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Prevalence and Genetic Profile of Duchene and Becker Muscular Dystrophy in Puerto Rico

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Abstract

Background—Duchenne and Becker Muscular Dystrophy (DMD and BMD, respectively), are common forms of inherited muscle disease. Information regarding the epidemiology of these conditions, including genotype, is still sparse.

Objective—To establish the prevalence and genetic profile of DMD and BMD in Puerto Rico.

Methods—We collected data from medical records in all Muscular Dystrophy Association (MDA) clinics in Puerto Rico in order to estimate the prevalence of DMD and BMD and to describe the genotypic profile of these patients. Patients selected for data analysis matched “definite”, “probable” and “possible” case definitions as established by MD STARnet.

Results—A total of 141 patients matched the inclusion criteria, with 64.5% and 35.5% being categorized into DMD and BMD, respectively. DMD and BMD prevalence in Puerto Rico was estimated at 5.18 and 2.84 per 100,000 males, respectively. Deletion was the most common form of mutation (66.7%) in the dystrophin gene, with exons in segment 45 to 47 being the most frequently affected.

Conclusions—This is the first report of the prevalence and genetic profile characteristics of DMD and BMD in Puerto Rico. Prevalence of DMD was similar to that reported worldwide, while prevalence of BMD was higher. Genetic profile was consistent with that reported in the literature.

Keywords

Duchenne muscular dystrophy; Becker muscular dystrophy; dystrophinopathy; muscular dystrophy; Puerto Rico

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Conflicts of Interest

The authors have no conflict of interest to report.

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Introduction

Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD) are X-linked recessive allelic disorders caused by mutations in the dystrophin (*DMD*) gene (1). These occur almost exclusively in males and are included in the conditions commonly referred to as dystrophinopathies.

The *DMD* gene is the largest protein-coding gene in humans, localized at Xp21.2 with a size of 2.4 Mb(2–4). RNA transcribed from the dystrophin gene is expressed predominantly in skeletal and cardiac muscle, with lower expression in brain tissue (5). This gene encodes for dystrophin, a cytoskeletal protein that bridges the inner cytoskeleton and the extracellular matrix of muscle fibers(6). Due to its large size, the rate of *DMD* gene mutations is high (7). Rearrangements such as gene deletions and duplications are the most common type of aberration found in the *DMD* gene, but 20% to 35% of DMD and BMD cases result from point mutations(8,9).

A recently published review of the epidemiology of DMD and BMD by Mah et al estimated the worldwide prevalence to be 4.78 and 1.53 per 100,000 males, respectively (10). However, there was a lack of studies from Latin America in the literature review that served as the basis to calculate these estimates.

This is the first publication that reports estimates for the prevalence and genotype description of DMD and BMD patients in Puerto Rico.

Materials and Methods

This is a retrospective chart-review study encompassing data from 141 patients attending the Muscular Dystrophy Association (MDA) neuromuscular clinics in Puerto Rico (4 clinics in total). These clinics are the only facilities in Puerto Rico dedicated to the care of patients with muscular dystrophy, and the community is well aware that they are sponsored by the Muscular Dystrophy Association. Thus, we estimate that over 90% of DMD and BMD patients in Puerto Rico are treated in these four clinics. The study protocol was approved by the Institutional Review Board (IRB) of the University of Puerto Rico Medical Sciences Campus (IRB ID: 0890113), and authorized by the Muscular Dystrophy Association Office in Puerto Rico.

Inclusion criteria followed case definitions as established by the Muscular Dystrophy Surveillance Tracking and Research Network (11). “Definite”, “probable” and “possible” case definitions were included for data analysis. “Definite” cases have symptoms referable to DMD or BMD and either (1) a documented DMD gene mutation, (2) muscle biopsy evidencing abnormal dystrophin without an alternative explanation, or (3) creatine kinase level at least 10 times normal, pedigree compatible with X-linked recessive inheritance, and an affected family member who meets criterion 1 or 2. “Probable” cases have symptoms referable to DMD or BMD, a creatine kinase level at least 10 times normal, family history consistent with an X-linked muscular dystrophy, and either (1) no DMD gene mutation analysis or muscle biopsy, (2) an inconclusive muscle biopsy, or (3) a negative DMD gene mutation analysis. “Possible” cases have elevated creatine kinase and documented clinical

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symptoms referable to DMD or BMD. A neuromuscular specialist in each clinic evaluated patients and conferred clinical diagnosis of either DMD or BMD following previously published diagnosis criteria(12), if applicable.

A complete record review of all active patients in the MDA clinics from the first to the most recent visit to the clinics was performed for all patients. Data collection included age at time of data collection and age at diagnosis (defined as age at which the physician documented a DMD or BMD diagnosis). Four cases were diagnosed when they were less than 1 year old. For statistical analysis, age at diagnosis for these cases was coded as 1 year old. In addition, data from confirmatory studies was collected, including results from muscle biopsy, creatine kinase (CK), DNA studies performed by certified clinical laboratories and/or family history of DMD or BMD. Details obtained from the DNA study report included type of mutation and the affected segment of the dystrophin gene.

This data was submitted and accepted for inclusion in the Leiden Open Variation Database for DMD (7,13).

Statistical Analysis

Statistical analysis was done using SPSS version 12 on Windows XP operating system, and R version 3.2.4(14), R Commander version 2.2-3(15) and the EZR plug-in for R Commander(16) on OS X 10.11.4. Frequency distributions and Mann-Whitney tests were used for data analysis. Denominators for the estimation of disease prevalence were obtained from population estimates of the male population in for Puerto Rico for the year 2012 (1,757,189 males for 2012) available online from the United States Census Bureau (17).

Results

141 patients (all male) met the inclusion criteria for the study. Table 1 displays general characteristics of the entire sample. 91 patients (64.5%) were classified as DMD while 50 (35.5%) were classified as BMD. Data about age at diagnosis, age at the time of the study (“actual age”) and CK levels were available for 111 patients (87.7%), 140 patients (99.3%) and 111 patients (79.3%), respectively. Median age at diagnosis was 6 years old. 84 patients (59.2%) had a molecular study available for analysis, while 37 (26.1%) had a muscle biopsy result on record. Based on 2012 census data (17) , and considering that the data was collected from all four major muscular dystrophy clinics on the island, the minimum prevalence of DMD and BMD in Puerto Rico was estimated as 8.02 per 100,000 males (141 DMD/BMD males/1,757,189 total males for 2012 in Puerto Rico). The minimum prevalence of DMD and BMD in Puerto Rico is 5.18 per 100,000 males (91 DMD males/1,757,189 total males for 2012 in Puerto Rico) and 2.84 (50 BMD males/1,757,189 total males for 2012 in Puerto Rico) per 100,000 males, respectively. Table 2 details the case definitions per DMD and BMD diagnosis using the Muscular Dystrophy Surveillance Tracking and Research Network criteria established above. The majority of patients were classified as a “Definite” case.

The genetic profile of patients having results of a molecular study in the medical record is displayed in Table 3. The majority of patients with a molecular study have evidence of a

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deletion, comprising 66.7%. Figure 1 shows exon mutations over a representation of the dystrophin exon sequence. The segment that was most frequently affected was segment 45 to 47.

Age and CK profiles of DMD and BMD patients are shown in Table 4. CK distribution for DMD and BMD patients is presented in Figure 2. Differences in age at the time of the study, as well as age at diagnosis were statistically significant ($p = 0.001$ and $p = 0.002$ respectively) suggesting that BMD patients were diagnosed later in life. CK levels at the time of diagnosis also demonstrated a statistically significant difference ($p = 0.001$) with patients diagnosed with DMD having much higher levels at the time of diagnosis.

Discussion

To our knowledge, this is the first study describing prevalence and genotypic characteristics of patients with DMD and BMD in Puerto Rico. After retrospective analysis of medical record data, one hundred forty one patients met the inclusion criteria for diagnosis of these conditions. Of these, 91 patients were diagnosed with DMD, 50 were diagnosed with BMD, and 19 patients were still pending final characterization of their conditions. As stated above, the minimum prevalence for both DMD and BMD in Puerto Rico is estimated as 8.02 per 100,000 males, while estimated individual prevalence of DMD and BMD is 5.18 and 2.84 per 100,00 males, respectively. As a reminder, the values reported by Mah in 2014 for estimated worldwide prevalence of DMD and BMD were 4.78 and 1.53 per 100,000 males, respectively (10).

It is noteworthy that the estimated minimum prevalence of BMD in Puerto Rico is 8% higher than that reported as the worldwide prevalence. Since there is no cure for this condition, a higher prevalence can be explained by an increased incidence, a lower mortality, or a combination of these two factors. Longer life expectancy (lower mortality) due to appropriate management of the disease and its associated complications might hence increase prevalence. Access to adequate diagnostic facilities may also result in a higher frequency of diagnosis, with a subsequent impact on the report of new cases of the disease. The island’s small size might also allow for less genetic variation and perpetuation of mutations resulting in more new cases of the disease.

Patients diagnosed with DMD were diagnosed at an earlier age than those with BMD (5.5 years old vs 9.0 years old, respectively). This may be explained by BMD patients presenting symptoms later in life. The difference in age at the time of the study (BMD patients older than DMD patients) might be due to better survival of BMD patients.

Molecular genetic testing of *DMD* can establish the diagnosis of either DMD or BMD without muscle biopsy in most individuals. Thus, the current guidelines for diagnosis establish that deletion/duplication *DMD* gene analysis should be done first and, on those whose result was non-informative, then *DMD* gene sequencing analysis should follow. If no *DMD* pathogenic variant is identified after performing both types of genetic analysis described above, then skeletal muscle biopsy is indicated for western blot and immunohistochemistry studies of dystrophin (18).

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Approximately half (59.2%) of patients diagnosed with DMD or BMD in our sample had a molecular (DNA) study available as part of their diagnosis. Although genetic testing is an important diagnostic tool for this condition, these were not commonly used until 1990 (10). Since the average age for our study population is 20.47 years old, this could partially explain the low number of patients having a result for a diagnostic DNA study. Although not all research articles about this condition report the usage of molecular studies as a requisite for diagnosis, there are some that do. In Canada, 74% of patients had genetic testing performed for their diagnosis (19). A recent study in DMD patients in the USA cites that 94.7% of their reported population had genetic testing available for analysis (20). Genetic testing can be costly for patients, and it is not usually covered by medical insurance, which could explain the low number of DNA studies available for analysis in our clinics. As detailed above, a *DMD* gene sequencing analysis should follow if the initial *DMD* gene analysis is non-informative. Not all patients have the economic means to more testing, raising the possibility of patients not being properly diagnosed if their initial testing is unremarkable. This reveals a health disparity issue amongst our patient population, primarily regarding awareness of diagnostic guidelines and/or coverage of expenditures associated to genetic diagnostic tests in individuals who meet clinical criteria.

Of those patients who underwent a genetic test, 58 patients had deletions or duplications, whereas only 7 of the remaining 26 patients (~27%) with uninformative results on the *DMD* rearrangement analysis were able to undergo reflex testing for *DMD* sequence variations. There is a large variability on the genomic mechanisms leading to DMD or BMD. Deletions of one or more exons account for up to 60-70% of pathogenic variants, duplications account for up to 10%, and the smaller genomic sequence alterations account for approximately 25-30%(9,21–24). Large rearrangements involving multiple exons cluster in two recombination hot spots. The proximal hot spot comprises exons 2-20 (30%) and the distal hot spot comprises exons 44-53 (70%) (25). Duplications preferentially cluster at the proximal hot spot with duplication of exon 2 being the most common (26)

In contrast to the expected rates, amongst those 58 patients with large pathogenic *DMD* rearrangements identified, 56 had deletions of one or multiple exons (~96.6%), whereas duplications were only seen in 2 patients (~3.4%). Both patients with duplications had involvement of exons within the proximal hot spot but only one of them affected exon 2. As observed in other populations, 31% of our patients had pathogenic rearrangements involving the proximal cluster whereas 69% involved the distal cluster.

Limitations in this study are mostly linked to the nature of retrospective data collection in a healthcare facility. Data was obtained from four different clinics that had not implemented electronic medical record systems. Multiple physicians evaluated patients throughout the time period analyzed. In addition, there was no standardized form to record results from the evaluation of patients with DMD or BMD, which made data collection more difficult. Our sample size is small when compared to other studies; however, this is to be expected based on the population size of Puerto Rico. We believe that collecting data from all four MDA clinics on the island allowed us to collect data from most patients suffering from these conditions in the island.

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Conclusion

This is the first report of the prevalence and genetic profile characteristics of DMD and BMD in Puerto Rico. The prevalence of these conditions in our population is similar to that reported worldwide, with a slightly higher prevalence of BMD patients. In addition, genetic characteristics were very similar to those reported in other parts of the world. There is a notable lack of genetic test data available in the population under study. This study also demonstrates the need for a standardized instrument to be used by clinicians following patients suffering from DMD and BM in Puerto Rico. The development of an island-wide registry for this population could be of benefit for future analysis, eliminating most of the limitations in the current study.

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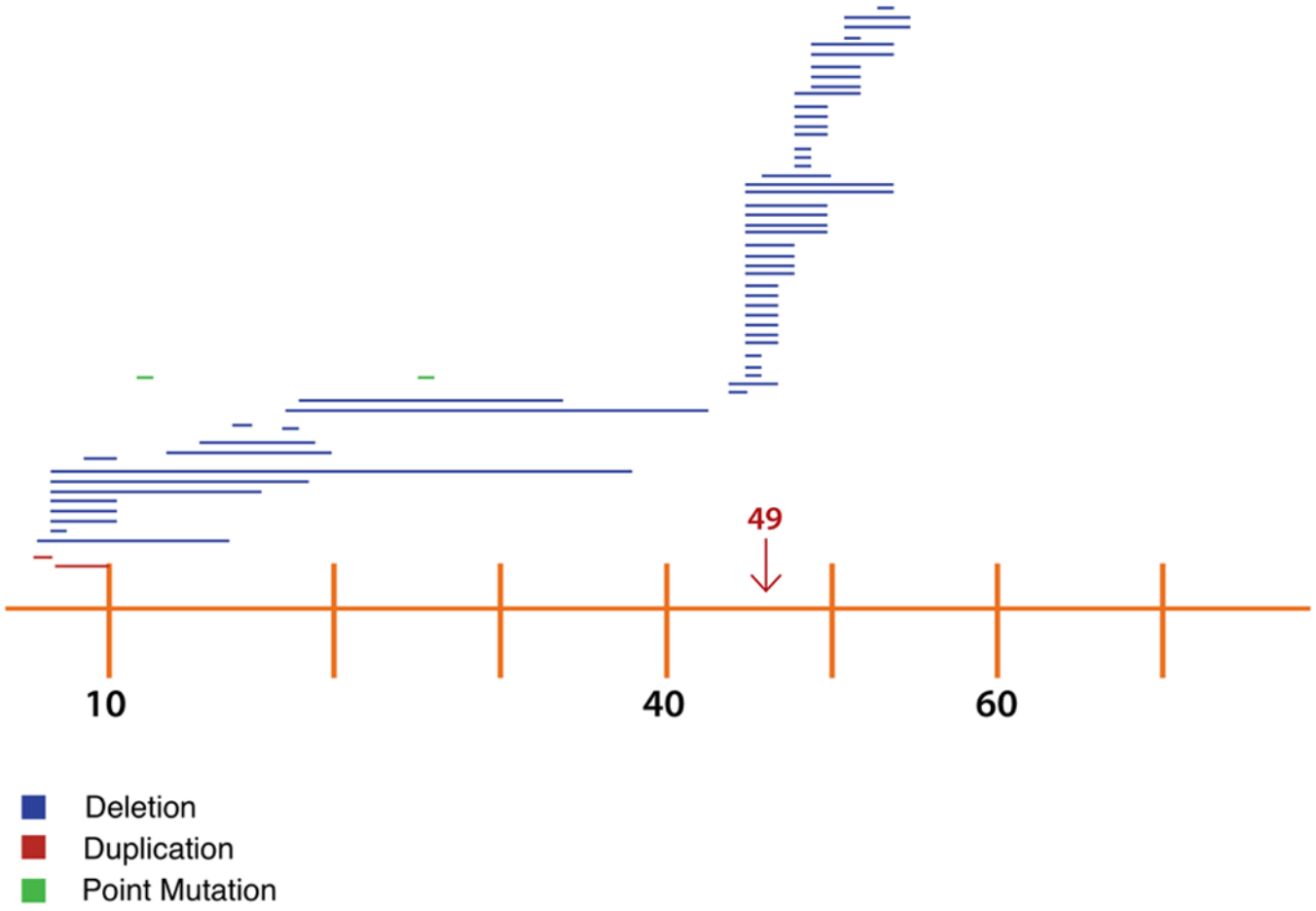

Figure 1.

Distribution of mutations along the DMD gene.

1 unit on the X axis represents 1 exon on the DMD gene, increasing sequentially from left to right.

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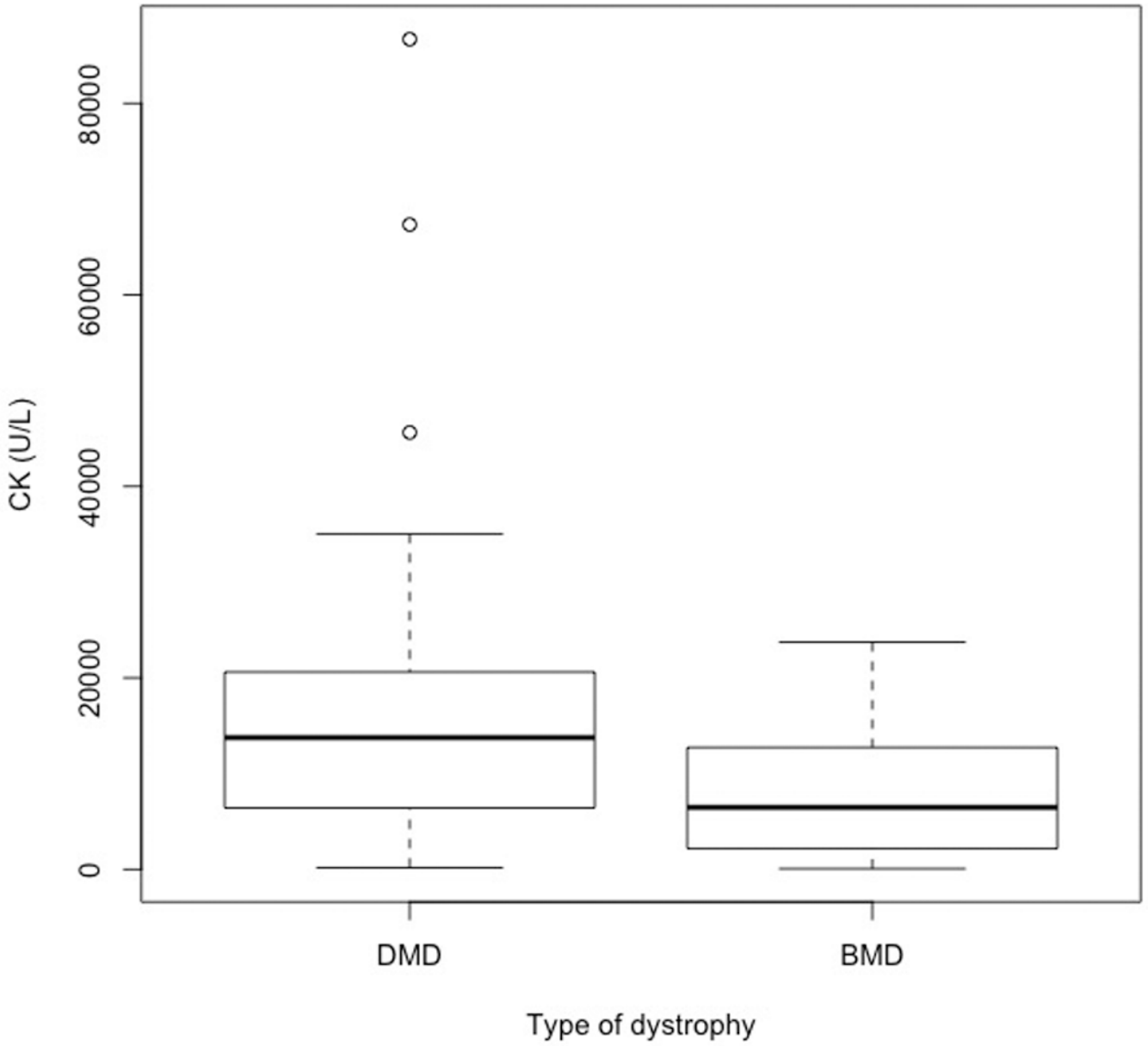

Figure 2.
Creatine kinase levels at time of diagnosis by type of muscular dystrophy

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Table 1

General and clinical characteristics of patients with DMD/BMD. Muscular Dystrophy Association Clinics, Puerto Rico. 2012.

Variable	**Median (range), or number (percent)**
Total cases	141
Actual age (median) n = 140	19 (4 to 72)
Age of diagnosis (median) n = 111	6 (1 to 59)
CK at diagnosis, units per liter (median) n = 102	11250 (63 to 86700)
Number of patients with Duchenne Muscular Dystrophy	91 (64.5%)
Number of patients with Becker Muscular Dystrophy	50 (35.5%)
Positive DNA study	84 (59.2%)
Positive muscle biopsy	37 (26.1%)
Positive family history	49 (34.5%)

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Table 2

Distribution by Case Definitions for DMD and BMD patients based on Muscular Dystrophy Surveillance Tracking and Research Network criteria.

	DMD Number (percent)	BMD Number (percent)
Definite	76 (83.5)	35 (70.0)
Probable	6 (6.6)	5 (10.0)
Possible	9 (9.9)	10 (20.0)
Total	91 (100.0)	50 (100.0)

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Table 3

Gene mutations in patients with DMD/BMD. Muscular Dystrophy Association Clinics, Puerto Rico. 2012 (n = 141)

Type of Mutation	Number (percent)
Deletion	56 (66.7%)
Duplication	2 (2.4%)
Insertion	1 (1.2%)
Other	6 (7.1%)
No mutation	19 (22.6%)

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Table 4

Age and CK profiles of DMD and BMD patients. In parenthesis is the number of patients from either DMD or BMD with available data for each variable.

	DMD	BMD	p-value (Mann-Whitney test)
Age at time of study median (n)	18.0 (90)	20.0 (50)	0.001
Age at Diagnosis median (n)	5.5 (74)	9.0 (37)	0.002
CK at Diagnosis (U/L) median (n)	13759.5 (68)	6476 (34)	0.001