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An incidence- and survival-based epidemiological model for the prevalence of Duchenne muscular dystrophy in nine countries

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Introduction

- Duchenne muscular dystrophy (DMD) is an X-linked, rare neuromuscular disease caused by mutations in the *DMD* gene that disrupt the production of functional dystrophin protein, leading to progressive muscle weakness, diminished quality of life and premature death.[1–3]
- Existing estimates of global prevalence of DMD are variable, ranging from 0.9 to 16.8 cases per 100 000 males across 15 studies published from 1982 to 2016, and from 0.7 to 7.7 cases per 100 000 in the general population across five studies published from 1993 to 2005.[4]
- There is a high degree of heterogeneity across existing estimates of global prevalence of DMD, highlighting the need for further high-quality studies to understand better the epidemiology of DMD and the resulting burden of disease.[4]

Objective

- To develop an epidemiology model of DMD, providing prevalence estimates of the total diagnosed population and selected subpopulations in Brazil, Canada, China, France, Germany, Italy, Japan, Spain and the UK.

Methods

- To estimate DMD prevalence, data on live male births were derived from the 2022 Revision of World Population Prospects (United Nations), a database presenting country population estimates from 1950 to present.
- A literature review was conducted to identify estimates of incidence, age at diagnosis and survival for each country.
- Survival was based on period of birth (pre-1970, 1970–1990, post-1990) and accounted for potential differences in median age of death by country.
- Data on the proportion of ambulatory and non-ambulatory patients by age group were derived from an international observational study of 440 patients with DMD.[5]
- Diagnosed incidences, considering historic trends in rate, were applied to the effective number of live male births and entered the model according to the age at diagnosis; survival curves were then used to estimate prevalence of DMD overall, by age group and by ambulatory status.
- To assess model validity, estimated DMD prevalence was compared with the published prevalence in studies from Italy,[6] Japan[7] and the UK.[8,9]

Results

Estimated total diagnosed prevalence of DMD

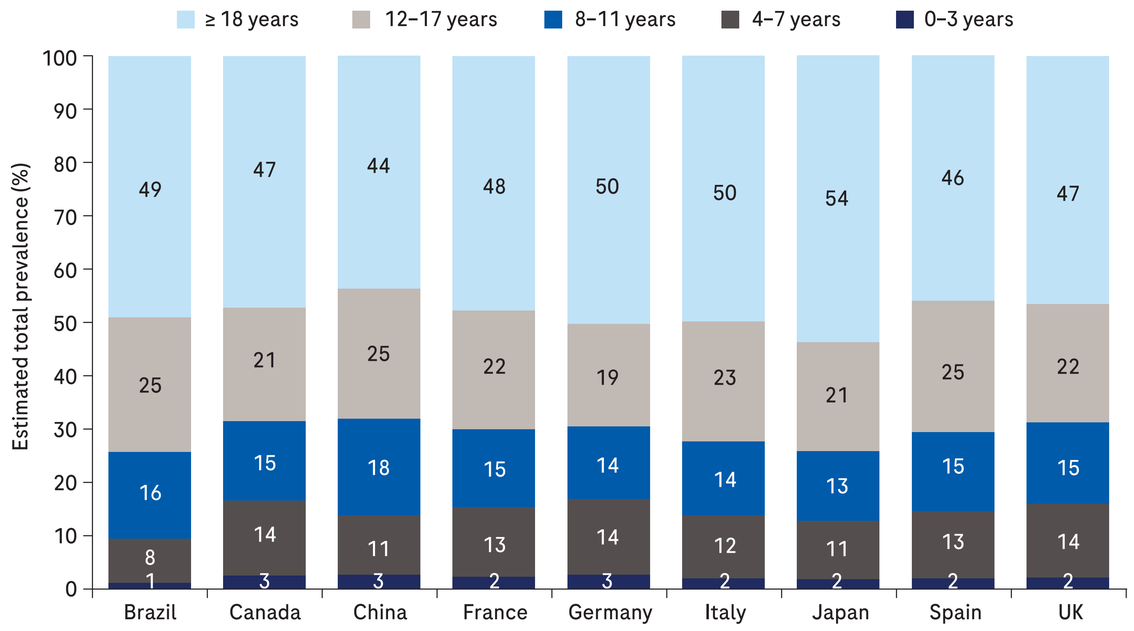
- The estimated total diagnosed prevalences of DMD for Brazil, Canada, China, France, Germany, Italy, Japan, Spain and the UK, for the overall population and by age group, for the year 2023 are presented in **Table 1**.
- Age group distribution was comparable across countries, with approximately an equal distribution of DMD prevalence between paediatric and adult populations (**Figure 1**).

Table 1. Estimated total diagnosed prevalence of DMD in nine countries, overall and by age group.

Country (male population, thousands[a])	Estimated total diagnosed prevalence of DMD, n	Estimated total diagnosed prevalence of DMD by age group				
		0–3 years, n	4–7 years, n	8–11 years, n	12–17 years, n	≥ 18 years, n
Brazil (106 241)	5249	67	437	844	1321	2580
Canada (19 275)	1003	25	141	149	215	473
China (727 173)	27 931	718	3141	4987	6872	12 212
France (31 279)	2082	47	272	306	463	994
Germany (41 109)	2071	53	296	281	402	1039
Italy (28 725)	1361	26	160	190	309	676
Japan (59 914)	3110	56	342	403	644	1665
Spain (23 292)	1169	24	147	173	290	535
UK (33 486)	2055	47	285	313	455	956

[a]Male population derived from United Nations, Department of Economic and Social Affairs, World Population Prospects 2022.
DMD, Duchenne muscular dystrophy.

Figure 1. Estimated contribution of different age groups to the total diagnosed prevalence of DMD in nine countries.

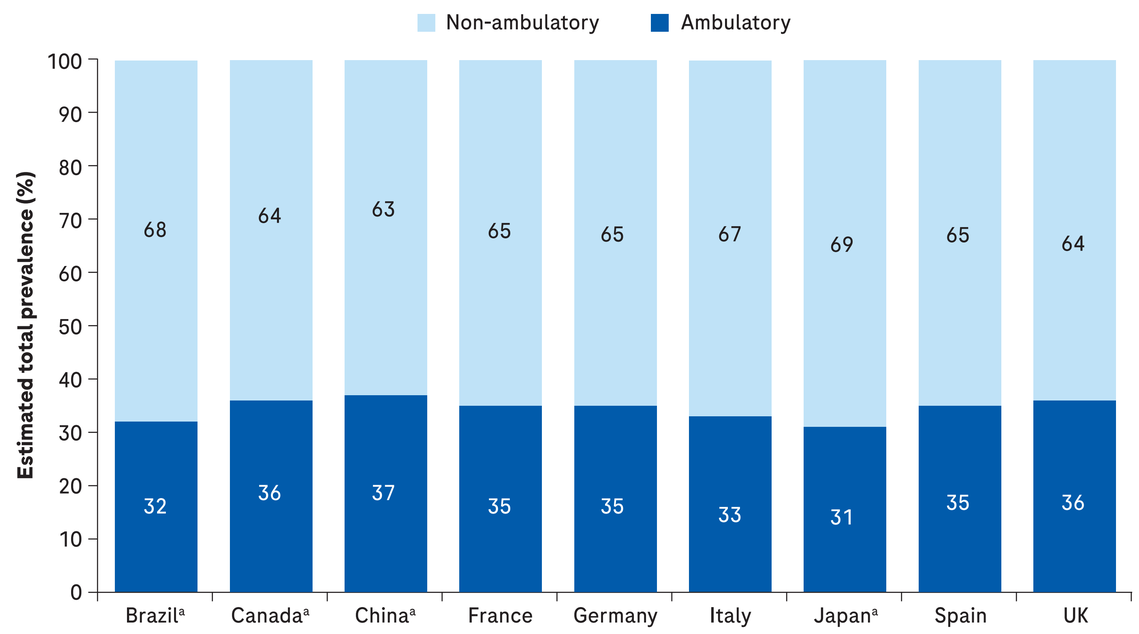


DMD, Duchenne muscular dystrophy.

Estimated ambulatory status of patients with DMD

- Of the total population of patients diagnosed with DMD, the estimated proportion of ambulatory patients ranged from 31% to 37% across countries (**Figure 2**).

Figure 2. Estimated total diagnosed prevalence of DMD in nine countries, by ambulatory status.

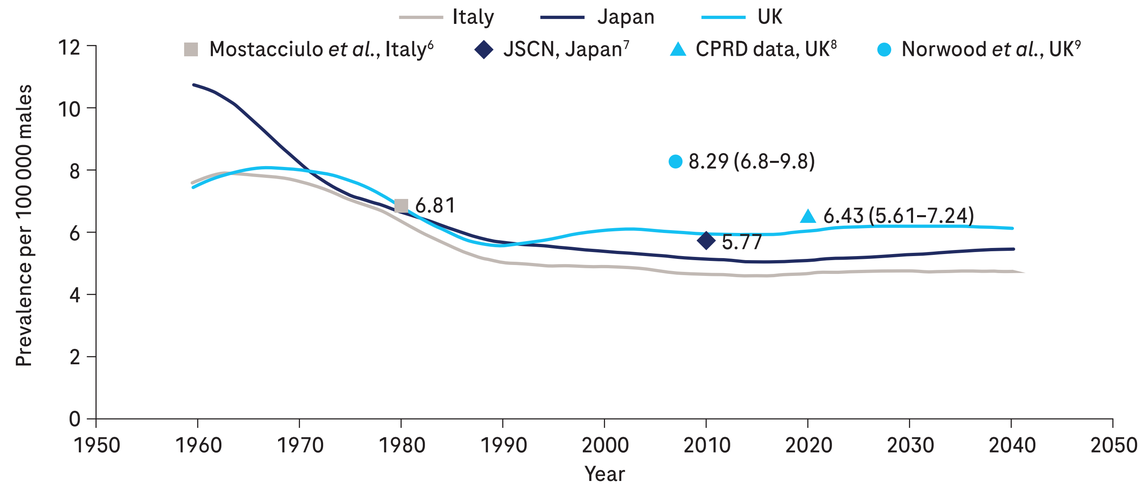


[a]Data on ambulation were derived from an international study with predominantly US centers; the applicability of these data to these countries is limited.
DMD, Duchenne muscular dystrophy.

Comparison of model-estimated prevalence with published epidemiology data

- The model-estimated overall prevalence showed good alignment with published prevalence in Italy, Japan and the UK, supporting the robustness of the methodology (**Figure 3**).

Figure 3. Comparison of estimated prevalence of DMD in Italy, Japan and the UK with literature-reported prevalence.



Lines indicate country-specific data. Markers indicate literature-reported prevalence with data (95% confidence interval), where reported.
CPRD, Clinical Practice Research Datalink; DMD, Duchenne muscular dystrophy; JSCN, Japanese Society of Child Neurology.

Limitations

- Country-specific incidences were assumed for selected countries. A trend curve considering historic trends in incidence of DMD was derived from UK studies and extrapolated to other countries.
- Although the median age at diagnosis for each country was determined from published studies, the distribution around the median for select countries was assumed.
- Model input data were based on the currently available but limited evidence in the literature. Acquiring claims and real-world data would strengthen the model.

Conclusions

- Estimates of the prevalence of DMD are required to understand disease burden in the population and to ensure that adequate resources are available to meet patient needs.
- This analysis applied incidence and mortality to provide robust prevalence estimates of DMD overall, by age group and by ambulatory status, contributing to a greater understanding of the epidemiology of DMD.
- The distribution of patients by age groups and ambulatory status was comparable across all studied countries; the adult population was a major contributor to the DMD-diagnosed prevalence.

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Author disclosures

CL is an employee of F. Hoffmann-La Roche Ltd. **AK** is an employee of ZS Associates India Pvt Ltd. **AT** is an employee of ZS Associates International Inc. **UO** is an employee of F. Hoffmann-La Roche Ltd. **MP** has nothing to declare. **VS** has been on advisory boards for Astellas Gene Therapies, Biogen, Edgewise Therapeutics, Ipsen, Kate Therapeutics, ML Bio Solutions, Novartis Gene Therapies, PepGen, Roche, Sanofi, Sarepta Therapeutics, Vertex Pharmaceuticals and Wave Therapeutics, has received speaking fees/honoraria from Novartis Gene Therapies, Pfizer, Roche, Sanofi and Sarepta Therapeutics and has research collaborations with Sarepta Therapeutics and Sanofi. **MCW** has participated in scientific advisory boards and reports receiving funding for travel or speaker honoraria or consultancies from Affinia, Argenyx, Aro, Audentes Therapeutics, AveXis Pharma, Biogen Pharma GmbH, BridgeBio, Colloquium Neurologicum, Edgewise, Fulcrum Therapeutics, GLG Consult, Grünenthal Pharma AG, Guidepoint Global, ML Bio, Neurix, Novartis Pharma GmbH, Pfizer Inc., PharNEXT, PTC Therapeutics, Roche Pharma AG, RTI Health Solutions, Santhera Pharmaceuticals, Sarepta Therapeutics Inc., Teva, Ultragenyx and Wave Sciences Technology and has received research support from Gamedis Study, GlaxoSmithKline GmbH, Grifols GmbH, Novartis Pharma GmbH, PharNEXT and Trophos AG (now Roche Pharma AG).

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