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Myotubular and Centronuclear Myopathy - Analysis of registry data to understand nutritional practices and difficulties and explore potential associations between liver health and nutrition

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Registry Background

The MTM-CNM Global Patient Registry is a key research database and the largest source of patient-reported data for individuals (n=614, living n =532, deceased n = 82, male n=384, females n=230) affected by Myotubular and Centronuclear Myopathies (MTM-CNM). The registry collects data from patients with variants in the following genes ***MTM1,DNM2, BIN1, RYR1 and TTN.***
Overall 56% participants had a genetic confirmation with 244 diagnosed with the MTM1 variant and 96 participants with other forms of CNM.

Comprehensive data collection
includes demographic, genetic, respiratory, liver function, mobility outcomes

Responsive to Research
adaptive to emerging clinical priorities and research questions (e.g liver function).

Global accessibility
*registrations from **57** countries, questionnaires available in **10** languages*

Collaborative Network
work in partnership with patients/families, clinicians, researchers and industry

Nutritional Health Overview

In 2024, data collection was expanded to include a nutrition section to capture dietary practices and nutritional health among individuals with MTM-CNM . The questionnaire was developed in collaboration with two patient advocacy organisations, Myotubular Trust UK and MTM-CNM Family Connection, USA , and the MTM-CNM Liver Collaborative, along with input from nutritionists and clinical experts.
From August 2026, **183** participants had completed the questionnaire to inform on such items as nutrition route, diet types and meal frequency. Analyses were conducted to explore associations between nutrition health and other factors such as liver function, mobility outcomes and ventilation.

These findings provide an overview of current dietary and nutritional health practices within the MTM-CNM community and may help inform clinical care and provide future nutritional guidance for patients, caregivers healthcare professionals.

Key Observations (Figures 1-5)

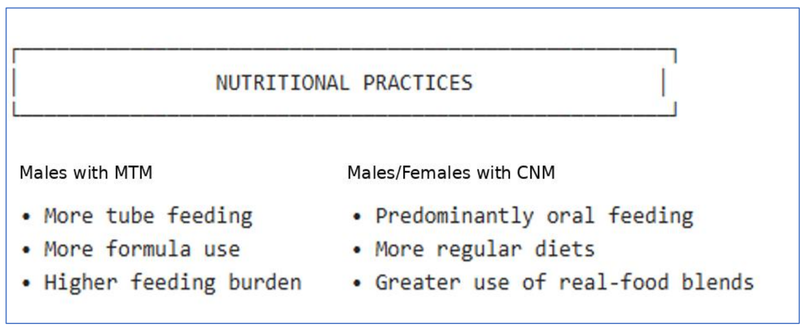
- Oral feeding was more common among CNM participants, whereas tube feeding (NG, G-tube, or J tube), either alone or combined with oral feeding, was more prevalent in ..
- Formula-based nutrition was reported more frequently in males, particularly among those with MTM (60%) compared with CNM (40%) and female participants.
- Standard enteral formula was the most commonly used nutritional formula, however real-food blends were reported more frequently in CNM participants than those with MTM.
- 'Other' feeds includes high-energy formulas (Nutrini, Infatrini and,Fortini) and misclassified feeds that should have been categorised as allocated as 'Standard' feed
- Feeding frequency was higher among younger participants, with the greatest feeding requirement in children with MTM
- Meal duration was typically less than one hour across both MTM and CNM cohorts.
- Most participants reported following a regular diet, high-protein and gluten-free diets were more commonly reported among females with MTM.
- Dietary guidance was most frequently provided by nutritionists (41%), followed by self-directed research (36%), primary care (29%), gastroenterologist(18%) and the patient community (7%)

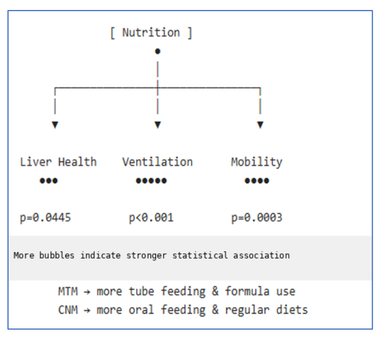
Associations : nutrition and disease severity (figures 6 – 9)

Among genetically confirmed MTM1 participants, significant associations were identified between :

- Nutrition route and liver disease (p = 0.0445)
- Liver disease and ventilation status (overall p = 0.0368; males p = 0.0427)
- Liver disease and wheelchair use in males (p = 0.0338)
- Nutrition route and ventilation use (overall p < 0.001)
- Nutrition route and wheelchair use (overall p = 0.0003)
- Diet type and wheelchair use (overall p = 0.0085)

These findings suggest that more complex nutritional requirements are present in patients with greater disease physical manifestation and functional impairment.





Results

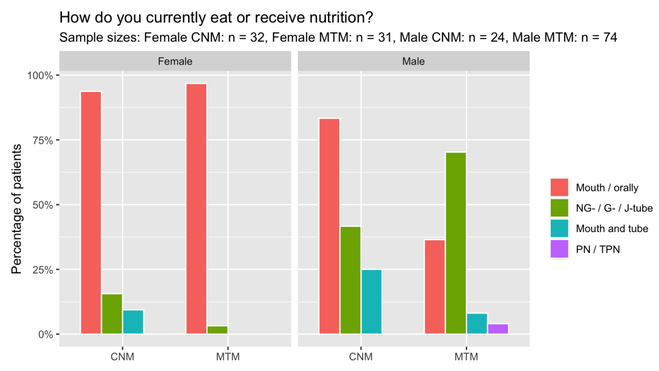


Figure 1 : Nutrition Route

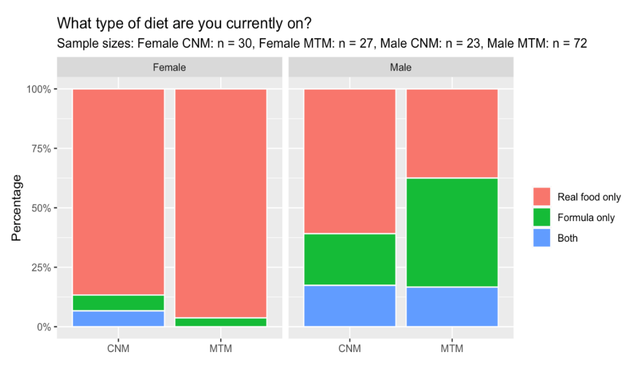


Figure 2 : Diet Type

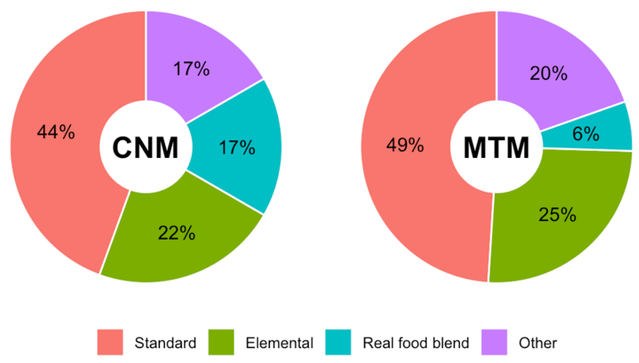


Figure 3 : Formula Type (CNM=18, MTM=51)

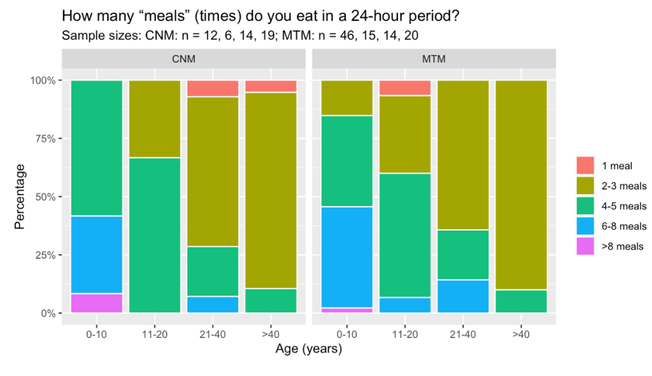


Figure 4 : Meal Frequency by Age

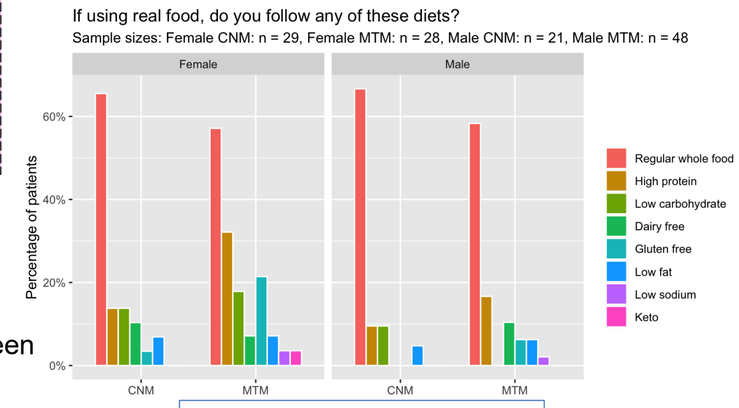


Figure 5 : Diet Type

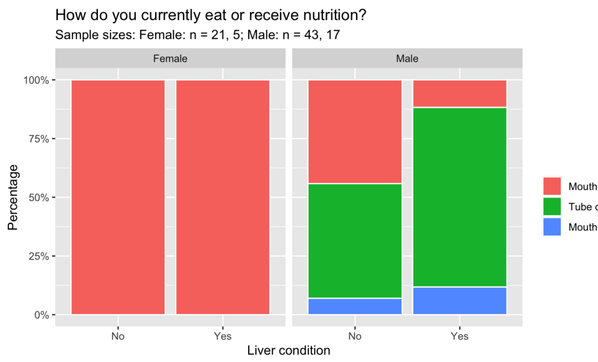
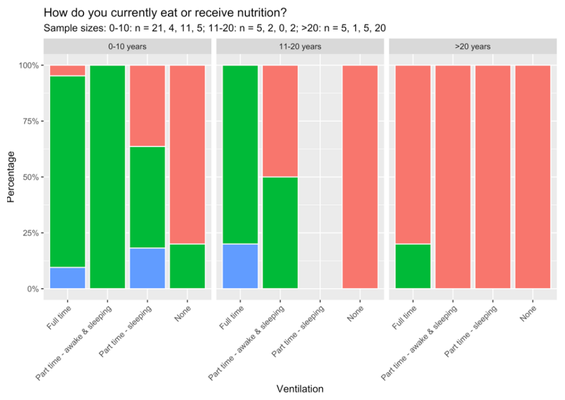


Figure 6 : Associations Nutrition Route V Liver Condition



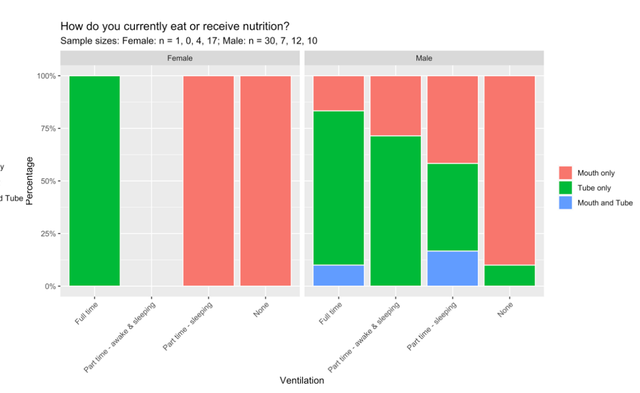


Figure 7 & 8 : Associations Nutrition and Ventilation by age and gender

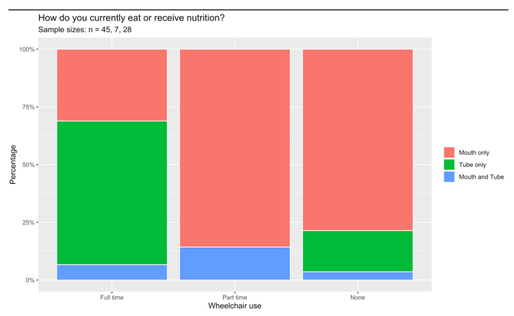


Figure 9 : Associations Nutrition and wheelchair usage

Conclusion

In this international registry cohort, individuals with MTM1-related disease demonstrated greater reliance on tube feeding and formula-based nutrition than other CNM subtypes. Significant associations were observed between nutritional factors, liver disease, ventilation requirements and wheelchair use, suggesting that nutritional complexity may reflect overall disease severity. These findings support routine multidisciplinary nutritional assessment and longitudinal evaluation of nutritional interventions in MTM-CNM. These data support the integration of multidisciplinary nutritional assessment into standard clinical care and provide a foundation for developing evidence-based nutritional guidance for patients and caregivers. Longitudinal registry analyses are needed to determine whether nutritional interventions influence disease progression, liver health and functional outcomes over time.

Acknowledgements

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