PCR257

Real-world impact of X-linked myotubular myopathy (XLMTM) on caregivers in the United Kingdom (UK), Germany, and Spain

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Background

- X-linked myotubular myopathy (XLMTM) is a rare, life-threatening congenital disease caused by mutation of the *MTM1* gene, which affects the normal development, maturation, and maintenance of skeletal muscle[1,2]
- XLMTM is estimated to occur in about one in 40,000–50,000 newborn males[3,4]
- Most newborns males with XLMTM, particularly those with severe XLMTM, experience symptoms from birth[4,5]
- All individuals affected by XLMTM, irrespective of long-term survival, require constant lifelong care[1,2]
- Currently, there are no approved disease-modifying therapies for XLMTM, and the current standard of care is based on general recommendations for congenital myopathies made in 2012[6]
- Despite the substantial caregiving needs for an individual with XLMTM, data regarding the economic impact and health-related quality of life (HRQoL) for these caregivers are lacking

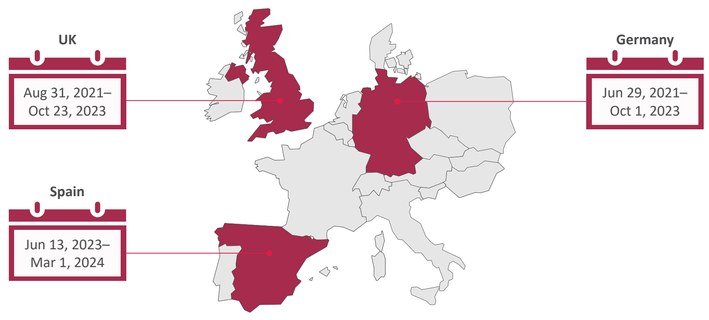
Objectives

- To evaluate:
  1. The economic impact of XLMTM, both work-related and financial, for caregivers of affected individuals
  2. HRQoL of caregivers of individuals with XLMTM

Methods

STUDY DESIGN

- A quantitative web-based survey was developed for caregivers of individuals with XLMTM, and was conducted in the UK, Germany, and Spain
  - The survey was reviewed and co-designed by patient advocacy leaders in Spain and the UK, who were also caregivers of individuals with XLMTM, in partnership with the survey team



- The survey was comprised of nine sections and 36 questions evaluating:
  1. The demographics of caregivers and individuals with XLMTM
  2. Physical characteristics and impact of the disease
  3. Caregiver HRQoL, as assessed using the EuroQol 5-dimension 5-level (EQ-5D-5L) and visual analog scale (VAS)
  4. Caregiver time and other costs

INCLUSION AND EXCLUSION CRITERIA

Participants were included if they were:

1. Caregivers or parents (≥18 years of age) of a male individual diagnosed with XLMTM who was not enrolled in the ASPIRO clinical trial (NCT03199469)
2. Residents of the UK, Germany, or Spain who were able and willing to provide written informed consent

STATISTICAL METHODOLOGY

- Average caregiver disutility was calculated by subtracting the caregiver-reported EQ-5D-5L index utility score from the EQ-5D population norm utility score; country- and age-specific EQ-5D population norm utility scores[7] were used to estimate the caregiver disutility for each respondent
- The number and percentage of hours of both paid and unpaid caregiving needs were calculated, summarised, and combined to determine the total hours of caregiving needs
- Annual unpaid caregiver productivity loss was calculated as:
  - Estimated productivity lost/year = (average hourly pay rate × (total employment hours reduction/week/family) × 52 weeks/year + (average hourly pay rate) × (days missed related to caring for an individual with XLMTM) × 8 work hours/day

ETHICS APPROVAL

- The study was reviewed and approved by an ethics committee in each country

Results

DEMOGRAPHICS AND CHARACTERISTICS IN THE UK

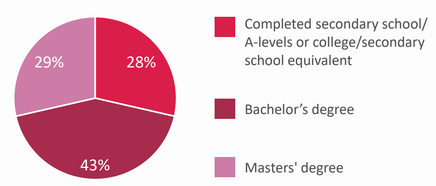
n=14 Caregiver respondents participated

Median age, years, (range): 44 (31–57)
Female, n (%): 9 (64)

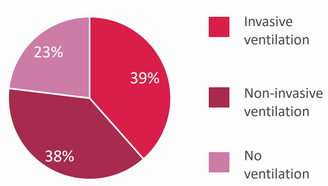
n=12 Individuals with XLMTM alive at the time of the survey

Median age, years, (range): N/A*

Highest level of education of primary household earner



Ventilation status†



DEMOGRAPHICS AND CHARACTERISTICS IN GERMANY

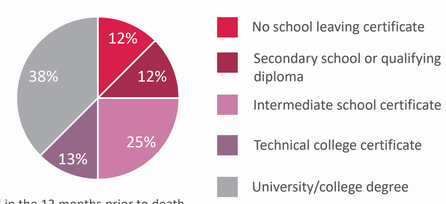
n=8 Caregiver respondents participated

Median age, years, (range): 32 (19–57)
Female, n (%): 7 (88)

n=7 Individuals with XLMTM alive at the time of the survey

Median age, years, (range): 10.5 (1–17)

Highest level of education of primary household earner



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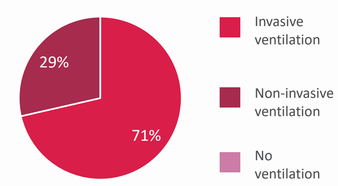


DEMOGRAPHICS AND CHARACTERISTICS IN SPAIN

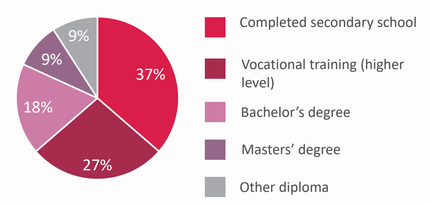
n=11 Caregiver respondents participated

Median age, years, (range): 40 (36–50)
Female, n (%): 7 (64)

n=11 Individuals with XLMTM alive at the time of the survey

Median age, years, (range): N/A*

Highest level of education of primary household earner



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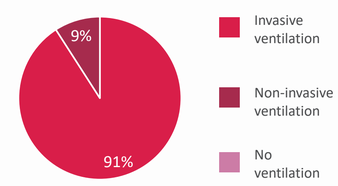
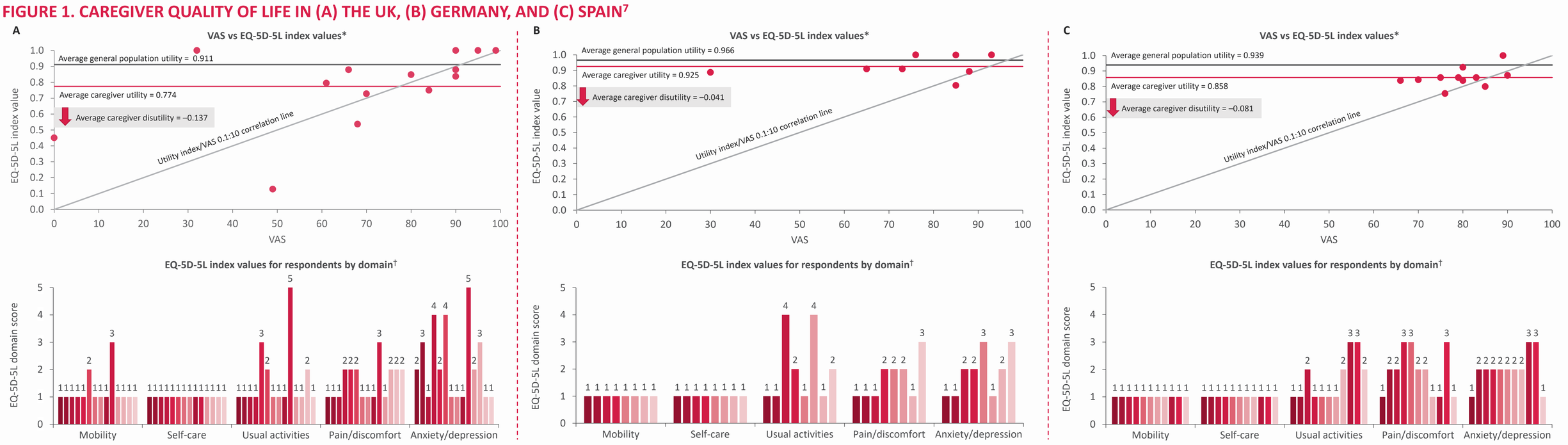


*The age of individuals with XLMTM was not captured due to an error in the LimeSurvey platform. †Current ventilation status or ventilation used in the 12 months prior to death.

HEALTH-RELATED QUALITY OF LIFE

- The estimated mean (standard deviation) disutility values in the UK, Germany, and Spain were −0.137 (0.25), −0.041 (0.07), and −0.081 (0.06), respectively (**Figure 1**), indicating reduced quality of life among caregivers of individuals with XLMTM compared with the general population
- Among caregivers in all countries, the HRQoL domains most impacted were anxiety/depression, pain/discomfort, and usual activities

FIGURE 1. CAREGIVER QUALITY OF LIFE IN (A) THE UK, (B) GERMANY, AND (C) SPAIN[7]

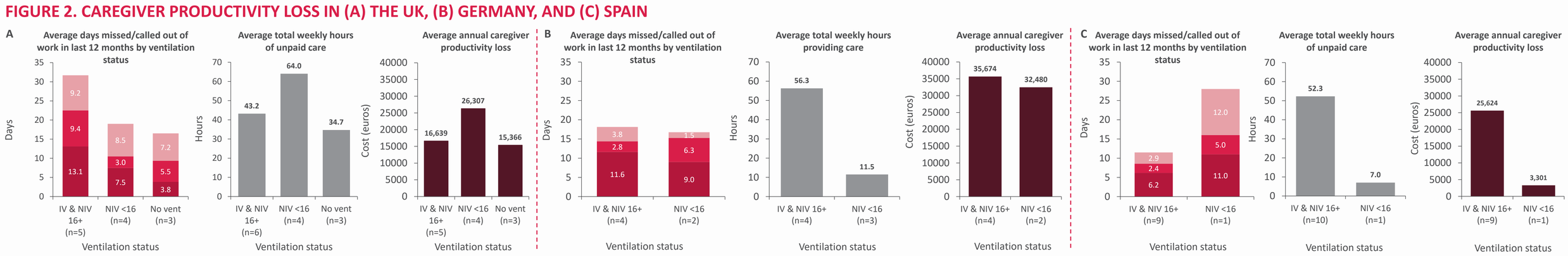


*For the EQ-5D index, a score of 1 represents the best possible health state and a score of 0 represents the worst possible health state. For the EQ-5D VAS, a score of 100 represents the best health imaginable and 0 represents the worst health imaginable.
†EQ-5D domain scores are reported as one of 5 levels: 1 = no problems; 2 = slight problems; 3 = moderate problems; 4 = severe problems; and 5 = extreme problems.

ECONOMIC IMPACT

- Across all countries, lost working hours and time spent on caring for the individual with XLMTM was substantial (**Figure 2**)
- Median (range) annual caregiver productivity loss in the UK, Germany, and Spain was €18,692 (€0–42,300), €24,915 (€13,137–79,728), and €29,113 (€1,400–36,783), respectively

FIGURE 2. CAREGIVER PRODUCTIVITY LOSS IN (A) THE UK, (B) GERMANY, AND (C) SPAIN



SUMMARY

- Caregivers reported substantial financial constraints, productivity loss, and poor HRQoL for both physical and mental health, regardless of location
  - However, due to the challenges posed by the small sample size in this rare disease population, interpretation of these data should be made carefully
- These real-world data highlight the need for improved support for caregivers, as well as targeted therapies for individuals with XLMTM

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ABBREVIATIONS EQ-5D-5L, EuroQol 5-dimension 5-level; HRQoL, health-related quality of life; MTM, myotubular myopathy; VAS, visual analog scale; XLMTM, X-linked myotubular myopathy.

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