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The **Lived Experience** of **Molybdenum Cofactor Deficiency Type A**

Funding was received from Sciensus BV to support this project

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BACKGROUND

Background

- **Molybdenum Cofactor Deficiency (MoCD)** is a **rare inherited metabolic disorder** with three forms: **type A, type B, and type C**. Although all types share similar symptoms, they are caused by **mutations in different genes** [1,2].
- MoCD type A is caused by a **mutation in the MOCS1 gene**, which is essential for producing a protein needed to create a molecule called molybdenum cofactor. This molecule is crucial for enabling enzymes (sulfite oxidase, xanthine oxidase, aldehyde dehydrogenase, and mARC) to **break down harmful substances that would otherwise become toxic [1,2].**
- The mutation in the MOCS1 gene means that the molybdenum cofactor cannot be produced properly, which **disrupts the function of these enzymes**. As a result, **toxic chemicals accumulate in the body [1,2].**
- **MoCD typically presents within a week of birth** and is often characterised by symptoms such as **excessive crying, poor feeding, and seizures**, all of which worsen over time[1,2].

1 MedlinePlus. (2025); 2 Orphanet. (2012)

Background

- There is **no cure for MoCD**; care focuses on **managing symptoms and improving quality of life**. A team of specialists is involved to provide relief for specific symptoms and ensure the highest possible quality of life is achieved [1,2].
- **A new treatment, Nulibry ® (fosdenopterin), is currently undergoing evaluation** by NICE (National Institute for Health and Care Excellence) in England and SMC (Scottish Medicines Consortium) in Scotland [3,4].
- Nulibry ® (fosdenopterin), is already available in some countries and has shown **promising impact in restoring molybdenum cofactor production**, which reduces the levels of toxic substances in the body and thereby the negative impact of the disorder.

Objectives of this study:

1. To gain a comprehensive understanding of the diagnostic journey, daily living experiences, treatment and management practices, and the perspectives of parents and caregivers of children with Molybdenum Cofactor Deficiency (MoCD) type A.
2. To expand the public evidence-base of the lived experiences of MoCD Type A and ensure the community's insights are represented in health technology assessment proceedings.

1 National Organization for Rare Disorders. (2024.); 2 Child Neurology Foundation. (2021); 3 More information on NICE evaluation: https://www.nice.org.uk/guidance/indevelopment/gid-hst10055; 4 More information on SMC evaluation: https://scottishmedicines.org.uk/medicines-advice/fosdenopterin-nulibry-ultra-orphan-smc2624/public-summary/

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METHODS

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Methods

- This report draws on data collected from a survey conducted by Metabolic Support UK (MSUK).
- The digital survey ran from January to April 2024
  - The survey contained questions on the following sections:
    - Diagnosis
    - Daily living with MoCD Type A
    - Treatment and management
    - Parent/carer perspective
    - New treatment
  - These sections were selected based on the NICE patient organisation submission template, alongside in-house knowledge of MoCD type A
  - MSUK utilised a dissemination strategy including social media posts, newsletter announcements, and direct mailing list outreach, to reach people caring for someone with MoCD Type A
  - A £25 Amazon voucher was offered to all participants

Methods (cont.)

- The digital survey was designed using Google Forms
- The data generated through the survey was analysed using a mixed methods approach:
  - Descriptive statistics, including means and standard deviations, were calculated to summarise quantitative data
  - Content analysis was applied to the qualitative data collected

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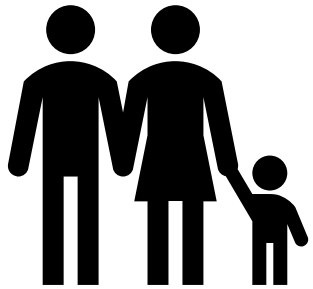
DEMOGRAPHICS

Survey respondents: Demographics



3 parents provided responses:

- 2 bereaved parents/carers
- 1 parent of a child living with MoCD Type A

Age of respondents' children:

- The average age: 3 years and 4 months
- Range: 1-6 years

All respondents reside in the United Kingdom

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RESULTS

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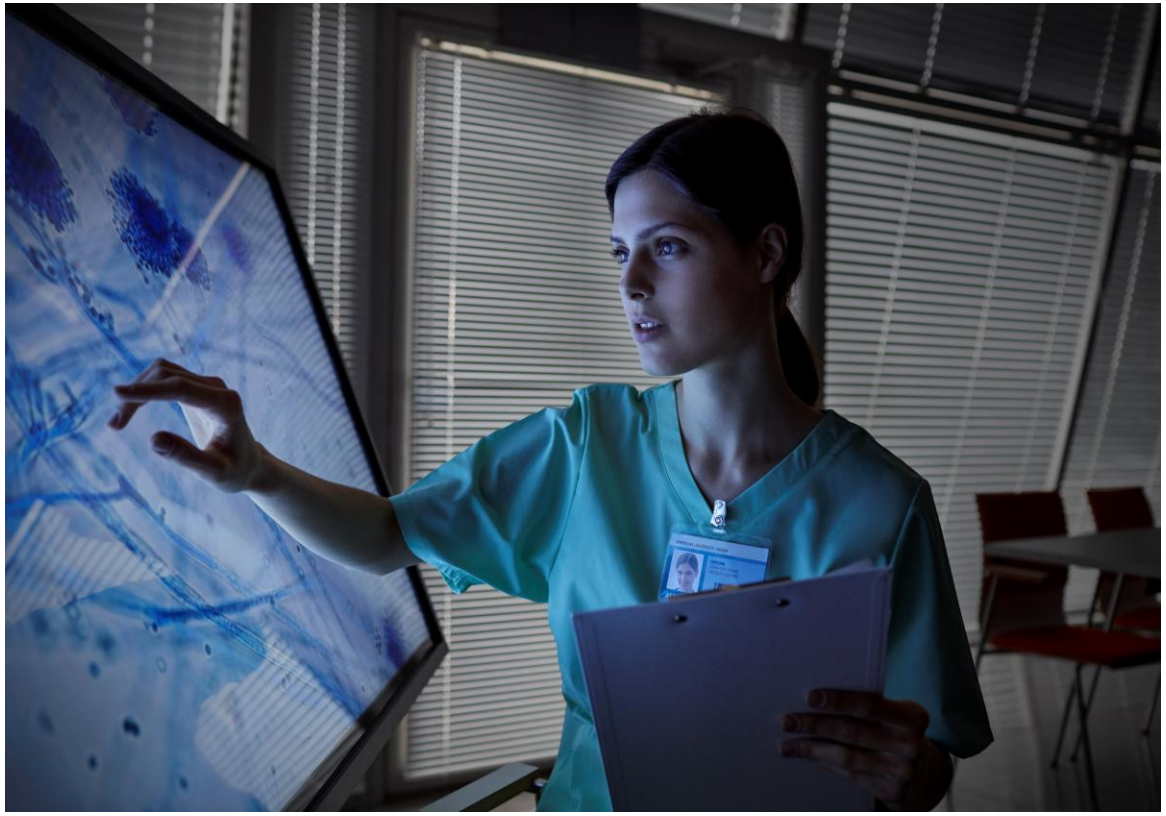
DIAGNOSIS

Diagnosis: Timing

- On average, MoCD Type A was diagnosed **within the first 4 weeks** after birth for all participants.

- Diagnosis was**, on average,** confirmed **3 days** after symptom onset.
  - Range: 2-4 days



Diagnosis: Experience

Most participants reported a smooth and straightforward diagnosis process, though a delay due to logistical challenges during Bank Holidays did occur.

2 participants reported **no challenges**:

- The process was "straightforward."
- The smooth process attributed to hospital admission under Metabolic consultants' care.

1 participant faced **challenges**:

- Their child had to wait 3 days for transport to another hospital to get tested. The delay was caused by a Bank Holiday.

Diagnosis: Most significant symptoms

Seizures	Dystonic movements	Lack of interest in feeding/unable to breastfeed
Jerkyness from the increased startle reflex	Vomiting	Excessive crying and irritability

Diagnosis: Access to support

- **2 participants** felt they **had** adequate access to support and contacts for questions.

- **1 participant** felt they **did not have** adequate access to support.

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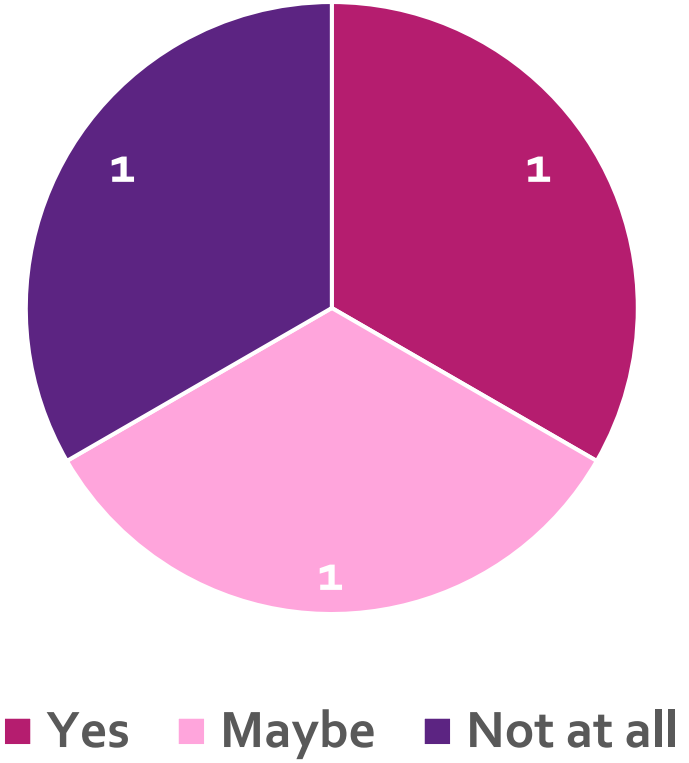
TREATMENT AND MANAGEMENT

Day-to-day management of MoCD type A

Is the condition easy to manage?



Treatment and management: Healthcare professionals involved

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All respondents have engaged with Consultant Neurologist, GP, Hospice Care, Metabolic Consultant, Physiotherapist, Specialist Dietitian

In all 3 cases:
- Consultant Neurologist
- GP
- Hospice Care
- Metabolic Consultant
- Physiotherapist
- Specialist Dietitian

In 2 cases:
- Clinical Nurse Specialist
- Ophthalmologist
- Social Worker

In 1 case:
- Palliative Care Team
- Personal Assistant

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Management: Appointments and care support

Appointments:

- On average, individuals attend 1–2 appointments per month, with the range varying from 0 to 8 appointments.
- Most respondents reported attending approx. 1 appointment per month, though some required more frequent visits.
- The range of appointments reflects diverse care needs, with individuals seeing various specialists such as GPs, physiotherapists, ophthalmologists, and others.

Home and hospice care:

- Overnight care for children with MoCD Type A is typically managed at home, with the use of hospice care during the day for additional support.

*The full table, detailing the average number of appointments for each specialist, can be found in the Appendix.

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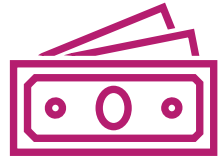
Appointments: Travel impact and cost

Commute times for appointments vary widely, **ranging from short trips of 5-20 minutes to longer journeys of up to 2 hours** for some participants.

All participants **use a car to attend appointments** with occasional use of a train or a taxi.



Monthly travel costs vary, with some participants spending as little as £5, while others report higher averages of around £25.

Management: Additional costs

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The additional costs of caring for a child with MoCD Type A varies widely, ranging from minimal expenses to significant investments in equipment, therapies, home adaptations, and specialised supplies.

Varying additional costs:

- Some participants faced **limited additional expenses** (e.g. only a freezer for medication).
- Others required **significant resources** for a variety of equipment and private therapies, including:
  - **Medical and mobility equipment:** e.g. sensory equipment, mobility aids, push-button toys, specialist creams
  - **Hygiene and sterilisation:** e.g. sterilisation equipment and drying rack for syringes and milk bottles or storage for medical equipment
  - **Comfort and adaptive home furnishings**: e.g. extra bedding and mattress protection, additional pillow and positioning wedge, specialist bath
  - **Private therapies** and specialised **home-based care tools**.

Home adaptations:

- Many participants needed **home modifications** to facilitate at-home care:
  - **Specialised seating equipment**: bath chairs, moulded POD chairs, special car seats, wheelchairs.
  - **Other:** nebuliser, sleep system.
- An extensive home adaptation **was already present** in one family's home, which the family deemed would have been required if not already in place.

Management: Challenges

The main management challenges shared by families related to accessing medication and dietary supplementation.

Medication accessibility issues:

Stock shortages: Medication and dietary supplements were sometimes unavailable due to stock issues.

Order errors: Medications were occasionally cancelled without notice or mislabelled due to human error.

Delays: Participants experienced delays in receiving critical medications.

Preventive measures:

One participant now orders medication **a week in advance** to avoid running out, highlighting the need for proactive management.

Management: demanding necessities

Caregivers emphasise the intensive and time-consuming demands of managing MoCD Type A, acknowledging the necessity of treatments while underscoring their limited scope.

- Respondents express that **few treatments** are available, and the **unpredictability** of the condition intensifies the complexity of care.
- Treatment requires **continuous attention**: home oxygen, NG feedings, and hourly monitoring. Families describe the process as **intensive and time-consuming**.
- Available treatment is described as **useful and essential** for the improvement of the children's life, but **limited in scope.**
- The **medication-centred approach** can feel restrictive, leaving parents feeling **disempowered**.
- **Beyond medications,** therapies like outdoor time, private physiotherapy, massage, and art/play therapy are highlighted as **transformative** for comfort, happiness, and quality of life.

Management: Home and hospice care

Overnight care for children with MoCD Type A is typically managed at home, with the use of hospice care during the day for additional support.

Overnight care at home:

- All participants indicate they provide regular overnight care as part of managing the condition.

Use of hospice care:

- Day hospice care has been used by two participants.

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IN YOUR OWN WORDS

All respondents answered this section

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Daily routine: Similarities and differences

Care routines differ in structure and intensity—ranging from scheduled care around activities to continuous hands-on management—but share common challenges, including demanding daily responsibilities like medication administration, addressing medical needs, and ensuring the child's comfort, well-being, and engagement.

Similarities:

- **A highly structured and demanding daily routine centred around managing the condition**
  - The management of the condition is physically and emotionally challenging.
- **Key responsibilities:**
  - Preparation and administration of medications.
  - Addressing specific medical needs.
  - Dedication to providing comfort and entertainment for the child.

Differences:

- Care routines vary in structure and intensity, with some children requiring scheduled care around external activities, while others need continuous, hands-on management and integration of therapeutic, educational, and social activities for overall well-being.

"Busy! Up early to prepare meds and make sure food is cooked…My most treasured part of the day was an hour in the morning bathing [child's name], massaging him, doing his physio and changing his medical tapes…Then we would go for a walk…or we would visit family/friends or the complex children's centre. Often, we would have family and friends visit us at home."

Anonymous respondent

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Gaps in available management and support

Addressing unmet needs for MoCD Type A includes enhanced parental support, specialised medical interventions, early sensory testing, and access to therapeutic resources

- Support for parents, including within community
- Medication to help with dystonic movements
- Eyes and ear testing for all babies at early stages
- An opportunity to benefit from complex children's centre and hydrotherapy pool for everyone
- Higher availability of activities for children
- More respite options

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Future treatment

Aspirations for future treatment:

- It should help with pain management at the early stages
- Medication should be easily transportable
- Increased focus more on physical therapy
- Reduce stress for children and families

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PARENT/CARER PERSPECTIVE:

[Two respondents answered this section]

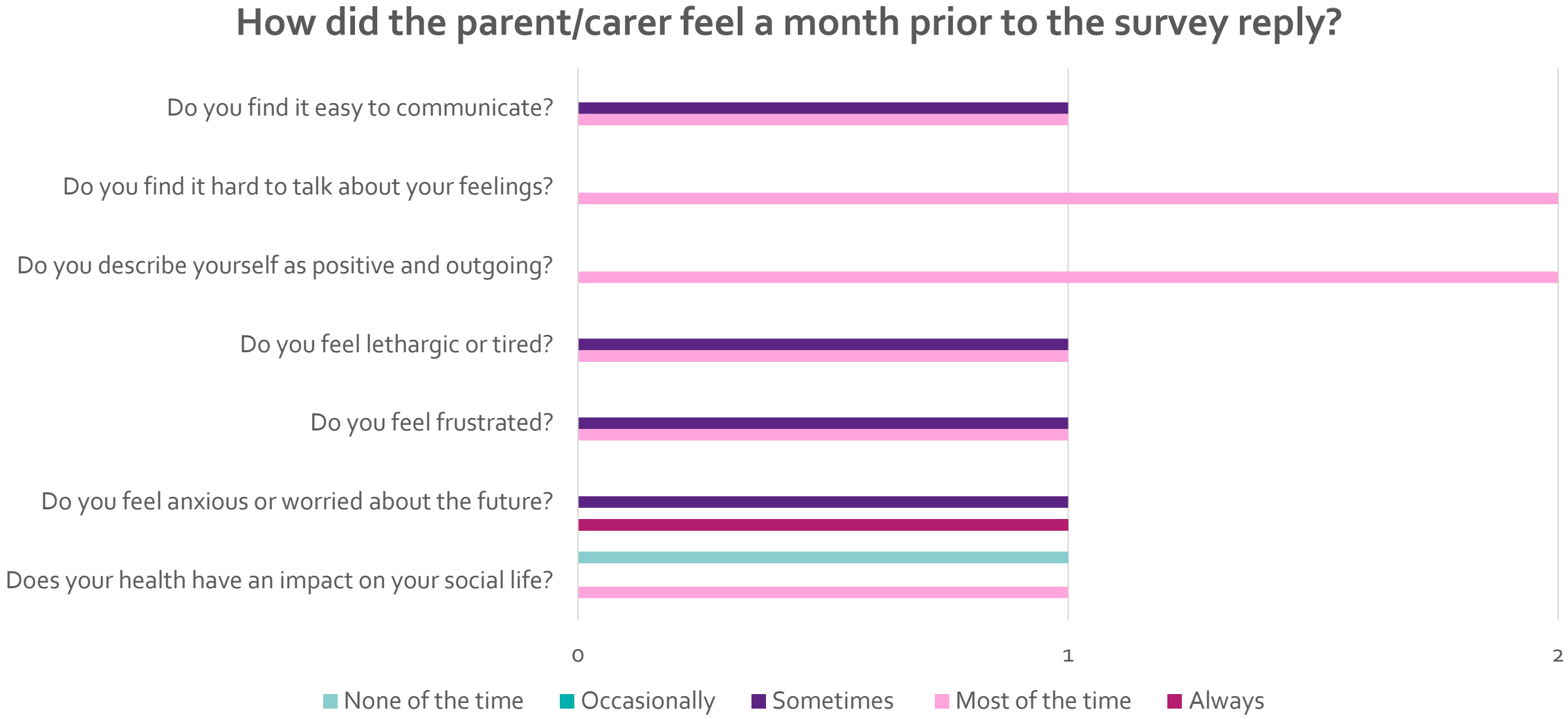
Carer perspective: General health and well-being

Caregivers reported good health prior to their child's birth, with the loss of their child leading to stress-related health impacts for one respondent.

Both respondents reported being in **good health and well-being** prior to their child's birth with no chronic conditions.

Following their **child's loss** to MoCD Type A, one respondent experienced **psoriasis**, which they attribute to **stress, poor diet, and self-care neglect** in the aftermath.

How did the parent/carer feel a month prior to the survey reply?



Carer perspective: impact on work and social life

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Caring for a child with MoCD Type A can affect work and social life, as parents may leave jobs and face limited social activities; support from family, friends, and community connections helps make a positive difference.

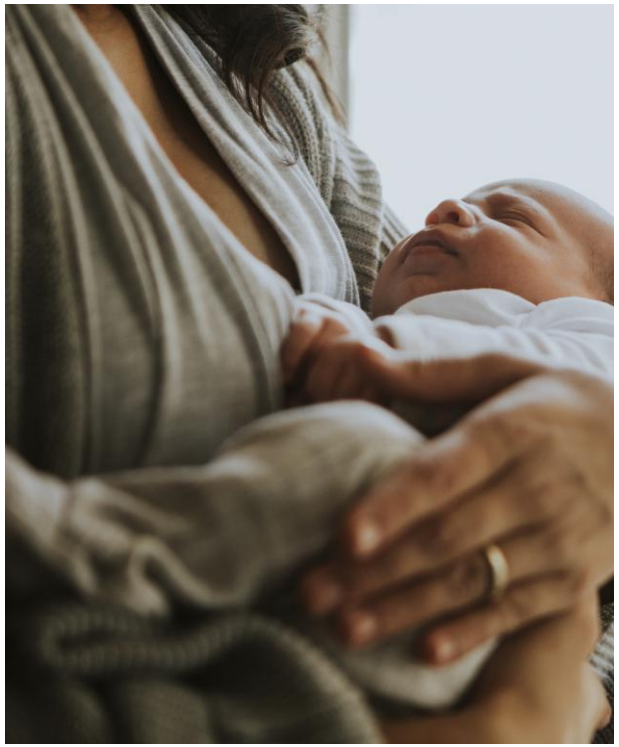
Negative impact of MoCD Type A on ability to work:

- *"I gave up work … due to not being able to fulfil my shifts due to needing to be available for my son at all times"* - Anonymous respondent

Mixed impact of caring for a child with MoCD Type A on a social life:

- *"It has limited it [social life] in some ways and enriched it in many others. I took [child's name] out a lot and I have a beautiful supportive friend and family network who included him in everything from park outings, zoo trips and holidays to art projects, religious gatherings and celebrations. I felt more obliged to go out and help [child's name] get out and experience different opportunities and have time around other children and outdoors. I also met some amazing people through our complex children's centre, made close friends with his carers and found my own family and friend relationships strengthened through us working together to support [child's name] and grieve collectively."* - Anonymous respondent

- *"I have no social life, I haven't been out with my friends since before [child's name] was born. We do not trust anyone to look after [child's name] in the way we do."* - Anonymous respondent



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DISCUSSION AND CONCLUSION

Discussion and conclusion

The current research lays the basis for creating a broader evidence base and understanding of the lived experience of MoCD type A:

- **Diagnostic journey:** diagnosis of MoCD Type A typically occurs within the first 4 weeks of life, with confirmation, on average, 3 days after the onset of symptoms.
- **Daily life:** management of MoCD Type A is intensive and highly demanding, requiring coordinated care from multiple healthcare professionals and significant parental involvement.
- **Treatment and challenges:** caregivers face difficulties with medication accessibility, additional costs, and limited scope of available interventions. The new treatment, Nulibry®, raises hope and shows promise.
- **Caregiver perspectives:** parents experience emotional and physical stress, yet highlight the importance of supportive networks and therapies beyond medication.
- **Future needs:** addressing gaps in care (e.g., earlier testing, better support for parents, broader access to therapies) and more comprehensive and accessible treatments to reduce the burden on families.
- **Future research:** should expand on the current research to create a broader evidence base of the lived experience of MoCD type A.

Acknowledgment

- Metabolic Support UK would like to **thank the members of the MoCD type A community** for their consideration of and participation in the survey and providing valuable input.

- Funding was received from Sciensus BV to support this project

- All the pictures and illustrations in the report were sourced from the Microsoft PowerPoint stock images and illustrations library. This approach was chosen due to the ultra-rarity of MoCD Type A, ensuring the anonymity of individuals living with the condition. Additionally, it avoids creating stereotypes about the disorder's visual characteristics, as the selected images may not be representative of everyone living with MoCD Type A.

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REFERENCES AND RESOURCES

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References and additional resources

- **References:**
  - Child Neurology Foundation. *Molybdenum Cofactor Deficiency Type A*. (2021) Retrieved from https://www.childneurologyfoundation.org/disorder/molybdenum-cofactor-deficiency-type-a/
  - MedlinePlus. *Molybdenum cofactor deficiency.* (2025)Retrieved from https://medlineplus.gov/genetics/condition/molybdenum-cofactor-deficiency/#causes
  - National Organization for Rare Disorders. *Molybdenum Cofactor Deficiency.* (2024) Retrieved from https://rarediseases.org/rare-diseases/molybdenum-cofactor-deficiency-mocd/
  - Orphanet. *Molybdenum cofactor deficiency.* (2012)Retrieved from https://www.orpha.net/en/disease/detail/833

- **Additional resources:**
  - Parent's blog about experiences of MoCD in the hope other families can learn from their insights. Please visit: https://molybdenumandmore.blogspot.com/
  - More information on NICE evaluation: https://www.nice.org.uk/guidance/indevelopment/gid-hst10055
  - More information on SMC evaluation: https://scottishmedicines.org.uk/medicines-advice/fosdenopterin-nulibry-ultra-orphan-smc2624/public-summary/

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APPENDIX

On average, how many appointments were/are required each month?

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	None	One	Two	Three	Four	Five	Eight
Clinical nurse specialist	2		1				
Consultant neurologist	1	2					
General practitioner	1	2					
Hospice care	1	1		1			
Metabolic consultant	3						
Ophthalmologist	1	2					
Palliative care team	2	1					
Personal assistant	2						1
Physiotherapist	1	1	1				
Social worker	1	2					
Specialist dietician		2		1			