# A case study of real-world health resource utilization and costs in a patient with neuronal ceroid lipofuscinosis type 2 (CLN2), an ultra-rare paediatric disease

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## Objectives

- Neuronal ceroid lipofuscinosis type 2 (CLN2) disease is an ultra-rare, neurodegenerative lysosomal disease with typically late-infantile onset. It follows a predictable clinical course characterized by frequent seizures, vision loss, and rapidly declining psychomotor function. Children affected by CLN2 disease have a typical life expectancy of 8–12 years[1]
- This case study aimed to better understand the healthcare resource use (HRU) associated with CLN2 disease under standard care, prior to the availability of enzyme replacement therapy (ERT)

## Methods

- HRU data (appointments, referrals, procedures/tests, and medications) for patients with a confirmed CLN2 disease diagnosis were collected retrospectively from medical records at Great Ormond Street Hospital, UK, using an electronic data capture (EDC) platform (**Table 1**)
- Reference costs were mapped on to each identified resource item and yearly costs calculated. Unit costs were sourced from the most recent (at the time of the study) NHS Reference Costs (2021/22), British National Formulary for Children (BNFC) (May 2022), and the Unit Costs of Health and Social Care Manual (2022) for hospital treatment costs, drug costs, and primary and community care costs, respectively[2,3,4]
- Analyses were stratified by stage of disease progression, as measured by total scores on the CLN2 Clinical Rating Scale (CRS)
  - The CLN2 CRS is a clinical measure of CLN2 disease progression consisting of four items (motor function, visual function, language, and seizures), each scored on a 0–3 scale to yield a 0–12 total summed score[5]
  - Where scores were not available for a patient visit, a consistent yearly rate of decline was mapped between any two scores entered in the EDC platform. Where no subsequent score was available, a decline of 2 points per year was assumed based on natural history[6]

### Table 1. Classification of HRU items

| HRU item | Details |
|---|---|
| Appointments and referrals | Specialist clinicians, specialist nurses, additional healthcare providers, complementary therapies, home and palliative care |
| Procedures and tests | Pulmonary hygiene, secretion management, feeding support, tests |
| Medications | Seizure management, dystonia, myoclonus, pain, management of mucus and secretions, breathing difficulties. |
| Additional costs | Adaptive devices, home adaptations, education |

## Results

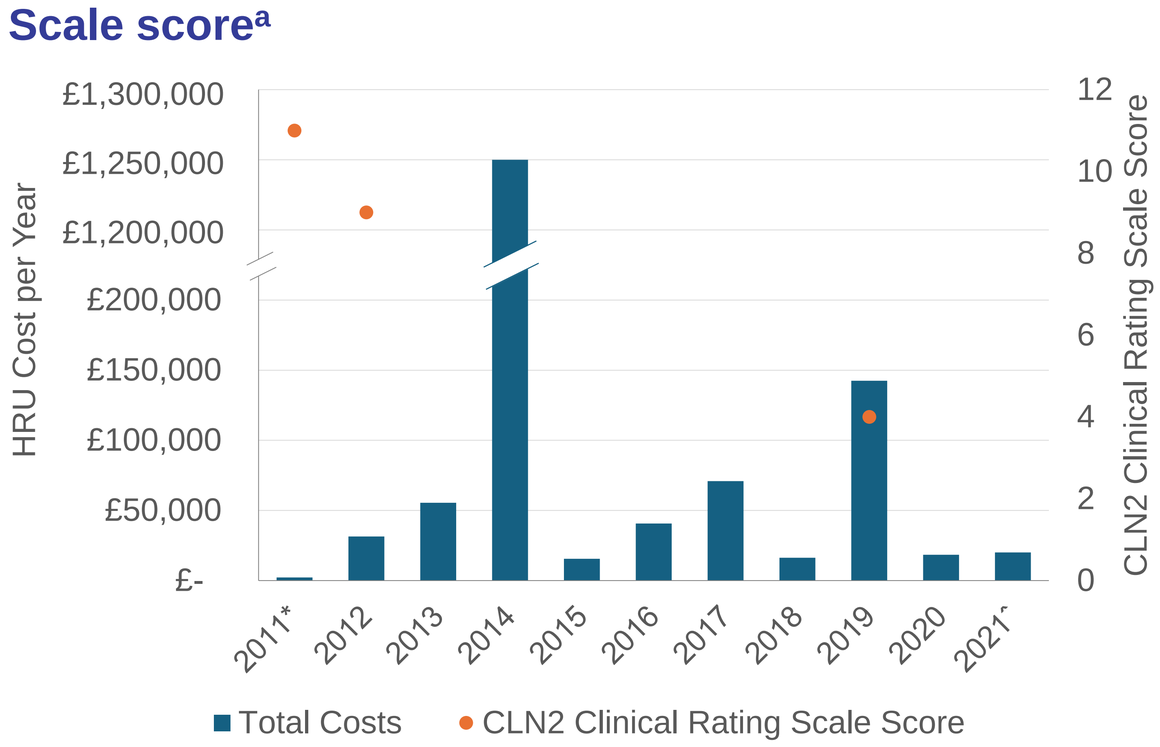
- Due to the ultra-rare prevalence of CLN2 disease and timing of availability of ERT in the UK, just one eligible patient was identified and enrolled. Data from hospital visits were collected over 9.92 years of follow-up until death (**Table 2**)
- On average, the patient had over 100 counts of resource use per year, of which 65 counts were medication related. This amounted to an estimated cost of £166,892.77 per year, of which £156,450.06 was for medications (**Table 2**)
- Costs varied considerably per year, with no clear trend identified between costs and disease progression (**Figure 1**)
  - The highest per-item medication costs were oxygen and intravenous- or rectally administered medications, all of which incurred an administration cost in addition to the drug cost. Significant use of such medication was noted in 2014, when the patient's CRS score was between 9 and 4

### Table 2. Mean annual HRU cost items and costs[a]

| | Cost items, n | Costs |
|---|---|---|
| Total follow-up time, y | 9.92 | |
| Appointments, per year | 13.00 | £3,268.91 |
| Procedures, per year | 5.44 | £3,643.49 |
| Additional cost items, per year | 3.33 | £3,520.04 |
| Referrals, per year | 13.51 | £10.27 |
| Medications, per year | 65.02 | £156,450.06 |
| **All HRU items, per year** | **100.30** | **£166,892.77** |

[a]Excludes HRU items that could not be costed (e.g. medications with missing dosage).

### Figure 1. Annual HRU costs and CLN2 Clinical Rating Scale score[a]




[a]Score as entered in the dataset for a visit during the calendar year *Start of follow-up in November 2011 ^Follow-up ended October 2021 upon recorded death

## Conclusions

- This study is the first of its kind in CLN2 disease, providing quantitative data on the estimated yearly costs associated with the management of a patient on standard care over long-term follow-up
- While data was only available for one patient, the patient was tracked across almost 10 years of hospital visits as their disease progressed
- There was no information about medication end dates within the available records. This may have led to overestimation of some medication costs. Disease progression was only recorded at three timepoints, thereby limiting analysis of HRU costs by disease stage.

### References

**1.** Steinfeld R; Heim P; von Gregory H et al. Am J Med Genet. 2002;112(114):347-54. **2.** NHS trusts and NHS foundation trusts. Available from: https://www.england.nhs.uk/costing-in-the-nhs/national-cost-collection/. **3.** National Institute for Health and Care Excellence. Available from: https://bnf.nice.org.uk/. **4.** Jones K; Weatherly H; Birch S et al. Available from: https://kar.kent.ac.uk/100519/. **5.** Wyrwich K; Schulz A; Kohlschütter A. JIEMS. 2018;6. **6.** Nickel, M; Simonati, A; Jacoby, D et al. Lancet Child Adolesc Health. 2018;2(8): 582-90.

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