# Healthcare resource drivers and cost among MCT8 deficiency patients in the United States (US)

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## Background

- **MCT8 deficiency** (Allan–Herndon–Dudley syndrome) is a rare, severely debilitating, and life-shortening X-linked disorder caused by mutations in ***SLC16A2***, the gene encoding **MCT8**.
- MCT8 is a key cell-surface transporter that enables the thyroid hormone (TH) T3 to enter cells and is widely expressed across tissues. It is the main TH transporter in organs such as the liver, thyroid, kidneys, heart, and lungs, and the predominant TH transporter in the blood–brain barrier endothelial cells and various brain cell types.[1-4]
- In MCT8 deficiency, survival is limited. A large natural history study reported a median overall survival of 35 years; 1 in 3 affected individuals died in childhood.[1,5]
- MCT8 deficiency is characterized by concomitant clinical presentations: neurodevelopmental impairment due to insufficient levels of TH (especially T3) in the brain, and systemic metabolic and cardiovascular disturbances due to persistently elevated circulating T3 (thyrotoxicosis) in the periphery.
- There is currently no approved treatment for MCT8 deficiency in the US.
- While MCT8 deficiency requires high levels of care, there is limited research characterizing healthcare burden and costs associated with the disorder.
- The lack of a dedicated ICD-10 code for MCT8 deficiency contributes to the challenge of conducting analysis utilizing real-world data. Further, confirmatory genetic testing results are not routinely available in claims or EHR data.

## Objectives

**In patients with MCT8 deficiency, using US-based real-world data sources:**

- **Evaluate the healthcare resource utilization and cost of care.**
- **Assess the clinical drivers of healthcare resource utilization and costs.**

## Methods

- **Phase 1:** The NorstellaLinQ US Real-World Data (RWD) Database, a patient-level linked dataset comprising claims, lab, and EHR data for approximately 45 million U.S. patients between January 2017 and April 2024, was interrogated using a Natural Language Processing (NLP) algorithm to identify patients with terminology suggestive of MCT8 deficiency. After additional analysis and manual clinical review of 155 putative patients, a diagnosis of MCT8 deficiency was confirmed in 15 patients.
- **Phase 2:** Diagnosed patients with confirmed SLC16A2 gene variants (GeneDx, n=32) among whom 30 patients had ≥2 entries in the NorstellaLinQ database: One patient was included among the 15 patients already identified in the Phase 1 cohort. Thus, 29 patients had both genetic and clinical findings consistent with MCT8 deficiency. In total, 44 unique patients with a confirmed MCT8 deficiency diagnosis were analyzed.
- The total study period was defined by the duration of available insurance activity episodes in the dataset.
- The per-patient-per-year (PPPY) costs and healthcare resource utilization (HCRU) were derived using both electronic health records and claims data.
- The average PPPY for costs and HCRU were determined by dividing the total costs and number of visits, respectively, across all patients by the cumulative number of active patient-years.
- Clinical presentation categories are not mutually exclusive. For instance, if both infection- and weight-related codes appeared on the same day, the costs associated with that visit were included in both categories.

## Results

**Figure 1A: Per-patient-per-year (PPPY) cost by setting**

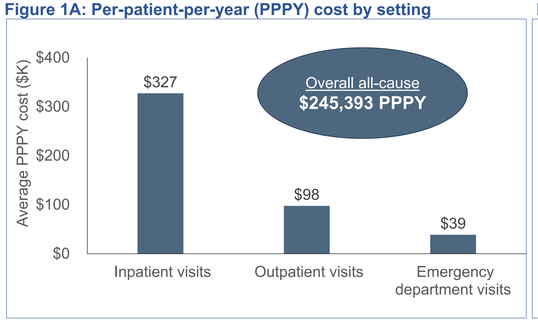
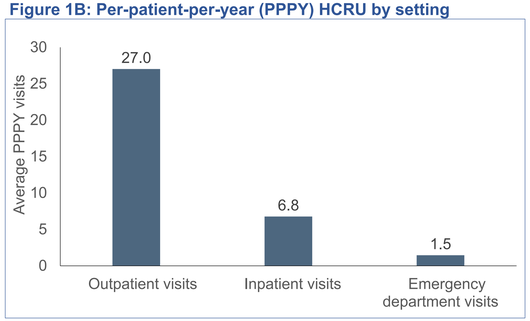
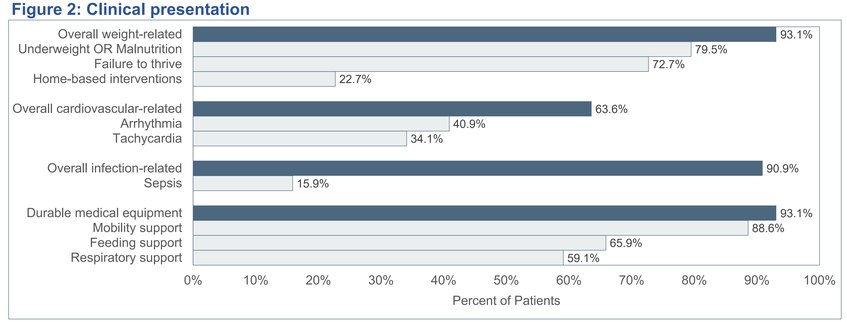



**Figure 1B: Per-patient-per-year (PPPY) HCRU by setting**




- The mean (SD) age of the patients was 5.9 (6.9) years, with a mean follow-up of 72.4 (20.8) months.
- Patients incurred substantial all-cause healthcare-related costs, with inpatient costs contributing disproportionately to the overall economic burden.
- Inpatient care was the largest contributor to healthcare costs, despite outpatient visits representing the highest-utilization setting on a PPPY basis (>2 visits per month).
- Emergency department utilization made a comparatively lower contribution to both PPPY cost and HCRU.
- Data not shown: Average PPPY pharmacy costs were $140,839 for inpatient care and $18,744 for outpatient care, respectively.

**Figure 2: Clinical presentation**




- Nearly all patients had weight-related claims, primarily characterized by underweight/malnutrition and failure to thrive.
- Cardiovascular claims affected approximately 64% of patients, with over 33% of patients experiencing tachycardia.
- Infection-related events were common, with 16% of patients experiencing sepsis.
- Durable medical equipment use was largely driven by mobility devices, with substantial use of feeding and respiratory support.

**Figure 3A – Per-patient-per-year (PPPY) cost – by clinical presentation**




**Figure 3B – Per-patient-per-year (PPPY) HCRU – by clinical presentation**




- Across clinical presentations, inpatient encounters contributed most substantially to the total costs despite relatively low utilization rates.
- The highest inpatient cost burden was observed for cardiovascular-related and sepsis-related visits, potentially reflecting greater acute care intensity and healthcare resource utilization, whereas weight-related presentations were associated with the greatest outpatient utilization, consistent with the need for chronic ambulatory management.
- Infection-related presentations showed moderate healthcare resource utilization but meaningful inpatient costs, indicating a substantial burden across both outpatient and acute care settings.

## Summary and Conclusions

**Cost Drivers and Care Burden:**

- The total cost of care for this cohort of patients with MCT8 deficiency was substantial, with costs reaching approximately $245,000 per patient per year (PPPY).
- Most patients were underweight or had failure to thrive, and more than 30% experienced tachycardia.
- Cardiovascular-related and infection-related encounters were key cost drivers; cardiovascular events contributed substantially to costs despite representing fewer visits, suggesting that each cardiovascular-related episode is associated with high costs.
- Inpatient visits related to cardiovascular complications, infections, or weight-related issues may be associated with severe clinical outcomes, including sepsis, which likely further increases costs and reflects the fragility of this patient population.
- Patients experienced nearly 7 inpatient visits per year, corresponding to approximately $327,000 PPPY in hospital-based costs, indicating substantial acute care utilization.
- Outpatient visits occurred more than twice per month, highlighting the high burden of care.

**Unmet Need:**

- These findings highlight a substantial unmet need for interventions aimed at improving outcomes and reducing disease burden in patients with MCT8 deficiency.
- Leveraging linked databases in the absence of a dedicated ICD-10 code, this study provides the first real-world estimates of healthcare utilization and economic burden in MCT8 deficiency, underscoring the need for a dedicated ICD-10 code to improve patient identification and support future research.

## Limitations

- Consistent with rare diseases, the relatively smaller sample size, may make the generalizability of the findings limited.
- Certain data elements, including cost information and admission/discharge dates, were incomplete, which may have affected the comprehensiveness of the analysis.
- Claims and EHR data were combined for this study; however, this integration may have introduced the potential for underestimation of cost due to differences in data capture across data sources.
- Cost estimates for infection-related, sepsis-related, cardiac-related, and weight-related categories may be subject to overestimation, as these categories were not mutually exclusive, potentially resulting in overlapping attribution of costs.

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## References

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