ОБЩЕСТВЕНИ КОМУНИКАЦИИ И ИНФОРМАЦИОННИ НАУКИ
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COMMUNICATION PROCESSES FOR THE EVALUATION OF THERAPEUTIC STRATEGIES FOR MCT8 DEFICIENCY: A PATIENT-CENTERED PERSPECTIVE

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Abstract: *The aim of this article is to identify patient-relevant therapeutic targets in the context of the serious rare disease MCT8 deficiency and to evaluate the effectiveness of existing treatment approaches based on clinical and patient-reported parameters. Using a systematic literature search and an exploratory survey of 21 international patient representatives, key treatment goals and satisfaction with the current care situation were recorded. The study quality was analyzed using the GRADE criteria. Quality of life, gross motor function, gastrointestinal health, cognitive development and immune stability were named as priority treatment goals. The desire for improved sleep was mentioned most frequently. Satisfaction with the current therapies was moderate in all areas. The results emphasize the need to systematically integrate patient-relevant objectives – especially quality of life – into clinical research on MCT8 deficiency and care. The development and application of standardized PRO instruments is recommended for future studies in order to improve the care situation of those affected.*
Keywords: *MCT8 deficiency; rare disease; patient-relevant outcomes; communication processes*

INTRODUCTION

Communication processes are essential for interdisciplinary collaboration in modern healthcare. Information systems (IS) support the collection, management and analysis of complex clinical data, enabling knowledge creation and decision-making. They consist of technological, human and organizational components and are particularly important in rare diseases, where case numbers are small, data are scarce and multiple actors are involved (Gelinas & Dull 2008).

MCT8 deficiency (Allan-Herndon-Dudley syndrome, ORPHA:59; OMIM:300523) is extremely rare, with around 320 documented cases worldwide (Schwartz 2017). It is caused by mutations in the SLC16A2 gene, encoding the MCT8 transporter, which regulates T3/T4 transport into cells and across the blood-brain barrier (Allan et al. 1944; Dumitrescu et al. 2004; Friesema et al. 2004; Schwartz et al. 2005). The condition combines severe neurocognitive developmental delay with peripheral thyrotoxicosis (Groeneweg et al. 2020).

As affected individuals cannot self-report, relatives are key informants, linking clinic, research and daily life. Patient-reported outcomes (PROs) allow care to be tailored to actual needs rather than biochemical markers alone.

No causal therapy exists (Neubauer & Schiebl 2019). The only approved drug is TRIAC/Emcitate (EMA 2025) for thyrotoxicosis. Supportive measures address motor limitations, spasticity, feeding difficulties, communication deficits or sleep disorders (e.g. physiotherapy, mobility aids, tube feeding, speech therapy, dietary therapy, symptomatic medication). While these interventions may help daily coping, their effectiveness is rarely evaluated systematically.

This study systematically analyses evidence-based therapeutic approaches for their clinical efficacy and relevance to PROs, alongside an exploratory survey of patient representatives to identify treatment goals and satisfaction. The aim is to improve patient-centered, interdisciplinary treatment concepts, supported by digital IS and participatory communication.

RESEARCH METHODOLOGY

Literature review

A systematic search (MEDLINE/PubMed, Google Scholar, ClinicalTrials.gov) using terms

[(SLC16A2) OR (MCT8) OR (AHDS)] AND [(therapy) OR (intervention) OR (treatment)] identified 2,590 studies. Inclusion: peer-reviewed human studies on MCT8 deficiency treatments (case reports/series, clinical trials, cohort studies); exclusion: preclinical studies, obsolete harmful therapies.

The results were critically evaluated with regard to methodological quality. After screening 11 relevant studies remained. The evidence quality of the treatment studies was classified according to the GRADE criteria (Schünemann & Reinap 2023) into the following categories: high (the true effect is close to its estimate), moderate (the true effect is probably close to its estimate), low (the true effect may differ substantially from the estimate), and very low (very little confidence in the estimate).

Explorative survey

In cooperation with MCT8-Forschung e.V., a questionnaire was developed and completed by 21 international patient representatives. Recruitment was voluntary; data were pseudonymized. The questionnaire, in German/English/Polish, assessed therapy goals and satisfaction. Responses were analysed descriptively and integrated with literature findings.

RESULTS

The 11 included studies comprised:

- TRIAC: phase II trial (Groeneweg et al. 2019, n=46), retrospective cohort (Van Geest et al. 2021, n=67), case series (Gazek et al. 2023, n=4), case report (Ünsal & Hayran 2023, n=1).
- DITPA: case series (Verge et al. 2012, n=4).
- Levothyroxine (LT4) + propylthiouracil (PTU): case reports (Visser et al. 2012, n=1; Wémeau et al. 2008, n=1).
- Liothyronine (LT3): case report (Zung et al. 2011, n=1).
- Prenatal LT4: case report (Refetoff et al. 2020, n=1).
- Levodopa/carbidopa ± botulinum toxin A: case series (Remerand et al. 2019, n=3; Tonduti et al. 2012, n=2).

Evidence quality ranged from low to moderate due to small sample sizes and methodological heterogeneity.

Effectiveness of different treatment strategies

Biochemical effects: TRIAC, LT4+PTU, DITPA, prenatal LT4 all reduced FT3 (and usually FT4) with concomitant TSH decreases. TRIAC and prenatal LT4 often reached reference ranges or clinically relevant reductions. Side effects were mild to moderate (GI symptoms, CK elevations, occasional infections), mostly dose-dependent. Levodopa/carbidopa and botulinum toxin had no effect on thyroid parameters.

Weight stabilization: Improvements most marked under TRIAC; prenatal LT4 also led to modest gains.

Neurological development: Isolated reports (TRIAC, prenatal LT4) noted motor milestone gains (head control, sitting) and improved myelination; quantitative improvements were generally small or absent.

Extrapyramidal symptoms: Partial, symptomatic relief with levodopa/carbidopa or botulinum toxin; effects not sustained.

Overall: good biochemical efficacy in several therapies (especially TRIAC), but limited and inconsistent neurological/motor benefits. PROs were rarely assessed.

Supportive interventions

Physiotherapy, occupational therapy, speech/feeding therapy, assisted communication, mobility aids, nutritional support (incl. tube feeding), and symptom-targeted drugs (e.g. for dystonia, seizures, spasticity, sleep) are widely used (Sarret et al. 2020; Refetoff & Dumitrescu 2007; Van Geest et al. 2021). Evidence is largely anecdotal; systematic PRO-based evaluation is lacking.

Patient survey

Cohort**:** 21 male patients with SLC16A2 mutation; median age 6.1 years (22 months–12 years). 75% had severe, 25% mild variants. All showed severe developmental delay; few achieved basic motor skills (e.g. 17% head control, 8% walking). None could speak.

Therapy goals:

- Quality of life: better sleep (4.06), more enjoyment (3.94), fewer hospital stays (3.43), reduced environmental sensitivity (3.44)
- Gross motor function: better body control (3.90), progress in sitting/walking (3.85)
- Gastrointestinal health: less constipation/diarrhoea (3.53), less painful burping (3.21)
- Cognition: mental development (3.57)
- Immune stability: fewer infections (3.28)

Therapy goals are presented on Fig. 1 below.

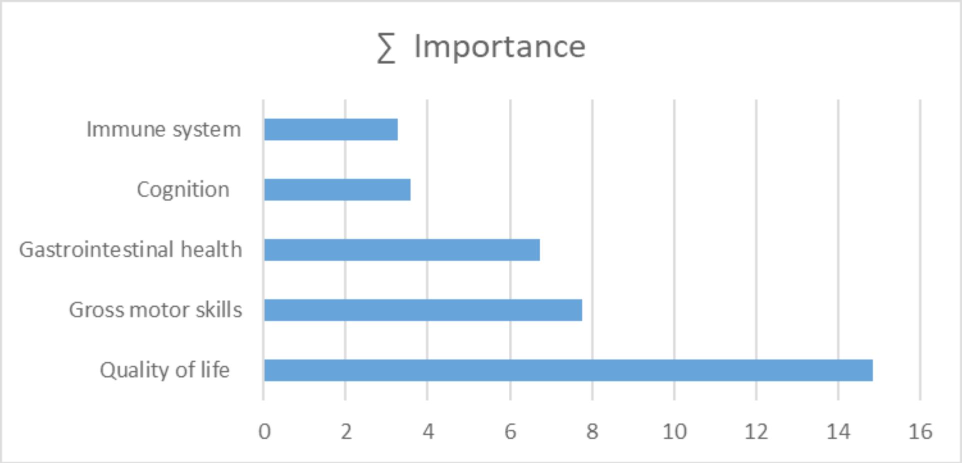

Fig. 1. Patient-relevant therapeutic goals ranked by aggregated importance (scale 1–5)

Satisfaction:

Scores were moderate (scale 1–5), mostly 2.1–2.6. Largest gaps between importance and satisfaction were in motor development and GI health; sleep improvement also lagged behind expectations. Figure 2 below compares therapy areas and therapy satisfaction.

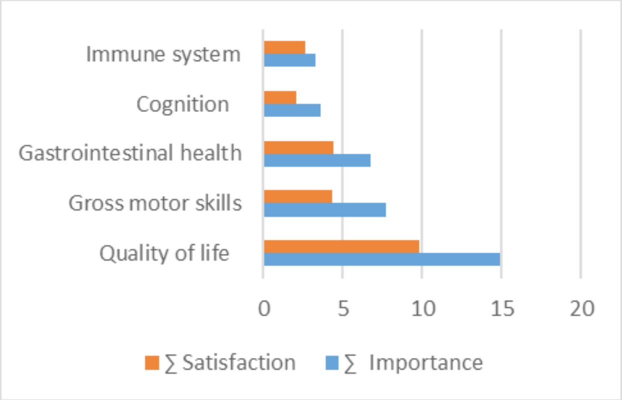

Fig. 2. Patient-relevant therapy areas and therapy satisfaction

DISCUSSION

The results underline the need for symptom-oriented, interdisciplinary care for MCT8 deficiency that is consistently oriented towards patient-relevant objectives (PROs). The parental prioritization of quality of life, motor development, gastrointestinal health and cognitive development makes it clear that existing treatment strategies have so far only inadequately addressed these needs. While individual treatment approaches such as TRIAC for peripheral thyrotoxicosis or physiotherapeutic measures can show selective effects, there is an overall lack of standardized, systematically evaluated treatment options.

The results of the systematic literature search make it clear that patient-relevant endpoints such as pain reduction, quality of life or development have hardly been considered in the studies published to date. The focus was predominantly on biochemical parameters. There was no systematic PRO-based evaluation of the suitability for everyday use and effect of the therapies. This gap makes patient-centered therapy development more difficult, as affected children are unable to provide information on subjectively experienced therapy goals due to their pronounced communication and developmental disorders. The parents‘ perspectives as primary caregivers therefore provide an indispensable supplement to the pure evaluation of clinical parameters.

Although supportive measures such as mobility and communication aids, specific nutritional therapies or symptomatic medication are used in practice, they often remain unstructured and without evidence-based validation. A more PRO-oriented evaluation of such interventions is necessary. Digital information systems can help to record and evaluate relevant findings from practice and research across all sectors and make them accessible to all stakeholders. In the context of rare diseases in particular, ethically sound research that guarantees transparency, participation and the involvement of those affected is also required. Taking their perspectives into account is not only methodologically sensible, but also ethically necessary in order to further develop the care situation in a fair and needs-based manner.

CONCLUSION

The results of this study underline the high relevance of a symptom-oriented, interdisciplinary therapeutic approach to MCT8 deficiency, which is consistently oriented towards the goals prioritized by patient representatives (PROs). The data collected show that quality of life, motor development, alleviation of gastrointestinal complaints and cognitive support are at the center of parental therapy expectations. Although some therapeutic measures – such as TRIAC for peripheral thyrotoxicosis or physiotherapy to promote mobility – show positive effects in individual cases, it is clear that the existing therapy options do not yet adequately cover the complex and multiple needs of people with MCT8 deficiency.

The role of communication processes and information systems is playing an increasingly central role here: structured digital information systems are crucial, particularly in the case of a rare disease such as MCT8 deficiency, with sufferers spread around the world, multiple specialist disciplines involved and limited clinical data. They enable patient knowledge to be systematically recorded, treatment data to be aggregated and evaluated on a multicenter basis and research findings to be efficiently passed on. The use of interoperable IS solutions can significantly improve the standardized collection of PROs, their analysis and the targeted exchange between science, practice and self-help organizations. Consistent integration of PROs into such systems also offers the opportunity to bring clinical research and care closer to the real needs of those affected. In order to meet the complex challenges of rare diseases such as MCT8 deficiency, future therapy and care concepts must not only be evidence-based, but also ethically reflected. The active involvement of those affected and their relatives is key to making research participatory, transparent and fair.

Particularly in the context of rare diseases, it is essential in the digital age to design communication processes as bidirectional knowledge flows. The systematic involvement of patient representatives – for example through targeted surveys – makes a decisive contribution to aligning evidence-based research and clinical care with real needs and thus improving them in the long term.

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КОМУНИКАЦИОННИ ПРОЦЕСИ ЗА ОЦЕНКА НА ТЕРАПЕВТИЧНИ СТРАТЕГИИ ПРИ ДЕФИЦИТ НА MCT8: ОРИЕНТИРАНА КЪМ ПАЦИЕНТА ПЕРСПЕКТИВА

Резюме: *Целта на тази статия е да се идентифицират терапевтичните цели, които са от значение за пациентите, в контекста на тежкото рядко заболяване дефицит на MCT8, и да се оцени ефективността на съществуващите подходи за лечение въз основа на клинични и съобщени от пациентите параметри. Чрез систематично проучване на литературата и експлоративно проучване сред 21 международни представители на пациенти бяха определени основните терапевтични цели и удовлетвореността от настоящото състояние на грижите. Качеството на проучването беше анализирано въз основа на критериите GRADE. Качеството на живот, грубата моторика, стомашно-чревното здраве, когнитивното развитие и имунната стабилност бяха определени като приоритетни терапевтични цели. Най-често споменаваното желание беше за по-добър сън. Удовлетвореността от настоящите терапии беше умерена във всички области. Резултатите подчертават необходимостта от систематично интегриране на целите, които са важни за пациентите, особено качеството на живот, в клиничните изследвания на дефицит на MCT8 и грижите за пациентите. За бъдещи проучвания се препоръчват разработването и прилагането на стандартизирани инструменти за измерване на резултатите, докладвани от пациентите (PRO), с цел подобряване на грижите за засегнатите лица.*

Ключови думи: *дефицит на MCT8; рядко заболяване; значими за пациента резултати; комуникационни процеси*

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