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RESEARCH

Prevalence of Duchenne muscular dystrophy in Italy: a nationwide survey

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Abstract

Purpose The availability of care recommendations has improved survival and delayed the progression of clinical signs in Duchenne muscular dystrophy. The aim of the study was to perform a nationwide survey investigating the prevalence, age distribution, and functional status of Duchenne muscular dystrophyin Italy.

Methods The survey was performed by collecting data from all 31 reference centers for Duchenne muscular dystrophy in Italy using a structured form. We assessed age distribution, motor function, and the need for respiratory and nutritional support to evaluate their prevalence in different age and functional subgroups.

Results The estimated prevalence was 1.65/100,000 (3.4/100,000 males). There were 972 boys and adults with a confirmed diagnosis of Duchenne, of age ranging between 6 months and 48 years (mean = 16.5). Over 59% were below the age of 18 years and the remaining 41% were adults. Over 43% were ambulant and 57% non-ambulant; 14.7% were steroids naive (mean 20.6 years), 75% are currently on steroids (mean 14.6 years) with 604 on the daily regime, 126 intermittent. Nearly 73% did not require any ventilatory support, 16% had NIV ≤ 12 h, 9% > 12 h, and 1.4% had a tracheostomy. More than 82% did not require any nutritional support, 13% required food modification/semisolid and 4.4% had a G-tube.

Conclusions: Our findings provide information to be used not only for epidemiological purposes but also for possible trial design to include older non-ambulant patients who until recently have been excluded and for whom clinical information is limited.

What is Known

- *Duchenne muscular dystrophy is a progressive disorder associated with reduced survival.*
- *As part of the disorder there is also a progressive loss of important milestones, including loss of ambulation, and increased need for respiratory and nutritional support.*

What is New

- *Our nationwide survey provides prevalence, age distribution, and functional status for Duchenne muscular dystrophyin Italy including both boys and adults.*
- *Our findings can be used for epidemiological purposes and for possible trial design.*

Keywords Prevalence · Duchenne muscular dystrophy · Respiratory

Abbreviations

DMD	Duchenne muscular dystrophy
CI	Confidence interval
SD	Standard deviation

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NIV Non-invasive ventilation
PEG Percutaneous endoscopic gastrostomy
CK Creatine kinase

Introduction

Duchenne muscular dystrophy (DMD), is an X-linked recessive disorder caused by mutations in the *DMD* gene causing deficiency of the dystrophin protein [1]. The clinical phenotype is characterized by progressive muscle weakness, leading to loss of ambulation and upper limb function, and to progressive cardiac and respiratory impairment with premature death. Historically, DMD was associated with loss of ambulation before the age of 12 years and reduced survival beyond 18 years [2]. Treatment with corticosteroids [3], and the introduction of cough-assist devices, mechanical ventilation [4, 5], and prophylactic cardiac medication use [6] have helped to slow down progression with increased survival and a delay of the age when loss of ambulation, cardiac and respiratory problems occur [7].

Over the years, several studies have reported epidemiological data on DMD [8, 9]. A systematic review and meta-analysis based on epidemiological studies reported that the pooled global DMD prevalence was 7.1 cases (95% CI 5.0–10.1) per 100,000 males and 2.8 cases (95% CI 1.6–4.6) per 100,000 in the general population, with a very high variability among studies [8]. A subsequent one using different criteria for data selection reported a prevalence of DMD at 4.8 per 100,000 people (95 CI 3.6–6.3 per 100,000 people)[9].

The high variability is partly probably related to the fact that the studies had been performed in different decades. As many of them report data collected between the 1970s and the 1990s [10, 11], some differences may be explained by changes in standards of care that have progressively led to increased survival. With the introduction of non-invasive ventilation and prophylactic cardiac treatment, together with the improvements in several aspects of management, survival has progressively increased and the mean survival is now approximately 29.5 years [7].

Data sources for the published studies were multiple, ranging from surveys to registries or electronic records and mainly focused on epidemiological data with little or no mention of their functional status [8, 9].

The aim of this study was to report the results of a nationwide survey investigating epidemiological data, from all 31 Italian reference centers for DMD in order to establish the prevalence and distribution of the whole cohort at the national level. We were also interested in providing information on the current survival data and on a number of functional endpoints, including the need for ventilatory and nutritional support, at different ages.

Materials and methods

The study includes all the 31 centers/units identified by the Italian government as referral centers for DMD throughout the whole national territory. As part of the Italian care system, only these centers are allowed to prescribe drugs or provide tertiary care for DMD.

The study is part of an ongoing prospective study aimed at establishing the natural history of DMD. The study was approved by the Ethics Committee.

The centers were asked to report the number of DMD male patients and to provide details on age, motor functional status, need for cardiac and respiratory support, corticosteroid treatment or other concomitant therapies, and the type and site of mutation in the *DMD gene*. Only patients currently followed or seen in the last 3 years (January 2021–December 2023) were included.

Patients were only included if the diagnosis of DMD was based on genetic testing using previously reported criteria [12] and had onset and early progression of clinical signs, such as weakness and motor delay, suggestive of a DMD phenotype [7]. In the absence of genetic testing, patients were only included if they had a muscle biopsy with absent dystrophin. As some centers only included data from DMD boys and male young adults and female symptomatic carriers had not been systematically collected in all centers, these were excluded from this study.

In order to avoid cases of patient duplicates for those who may be seen in more than one center, a unique global identifier number was generated and the results from each center were centrally reviewed. The study is adherent to the relevant Strobe checklist.

The period prevalence was calculated as the proportion of persons affected by Duchenne divided by the Italian population reported by the Istituto Nazionale Statistica (ISTAT) for 2023 (58,850,717 persons). As in this study, we only included males, we also calculated the prevalence of persons affected by Duchenne divided by the Italian male population (28,762,901 persons).

Results

Nine hundred seventy-two DMD patients were reported to be currently followed in the 31 participating centers. Consent to participate in this study was obtained in all 972. The estimated prevalence for all cases of DMD divided by the whole Italian population is 1.65/100,000. When calculating for males only the prevalence was 3.4/100,000 males. Their age ranged between 6 months and 48 years (mean = 16.5; median = 16; SD = 8.2).

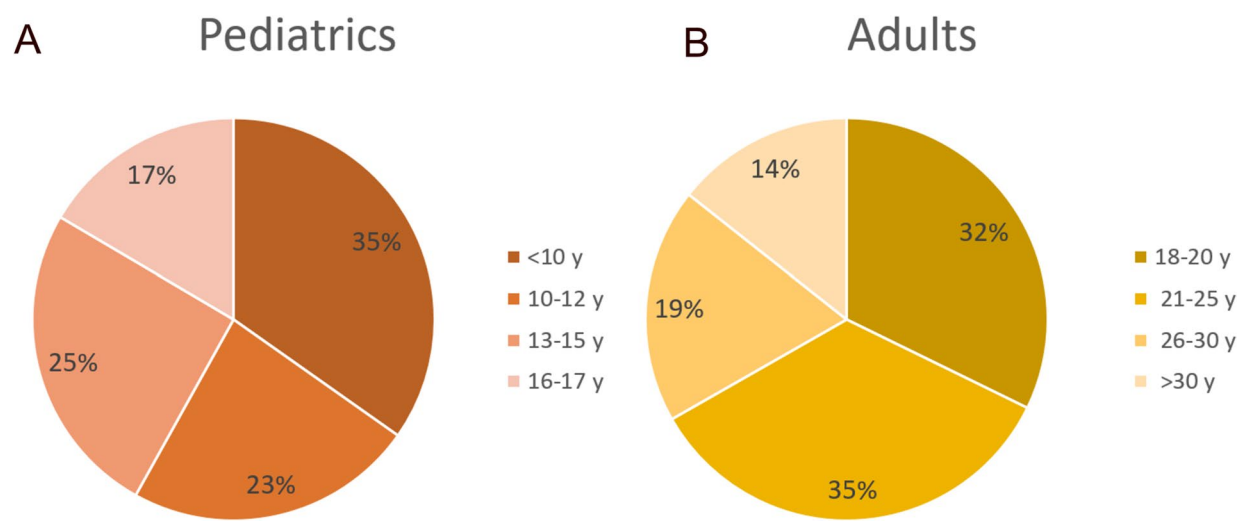

Fig. 1 Distribution of pediatric (**A**) and adult (**B**) DMD patients according to age groups

Table 1 Characteristics of the cohort

Age group	Number of subjects	Ambulant	Non-ambulant	Ventilatory support	Nutritional support	Steroids
< 3	16	16	0	No support: 16 NIV < 12 h: 0 NIV > 12 h:0 Tracheostomy: 0	Solid: 16 Semisolid: 0 G-tube: 0	No: 15 Intermittent: 0 Daily: 1 Previous: 0
3–6	91	91	0	No support: 91 NIV < 12 h: 0 NIV > 12 h: 0 Tracheostomy: 0	Solid: 91 Semisolid: 0 G-tube: 0	No: 22 Intermittent: 9 Daily: 60 Previous: 0
7–12	227	180	47	No support: 226 NIV < 12 h: 1 NIV > 12 h: 0 Tracheostomy: 0	Solid: 226 Semi-solid: 1 G-tube: 0	No: 10 Intermittent: 13 Daily: 204 Previous: 0
13–15	146	64	82	No support: 137 NIV < 12 h: 9 NIV > 12 h: 0 Tracheostomy: 0	Solid: 142 Semi-solid: 4 G-tube: 0	No: 6 Intermittent: 10 Daily: 126 Previous: 4
16–17	95	21	74	No support:77 NIV < 12 h: 17 NIV > 12 h: 0 Tracheostomy: 1	Solid: 87 Semi-solid: 7 G-tube: 1	No: 6 Intermittent: 18 Daily: 68 Previous:3
18–20	128	20	108	No support:87 NIV < 12 h: 35 NIV > 12 h: 4 Tracheostomy: 2	Solid: 111 Semi-solid: 11 G-tube: 6	No: 12 Intermittent: 31 Daily: 75 Previous:10
21–25	137	10	127	No support:58 NIV < 12 h: 55 NIV > 12 h: 22 Tracheostomy: 2	Solid: 85 Semi-solid: 42 Peg: 10	No: 19 Intermittent: 33 Daily: 50 Previous: 35
26–30	75	2	73	No support: 12 NIV < 12 h: 29 NI V > 12 h: 31 Tracheostomy: 3	Solid: 30 Semi-solid:32 G-tube: 13	No: 27 Intermittent: 9 Daily: 15 Previous:24
> 30	57	0	57	No support: 6 NIV < 12 h: 9 NIV > 12 h: 36 Tracheostomy: 6	Solid: 14 Semi-solid: 30 G-tube: 13	No: 32 Intermittent: 3 Daily: 5 Previous: 17

Of the 972 (59%), 575 were below the age of 18 years and 397 adults (41%) (Fig. 1A, B).

Six hundred twenty-nine of the 972 (65%) had deletions in the *DMD* gene, 104 (11%) duplications, and 206 (21%) small mutations, in 33 (3%) mutation status was unknown. Sixty-six of the 629 with deletions (10.5%) had mutations eligible for skipping exon 44 (6.8% of the whole DMD cohort), 94 (14.5% of deletions, and 9.7% of whole cohort) eligible for skipping exon 45, 122 (19.4% of deletions and 12.5% of whole cohort) eligible for skipping exon 51, 94 (14.9% of deletions and 9.7% of whole cohort) eligible for skipping exon 53. All these findings are in agreement with the results of our published genetic study [13]. Table 1 shows the cohort characteristics according to age groups.

Functional status

At the last assessment 425 (44%) of the 972 patients were ambulant and 544 (56%) non-ambulant; 3 patients were too young to acquire ambulation (< 18 months of age). In the non-ambulant subgroup, the age at loss of ambulation ranged between 6 and 25 years (mean 11.9, median 11.5 years; SD 2.98).

The percentage of patients losing ambulation before the age of 12 years was higher in the boys carrying deletions than in those with duplications or small mutations (Fig. 2A, B reports details of age at loss of ambulation).

Respiratory support

Seven hundred ten (73%) DMD individuals did not require any ventilatory support (age range 0.5–34.2 years, mean 13.3 years, median 13.0, SD 5.97), 155 (16%) had NIV ≤ 12 h (age range 10.1–39 years, mean 22.2 years, median 22.0, SD 4.99), 93 (10%) had NIV > 12 h (age range 19–48 years, mean 29.7 years, median 28.0, SD 6.61), and 14(1%) had tracheostomy (age range 16–45 years, mean 29.0 years, median 29.2, SD 7.82) (Fig. 3A, B).

The mean age when ventilation was started was 19.1 years (range 7–28.7 years, median 18).

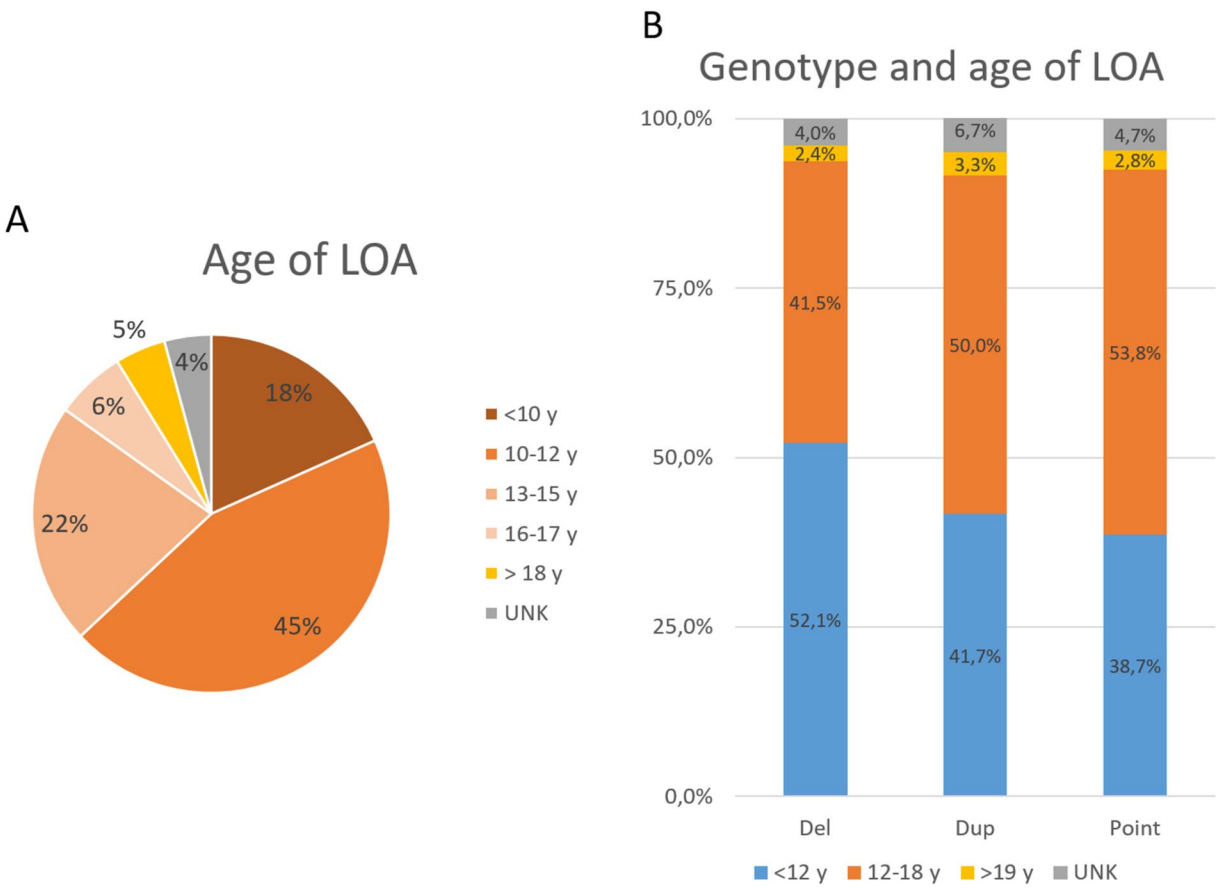

Fig. 2 Details of loss of ambulation (LOA) subdivided into age groups (**A**) and according to genotype (**B**)

Nutritional support

Seven hundred ninety-seven (82%) DMD individuals did not require any nutritional support (age range 0.5–47 years, mean 14.5 years, median 14.0, SD 6.66), 43 (13%) had PEG (age range 17.5–48 years, mean 28.1 years, median 26.0, SD 7.56), 127 (5%) required food modification /semisolid (age range 10–47 years, mean 26.0 years, median 25.2, SD 6.55) (Fig. 4).

Steroids

One hundred forty-three (15%) DMD individuals were steroids naive (age range 0.5–48 years, mean 20.6 years, median 21.56, SD 12.5), 730 (75%) are currently on steroids (age range 2.8–35 years, mean 14.6 years, median 14.0, SD 5.98) with 604 on daily regime and 126 intermittent, and 93 (10%) used to take steroids in the past (age range 13–45 years, mean 25.3 years, median 24.7, SD 5.86). For six patients this data was not available (Fig. 5).

Other drugs (clinical trials)

Of the 972 patients, 149 (15%) are currently or have been previously enrolled in clinical trials.

Discussion

Our results, obtained as part of a nationwide survey, include 972 DMD patients with an estimated prevalence of 1.65/100,000. This value is somehow lower than previous reports. In two recent systematic reviews, the pooled global prevalence for DMD was estimated to be 4.6:100,000 [9] and 2.8:100,000 [8], respectively. Both articles however highlighted a significant heterogeneity among the cohorts studied. In the two reviews, the prevalence values ranged from 0.7 to 7.7: 100,000 and between 0.7 and 16.7: 100,000 respectively. In both papers, the discrepancies were thought to be likely due to different study designs and inclusion criteria for a DMD diagnosis, with the older studies not based on genetic analysis and the more recent ones often including patients identified by multiple tools, such as increased CK levels, sometimes with no clear indication of possible overlaps with other dystrophinopathies or even other muscular dystrophies. This led the authors to wish for a commonly shared methodology, together with the call for nationwide approaches in order to mitigate the risk for biases.

When we compared our nationwide results to the previously published studies performed in Italy we found three studies [10, 14, 15] with a global prevalence estimated to be 1.69, 3.31, and 3.4:100,000 respectively. The comparison

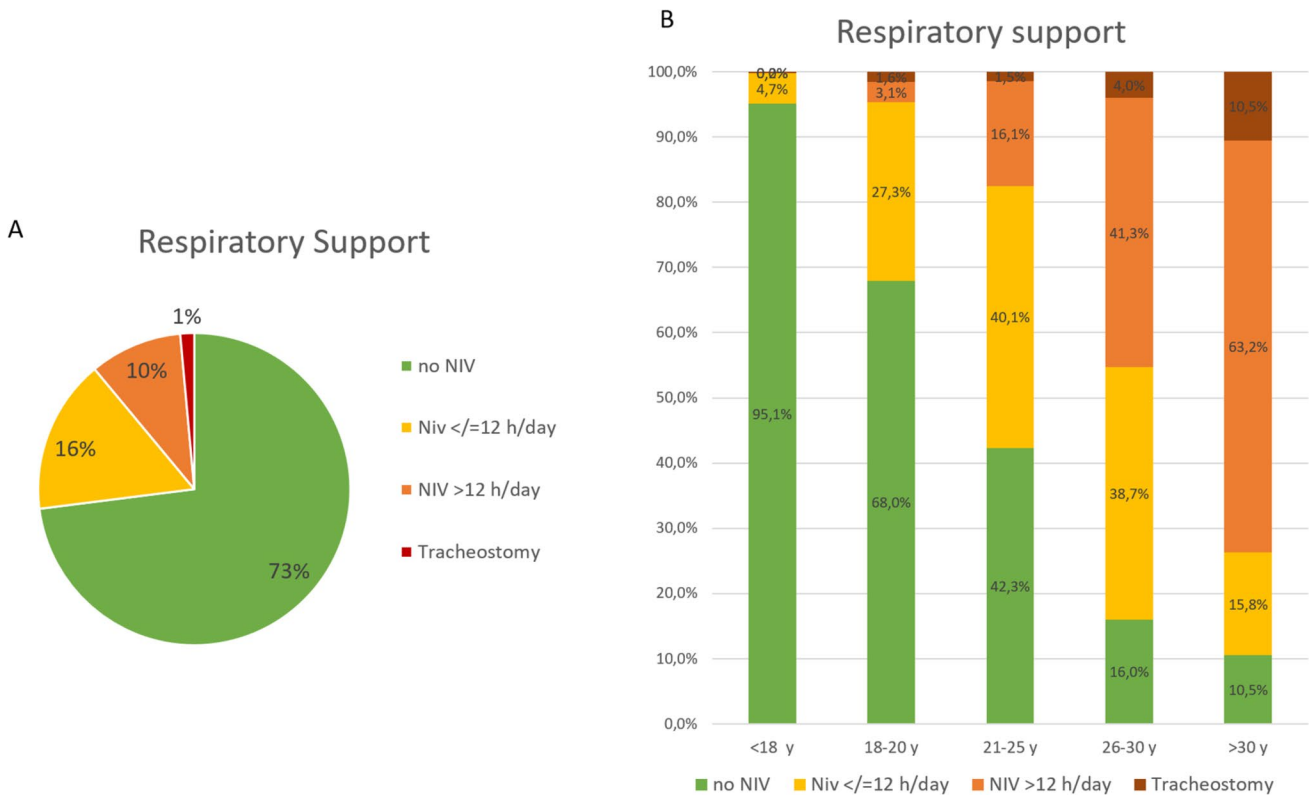

Fig. 3 Distribution of the need for respiratory support (**A**) also in relation to age groups (**B**)

is however difficult, as all the studies were performed over 20 years ago and 2 of the 3 studies did not include genetic testing in the inclusion criteria for a DMD diagnosis. Furthermore, while we report nationwide data involving all of the reference centers with a cross-sectional interpretation of prospectively collected data, all three previous studies only reflected regional data, with sample sizing calculated over regional population data.

The survey also allowed us to draw a picture of the number of patients treated with steroids with a clear reduction of patients on steroids with increasing age. It is of note that while until two decades ago steroids were usually stopped at the time when patients lost ambulation, in our cohort nearly 2/3 of the non-ambulant patients between the ages of 18 and 25 were still on steroids, in agreement with the more recent recommendations [7] and clinical evidence of a beneficial effect on upper limb [16] and respiratory function [17, 18].

The survey also allowed us to establish the frequency of respiratory and nutritional difficulties in different age groups. More than 70% of our patients did not need ventilatory support. Not surprisingly the percentage of patients requiring NIV was very low in the pediatric patients (approx 5%) and progressively increased with age: the percentage rose to approximately 30% between 18.1 and 20.11 years, and by the age of 26 years only 14% were ventilation free. Although the results cannot be easily compared, these values are in line with previous papers reporting the need for respiratory support in 21.2% [19] and 39.6% [3] of patients by the age of 20 and with a recent study from an Italian center reporting that all patients between 25 and 48 years were mechanically ventilated with continuous invasive ventilation via tracheal tube in 26% of the cohort [20].

The median age at any respiratory support in our whole cohort was 18 years (mean 19.12), also in line with the previously reported age at NIV that ranged from 18.0 years (reported median value) [19] to 22.3 years (reported mean value) [21].

A similar progression was also found when analyzing the need for food adaptations or nutritional support even though, at variance with respiratory support, the age when these were needed was shifted to after the age of 20 years rather than 18. The need for cardiological therapies was also recorded but as the participating centers had different policies on prophylactic treatments, this requires further details. The increased survival and the relatively late onset of the need for respiratory and nutritional support are likely to

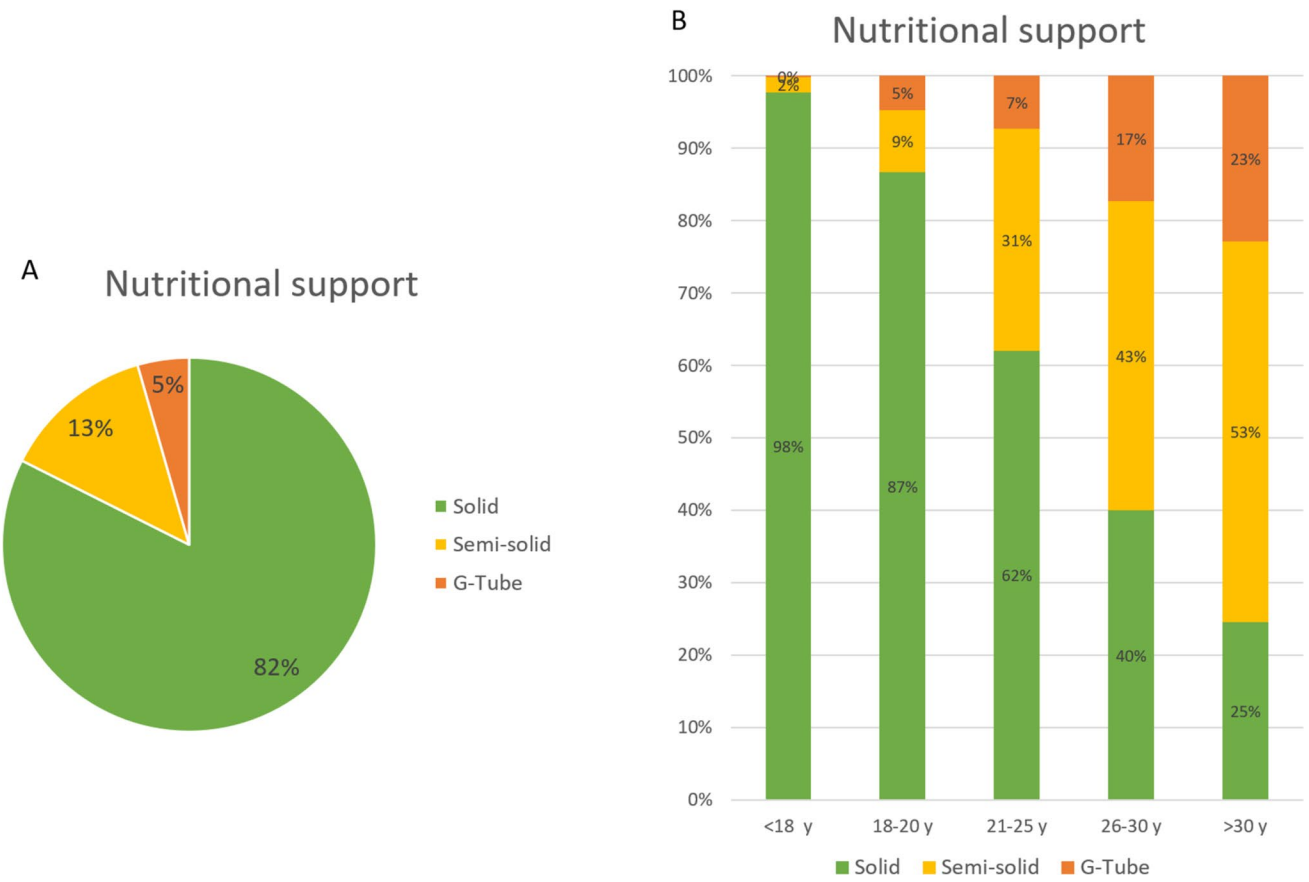

Fig. 4 Distribution of the need for nutritional support (**A**) also in relation to age groups (**B**)

be the result of the introduction of steroids and improvements in standards of care (non-invasive ventilation, cough machine) in patients born in the last three decades [4, 5].

As the Italian Health system allows follow-up and prescription of drugs only in specialized tertiary care centers for DMD, having included all of them and having had all the patients consenting to participate in the study, we believe that our cohort reflects the total number of DMD in Italy. We cannot however exclude that a number of adult patients may not be followed in the participating centers or that, especially at the time of COVID, a number of them may have electively decided not to attend follow-up appointments in a tertiary care center. While we acknowledge that our survey may not capture the totality of adults, it is more unlikely that younger boys who had a diagnosis did not reach a tertiary care center.

Even with these limitations, our nationwide survey provides the largest cohort so far reported in epidemiological studies at the national level. Our study has the advantage that it had strict inclusion criteria to define DMD that were shared across the centers and even if retrospective in nature, the data had been collected as part of a long-term prospective data collection. The relatively lower prevalence found in our cohort compared to older studies [8, 9] may be justified by the stricter inclusion diagnostic criteria, excluding other dystrophinopathies. Our findings, also providing details of ambulant status and need for respiratory and nutritional support will be of help to better identify subgroups of patients who may be enrolled in clinical trials or may benefit from new drugs or other types of intervention.

Although the survey was not designed to specifically capture details of progression over the years or to cross-reference different clinical findings, the results suggest that, when compared to previous studies reporting motor,

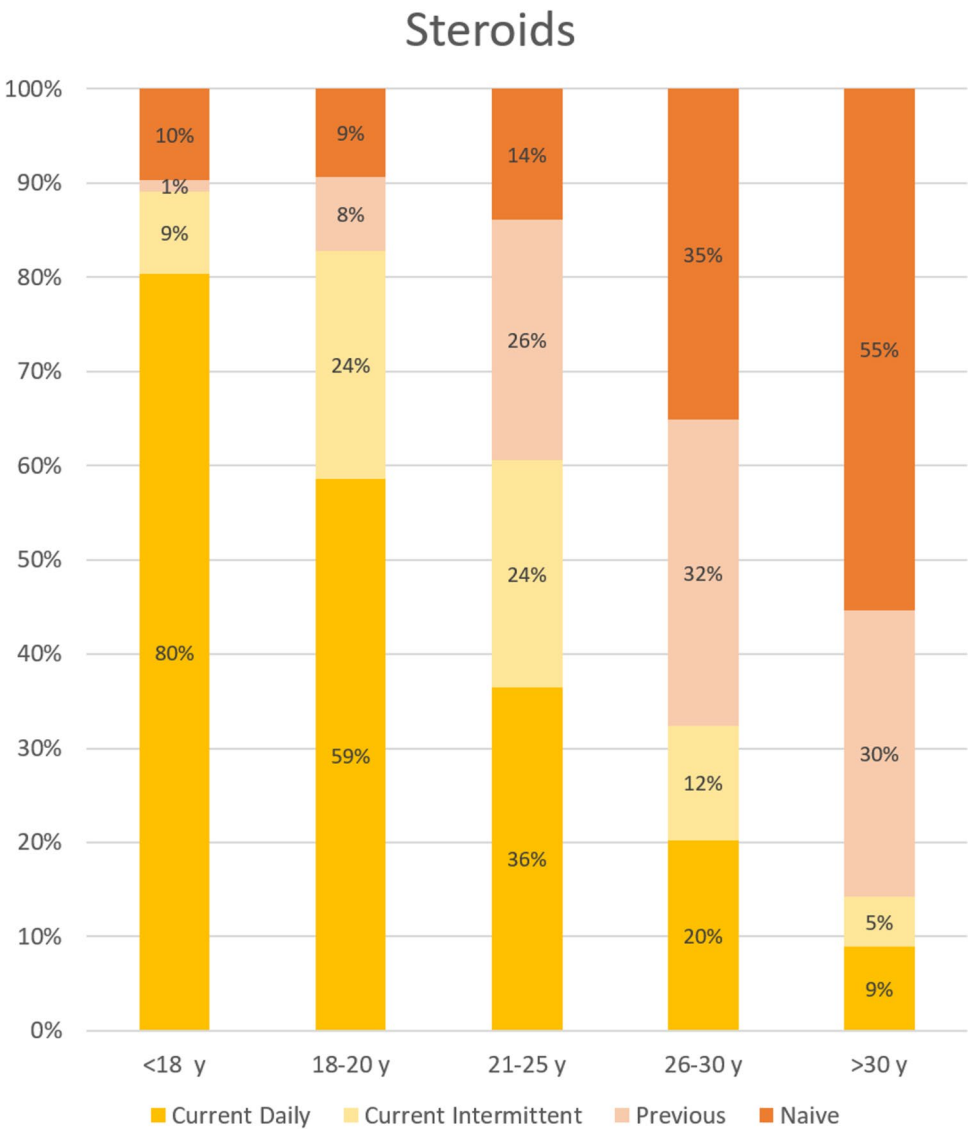

Fig. 5 Distribution of steroid use in relation to age groups

respiratory, and nutritional involvement, there is a wider variability that is likely to be the result of evolving standards of care. A more detailed analysis aimed at establishing the possible effect of participation in clinical trials or other variables is ongoing also to better understand which are the factors that may contribute to delayed loss of ambulation in boys who had mutations and early clinical findings suggestive of DMD.

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Author Contribution All authors contributed to the study conception, design and data collection. Material preparation and analysis were performed by AC, GC, MR, MP, GCor, AB, RM, MGDA, EP, ADA, SM, VAS, CB and EM. The first draft of the manuscript was written by AC, GC, MR, MP, GCor and EM and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript. The Italian DMD working group participated in the acquisition of data.

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Data Availability All data will be made available on request.

Declarations

Ethics approval This study was performed in line with the principles of the Declaration of Helsinki. Approval was granted by the Ethics Committee of the Catholic University of Rome.

Consent to participate Informed consent was obtained from all individual participants included in the study.

Competing Interests The authors declare no competing interests.

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