved in their care and transition need to take time to look at it to try and fully understand the young person.

Siloed healthcare transition should be avoided. Many young people will have several disciplines involved in their care, such as primary and social care, but also education. In some cases, no recognised equivalent service may be available. Close communication and collaboration between these teams, the young person, and their families/carers will lead to an overall better experience in the transition pathway for all involved parties.

Finally, using a framework to discuss and outline the transition pathway encourages and empowers the young person and their families/carers. An example of such a framework is "ready, steady, go". This framework can be used to promote positivity around the transition, through for example the encouragement of joint appointments between children and adult services where possible whilst not making it daunting for the young person.

Roald Dahl Nurses work alongside the MDT to ensure that a holistic, individualised healthcare transition pathway is implemented and evaluated.

Read more about the Roald Dahl charity at: roalddahlcharity.org

Mental health in the IMD community

A lack of mental health support was highlighted by the participants within our research as a significant 'gap', impacting their quality of life. When participants described situations of 'falling through the gap' in their 'net', they often provided stories about "*traumatic*" diagnostic and treatment pathways or ongoing mental health issues either caused or exacerbated by living with an IMD. Although this gap in the net may be perceived to be related to healthcare, we recognise that medical, employment, and social needs cannot be considered in silo. They impact one another intrinsically.

The negative impact on mental health to individuals living with a rare disease is well documented (7,43,44). Our research shows that living with an IMD can cause stress

and other mental health problems and that it is in the intersections of a person's 'net' where people can become most susceptible. They have told us how this is exacerbated by a lack of adequate support.

One participant shared how gaps across their net ultimately led to the "lowest [they've] ever been" as they began creating a plan to take their own life: ***"...my life became work, pain, fatigue, you know, trying to do some life stuff, like, take your laundry, or do bank stuff or something. But it just, it was really bad. And it impacted my mental health And when it got really bad, I was at the point where I was planning my suicide because I was like, I just can't do it anymore. And I couldn't see a way out."*** **(TiA participant, 2023)**

Our research has highlighted many situations where participants are psychologically vulnerable, particularly within contexts of scepticism or disbelief about their IMD and its impact on their daily lives.

One participant described how he felt if he had been living with cancer, he would have been treated very differently: ***"I'm 34 years old, and I've only been offered any kind of counselling now. And I begged for it. Cancer patients get counselling straight away, it's part of their treatment, essentially."*** **(TiA Participant, 2023)**

Mental health in the IMD community

Our research suggests that how people restructure their 'net' to avoid further gaps created by poor mental health, appears to be dependent on the services, or lack thereof, in their area. It is also dependent on their financial abilities to access support privately, when no access is provided on the NHS, or waiting lists are long. When accessing mental health support, participants shared issues and frustrations around being understood by HCPs. They indicated that they felt the people who were supposed to give them support did not have adequate knowledge of IMDs or the impact of living with a rare disease.

One participant shared their one interaction with mental health support: "They sent once a mental health specialist to my bed. And they just talked about my

childhood. And I said, it's got nothing to do with my childhood, for God's sake. And then they left. … my child[hood] has got nothing to do with my medical problem. Yeah, no help. When I was literally having a breakdown in hospital, crying," (TiA Participant, 2023)

In contrast to interactions with HCPs, some participants talked about the positive impact of socialising with others who have an IMD or a similar lived experience, thereby understanding them more deeply. **Many spoke about the power of online communities, sharing knowledge, understanding, and a lived experience: "It can be a very lonely, stressful time. So the [online community] sometimes is just the only place where you can feel and just sit there and have a rant."**

Nonetheless, other participants who had the same condition, had different experiences of mental health and even everyday quality of life outside of that. This is similar to the findings reported in a recent report by National Voices, a leading coalition of health and social care charities. It highlighted that people with the same long-term conditions may have different mental health experiences. This is due to a number of factors, including the financial structures of their 'net'(45). This is a very worrying theme to emerge from our research, especially in the backdrop of data suggesting a higher rate of depression within communities most impacted by the cost-of-living crisis.

As a support organisation, MSUK receive an average of 23 mental health-related queries per month. This is unsurprising, considering a 2019 review of mental health services for people living with IMDs across England and Wales (49), found that current service provisions were failing to meet international guidelines, with inequality across those services that do exist.

Recommendations: Mental health

Given the significant challenge of improving mental health within the UK, it is unsurprising to find that specific policy does exist that will attempt to address the challenges our community face directly. The NHS Long Term Plan in England has committed an additional £2.3billion per year to improve mental health services in England by 2024 (50). There is also a focus within the action plans of each of the four nations on improving mental health for people living with rare diseases as part of their delivery against the UK Rare Disease Framework (41,48,49,50).

Each of the action plans recognises the interdependence of mental health and all aspects of an individual's life and suggests the need for wider policy alignment in achieving improvements. However, a recent report suggests that we have a long way

to go in ensuring mental health is given the same priority as physical health by healthcare professionals managing their care (42).

MSUK will:

- Monitor the direct impact of the UK Rare Disease Framework action plans on our IMD community through an annual evaluation survey
- Provide our team with ongoing professional development training on mental health and rare diseases
- Offer mental health training to all our community advocates and through our peer support scheme - Metabolic Connect
- Continue to develop our community-based provisions, both online and offline, to create accessible and supportive spaces
- Work alongside our Metabolic Advisory Council (MAC) to coproduce content on lesser spoken about aspects of living with an IMD
- Engage with National Voices and wider mental health charities to monitor the delivery of new mental health services promised within the NHS Long Term Plan in England.

What we would like to see:

- An online centralised mental health referral and support service specific to the IMD community
- A list of available private mental health care specialists with experience in rare disease that provides varied geographical, topical and economic options
- Emotional Support pathways included as standard arm of patient pathways
- Training delivered in Rare Disease for all NHS mental health support workers

Access to treatments and clinical trials

Although not identified in our research as one of the key 'gaps' in the 'net', access to and knowledge about treatments and clinical trials was something participants mentioned in connection to quality of life. They expressed a desire to be informed about treatments and trials, but found information was difficult to access and or understand.

This can be true even when the person living with an IMD, or their parent/carer, have a high level of understanding: ***'I'm a scientist and I love to know the research but it's so hard to find out what has been done. Sometimes things come up and you think ok, they've done this in this country. I'm sure other parents would want to***

know what other what researchers are doing. It'd be really nice to have a collective place to have (information). It's maybe very, very little, but there is stuff going on. Sometimes it isn't. I've had information in different languages, and I've just Google translated!'' **(TiA Participant, 2023)**

Better information about what is happening across the world in treatment development and clinical trials was a key issue in improving quality of life for our community: ***''My son has absolutely no faith in the health system. He tolerates the appointments to basically appease me. He just doesn't believe anyone's going to be able to help him. I think it really would be helpful (to know) where are the places around the world that people are doing things? And are there opportunities to be part of trials?''*** **(TiA Participant, 2023)**

It was important to our participants to highlight their sometimes-complex journey even when there is an approved treatment for their conditions or symptoms. This can impact quality of life and it is important that their 'net' offers continuous support throughout this journey. Participants reported on treatments not having the expected impact and what happens then.

One participant shows the importance of being prepared for negative outcomes shared about their child: ***''Unfortunately, she wasn't responsive (to medication). If she had responded she wouldn't need her substitutes or diet. But there's only about 30% of people who respond to it, unfortunately. It is what it is. But I'd already done some research on (a different IMD) as being the closest condition to it. And there's much more available there''*** **(TiA Participant, 2023)**

Whilst access to treatments was not identified as a key 'gap' in our research, knowledge and feeling informed about developments was important to them. Our research highlights the huge opportunity for organisations like MSUK to do more to get information about treatments and trials to IMD communities more quickly, and present this in a centralised location. With the continued work in drug repurposing having potential impact across diseases, knowing more about what is happening outside specific conditions can potentially be useful information for all of the IMD community. We would like to support our IMD community to be **'Research Ready'** and to develop an interest in contributing to research at all stages of development for individual and for wider community benefit.

Recommendations: Access to treatments and clinical trials

In a world where very few people living with IMDs have access to effective treatments it is vital that we campaign and advocate for greater investment, research and treatment approval. However, this needs to be balanced with the need to support quality of life needs as identified in this research and ensure we are doing enough to help our community live well right now. MSUK is committed to proportionally balancing both its services and funding model to better reflect these needs. There can be clusters of interest in particular disease areas - particularly with the rise of Advanced Therapy Medicinal Products such as gene therapy and, whilst we must and do support these advances, we intend to also keep energy and focus in other areas such as drug repurposing.

MSUK will:

- Launch a ‘Clinical Trial Hub’ an online hub providing a central place for independent information on what is happening, where, and for whom, for everyone living with an IMD
- Develop and deliver our ‘Research Ready’ programme which will inform and educate our IMD community on all aspects of clinical trials and treatment access
- Commission and undertake research into unmet needs in underserved IMD diseases

What we would like to see:

- Mandatory inclusion of patient advocates and/or patient organisations in the development of clinical trials
- Quality of Life measures that reflect the needs of the IMD community more specifically
- New guidance, co-produced with patient groups on funding from industry and declarations of interest

Guest contribution: Beacon for Rare Diseases

Drug Repurposing: An effective route to treatment in rare diseases

Drug repurposing, taking a drug that has been approved for use in one disease and using it in an entirely new condition, is increasingly gaining momentum and interest from rare disease researchers and patient groups. While gene therapies offer hope of curative treatment in the long-term; drug repurposing offers a quicker, cheaper, and more accessible route to treatments that slow disease progression or improve symptoms.

Repurposing a drug enables steps in the drug development pathway to be shortened or removed. Researchers already know the drug’s target, mechanism of action and it’s off-target effects and side effects. This means repurposed drugs can move quickly

into phase II/III clinical trials which, if successful, can see a faster route to regulatory approval and clinical use. New therapy trials have rigorous eligibility criteria, meaning many subsets of patients cannot participate. Pre-existing data relating to repurposed drugs means that eligibility criteria can be more flexible, lowering barriers to participation and creating more inclusive trials.

Drug repurposing often focuses on slowing disease progression or improving symptoms. Several successful repurposing projects have been led by clinicians or researchers in collaboration with patient groups. This collaboration ensures the drug targets symptom(s) that are most meaningful to patients and families. Success has been seen in EMA approval for use of nitisinone in alkaptonuria (AKU), a repurposed drug that slows the disease's progression. The project was driven by patient group, The AKU Society, in collaboration with an enthusiastic and committed group of researchers.

Using generic drugs for repurposing, those currently not under patent and can be manufactured and marketed by multiple companies, means they are cheaper to source. This creates a more attractive reimbursement model for healthcare systems than other treatments, particularly high-cost cell-based therapies. Despite this, getting repurposed drugs to market in this context can be challenging through lack of interest from pharmaceutical companies due to a seemingly weak business model with poor financial return. Projects like REMEDi4ALL bring together experts to address these issues and encourage collaborative discussions across stakeholders on why repurposing is a viable option for them.

While use of pre-existing drugs, that perhaps can even be purchased from your local supermarket, may not seem innovative, the drug repurposing journey sees the drug development pathway altered to bring more treatments to patients faster; surely doing something differently to improve outcomes is the very definition of innovation?

Final thoughts

The Thoughts into Action project was created with one key question in mind; What is quality of life as defined by the community of people living with an Inherited Metabolic Disorder? The lived experience shared in this research created the theory; "Taking directed action to live in different contexts of belief". Simply put, our community need to feel understood, believed and have access to services and support that will improve their everyday lived experience.

They are a community who, when not supported, can feel emotionally and physically exhausted from the effort of creating and maintaining a safety 'net', built from the social, financial and healthcare structures required to stay safe. They have identified systems and social structures that are inaccessible to their needs, dependent on the views of the people that hold power in those spaces. And importantly, that 'living

well with an IMD' is dependent on a combination of positive experiences across their lives - not simply from the management of their condition or the hope of an effective treatment.

In the current landscape, where only 5% of rare diseases have a disease-modifying treatment and very few have a 'cure', a great deal of work remains to change the narrative from despair and concern to how people can live well with an IMD when they have the right support. Our research tells us that we need to rebalance the focus on living with an IMD as 'life-limiting' and 'life-threatening' with no 'cure' and move towards a focus on living well with access to support that improves the everyday lived experience.

For MSUK this means a frank appraisal of our current work streams and a plan to balance and evaluate the areas in which we place our best efforts - both with and on behalf of the community. The reality for patient organisations is that our opportunities for funding can impact our work. Securing funding to support meaningful patient advocacy is not easy; in a context where we must rightly remain independent of all outside influences to be considered credible stakeholders, the opportunities for financial support are few. This is a conversation that needs to be had openly and repeatedly so that everyone working in rare disease can contribute to the solutions.

As we focus on supporting our community to 'live well with an IMD', MSUK intends to strengthen our understanding of the commonalities between the over 500 IMDs to increase our ability to provide a 'whole of government' approach to increasing quality of life.

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