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Introduction

- Duchenne muscular dystrophy (DMD) is a rare disease with an estimated incidence of 1 in 3,300 male births. It is a neuromuscular disorder with X-linked recessive inheritance, and for this reason, females are typically asymptomatic and considered healthy carriers (1-3).
- DMD represents a significant burden in terms of healthcare and non-healthcare resource utilization.
- Despite notable therapeutic advancements (4,5), ensuring economic sustainability and operational efficiency remains a critical challenge for the National Healthcare Service.
- This study aimed to assess the clinical and non-clinical burden of DMD from the perspective of the Italian National Healthcare System (NHS).

Methods

- An observational, multicentre, cross-sectional study is ongoing across five Italian DMD centres.
- Data on ambulatory and non-ambulatory patients, including clinical, demographic, and healthcare service use, were collected via questionnaire.
- Subjects with a confirmed diagnosis of DMD for at least 12 months, who will visit the participating centers for a follow-up appointment between May 2023 and June 2025, or within 6 months following the approval of the protocol by the ethics committee, were enrolled.
- In the study the follows information were collected:
  - Demographic data (i.e., age, gender, household composition, education, and employment status);
  - Clinical data (i.e., severity of the disease, date of diagnosis, etc.);
    - Data on disease severity were collected using the follows validate scale: North Star Ambulatory Assessment (NSAA), Time to Rise from the Floor (TRF), 6-Minute Walk Distance (6MWD), 10-Meter Walk/Run Test (10MWT), Supine Up Time, and Performance Upper Limb Scale (PUL).
  - Hospital care (i.e., hospitalizations, day hospital admissions, long-term admissions, and rehabilitation);
  - Outpatient care (i.e., visits, exams and tests, medications), equipment, aids, medical devices, home modification interventions, and purchase of transport vehicles, community care (home nursing visits, home care, transportation), and family assistance (informal care).
  - Data on the resource used by patients in the 12 months prior the enrolment date were collected through a dedicated questionnaire.
- The patients' demographic and clinical data were reported as frequencies and percentages for categorical variables, as mean and standard deviation for continuous variables, or as median (interquartile range) for discrete variables.
- Here preliminary data from interim analysis were reported.

Conclusions

- The preliminary findings provide a glimpse into the considerable burden of DMD and emphasizing the urgent unmet need for disease-modifiers.
- These findings highlight the importance of tailoring broader societal intervention to mitigate the DMD impact for patients, caregivers and society.

Reference

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Results

- In the study twelve patients were included: 2 ambulatory and 10 non-ambulatory.
- The mean age of ambulatory patients was 16.00 years (SD: 2.83), and 21.10 years (SD: 5.84) for non-ambulatory patients. 50% of ambulatory patients had completed lower secondary education; among non-ambulatory patients 40%completed lower and 40% upper secondary education. All patients live with parents.
- All ambulatory patients were Stage 2. Non-ambulatory patients ranged from Stage 4 (early non-ambulatory) to 8 (full ventilation): 3 were stage 4, 3 were stage 7, 2 were stage 8, 1 were stage 5, and 1 were stage 6.
- Among the two groups, half of patients were unable to perform the “Time to Rise from Floor” test; the others completed the 6-Minute Walk and 10-Meter Walk/Run tests. Median NSAA was 5.00 (IQR: 3.50-6.50).
- Among non-ambulatory patients, nine completed the Upper Limb Module, median NSAA 0.00 (IQR: 0.00-0.25).
- In the previous year, 80% of non-ambulatory patients were hospitalized, totalling 168 nights (across internal medicine, neurology, and surgery wards).
- High healthcare utilization was reported in non-ambulatory patients, including medical consultations and diagnostics tests such as ECGs (80%), pulmonary function tests (70%) and other such as CT scans, MRIs, and DEXA.
- All ambulatory patients used assistive devices and adapted vehicles; most non-ambulatory used wheelchairs and home modifications.

Table 1. Demographic, Clinical characteristics, and resource used by DMD patients.

Variable	Ambulatory	Non-Ambulatory	Variable	Ambulatory	Non-Ambulatory
Demographic variables			Clinical Variables		
Age (mean, SD)	16.00 (2.83)	21.10 (5.84)	Time to rise from the floor - (N, %)		
Sex, male (N, %)	2 (100.00%)	10 (100.00%)	Not able to perform the test	1 (50.00%)	5 (50.00%)
Weight (mean, SD)	46.00 (11.31)	52.30 (19.71)	6- Minute Walk Distance (6MWD) - (N, %)		
Height (mean, SD)	145.00 (26.87)	158.70 (12.30)	Able to perform the test	1 (50.00%)	10 (10.00%)
Caregive, Parents (N, %)	2 (100.00%)	10 (100.00%)	10-Meter Walk/Run Test (10MWT) - (N, %)		
Resource utilization			Able to perform the test	1 (50.00%)	5 (50.00%)
Hospitalization in the last 12 months (N, %)	1 (50.00%)	8 (80.00%)	Performance of the Upper Limb Module for DMD (PUL for DMD)		
Number of night in hospital by ward (sum)			Preferred arm - (N, %)		
Nights in Internal medicine	0	50	Right	2 (100.00%)	9 (100.00%)
Nights in surgery	0	3	Left	0 (0.00%)	0 (0.00%)
Nights in neurology	0	27	Elbow extension ROM Right arm - (N, %)		
Nights in other ward	2	88	Full	1 (50.00%)	4 (44.44%)
Aids and devices (N, %)			3/4	1 (50.00%)	1 (11.11%)
Support devices	2 (100.00%)	3 (30.00%)	1/2	0 (0.00%)	1 (11.11%)
Wheelchair	1 (50.00%)	7 (70.00%)	1/4	0 (0.00%)	3 (33.33%)
Adjustable bed	0 (0.00%)	2 (20.00%)	Elbow extension ROM Left arm - N, %		
Anti-decubitus devices	0 (0.00%)	2 (20.00%)	Full	1 (50.00%)	4 (50.00%)
Stair lift	1 (50.00%)	4 (40.00%)	3/4	1 (50.00%)	0 (0.00%)
Shower chair/reclinig chair for personal hygiene	0 (0.00%)	6 (60.00%)		0 (0.00%)	0 (0.00%)
Car adaptation	2 (100.00%)	5 (50.00%)	1/2		
Other devices	0 (0.00%)	2 (20.00%)	1/4	0 (0.00%)	4 (50.00%)
			NSAA score (Median, IQR)	5.00 (3.50 – 6.50)	0.00 (0.00 – 0.25)

Acknowledgements

- The study was supported by an unrestricted grant by Roche S.p.A.
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