Analysis of Italian Administrative Healthcare Data to Identify Rare Diseases and Describe Their Clinical Burden: The Case of Duchenne Muscular Dystrophy

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BACKGROUND AND AIMS

Duchenne muscular dystrophy (DMD) is a rare disease that causes a progressive loss of muscle function in males, presenting at the age of two years, and involving respiratory and heart function starting from teenage years. The identification of DMD through real-world data (e.g., administrative healthcare data) is still challenging.

→ **AIMS**: To identify patients potentially affected by DMD (prevalence) and describe the demographics and clinical characteristics (i.e., comorbidities) .

METHODS

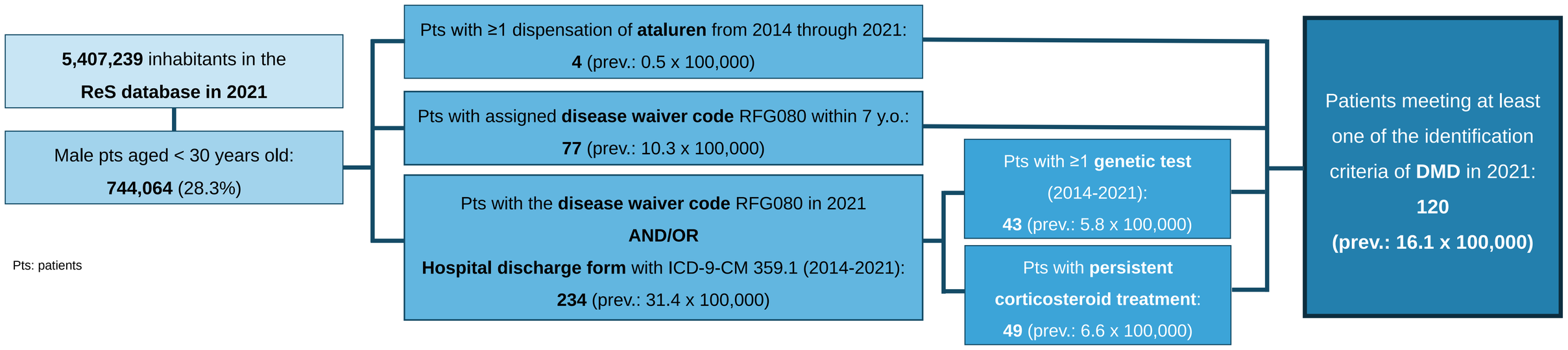
Retrospective observational and longitudinal cohort study through the **administrative healthcare database** of Fondazione Ricerca e Salute (ReS)

CRITERIA for the IDENTIFICATION of the study population

- Male patients aged < 30 years old during the accrual period (from 1st January to 31st December 2021)
  - Patients meeting at least one of the following criteria:
    - At least one reimbursed **dispensation of ataluren** (ATC code: M09AX03) during the accrual period and/or the available look-back period (max. 7 years);
    - Assignment of the **disease waiver code from co-payment** of fee related to muscular dystrophies (**RFG080**) **within 7 years of age**;
    - Presence of the **disease waiver code** RFG080 in the accrual period AND/OR **admission to hospital/emergency department** with diagnosis of hereditary progressive muscular dystrophy (ICD-9-CM code: 359.1) during the accrual period and/or the 7-year look-back period, together with:
      - A previous **genetic test** performed in local outpatient specialist settings (Italian codes);
      - AND/OR a **persistent** (i.e., ≥3 months) treatment with **corticosteroids for systemic use** (i.e., prednisolone - ATC code: H02AB06; prednisone - H02AB07; deflazacort - H02AB13) during the year of follow-up;

RESULTS

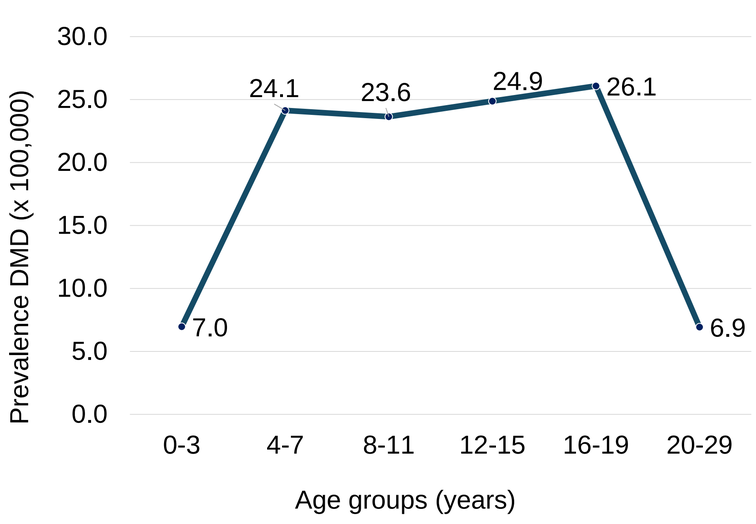
Identification of the study population



Pts: patients

Demographics

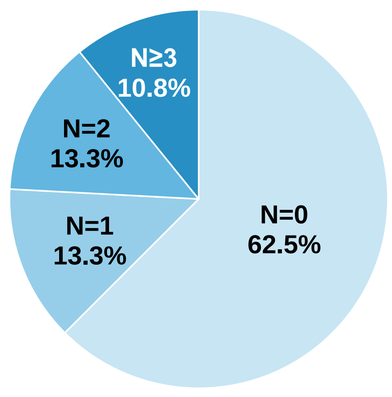
Prevalence (x 100,000 inhab. aged <30 y.o.) of DMD, by age group



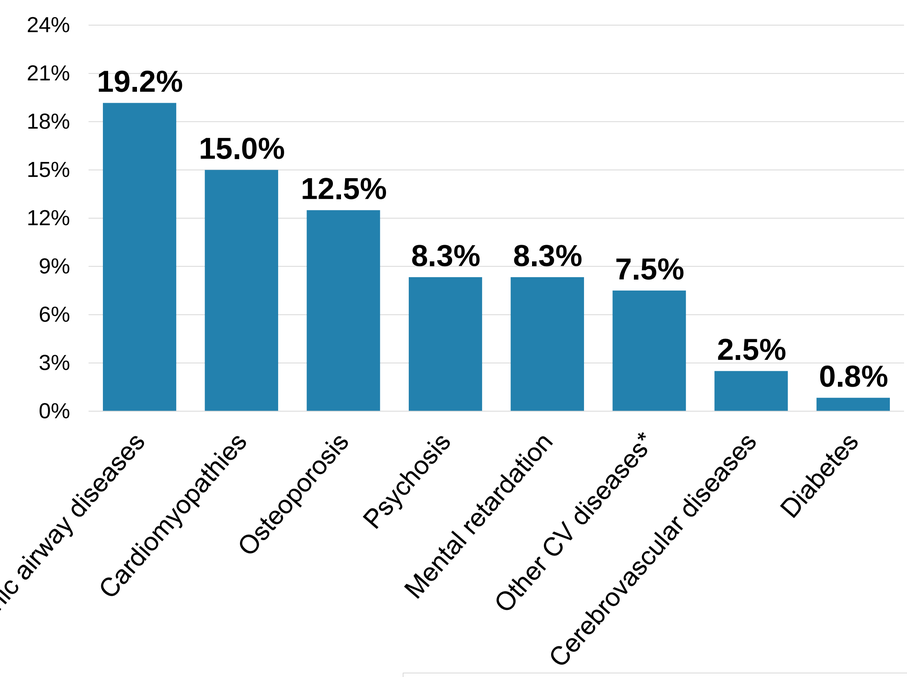
Mean age (SD)	**13 (6)** years
Median age (Q1; Q3)	**13 (9;18)** years

Comorbidities

Patient distribution (%), by N° of comorbidities



Patient distribution (%), by type of comorbidity



CV: cardiovascular
*heart failure, coronary artery disease, arrythmias

CONCLUSIONS

DMD is a rare disease. Its **epidemiology** and the **clinical burden** of patients potentially affected by DMD have been realibly identified through **real-world data**, such as administrative healthcare data, despite the need for different algorithms and proxies. These findings are useful for Italian **pre-marketing regulatory** steps, thanks to the quantification of patients elegibile to innovative therapeutics for DMD.

Scan and download the poster!

Conflict of interest: Dell'Anno I is an employee of Fondazione ReS

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