Decode Duchenne:

Impact and Trends in a Sponsored Genetic Testing Program

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Background

The Decode Duchenne (DD) industry-sponsored genetic testing program provides testing for individuals with a suspected or confirmed diagnosis of Duchenne or Becker muscular dystrophy (DMD/BMD). DD began with diagnostic testing in 2013 and added carrier testing in 2018. DD is administered by genetic counselors at Parent Project Muscular Dystrophy. Testing was initially performed at Emory/EGL and the transferred to PerkinElmer Genomics in 2018.

Methodology

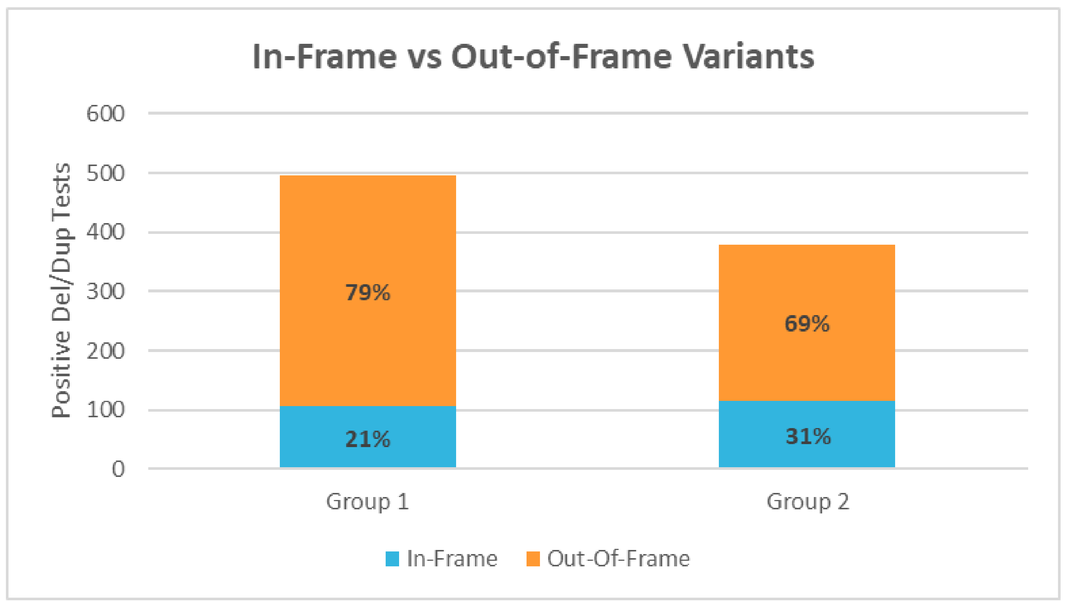
We analyzed DD data including patient demographics, ordering provider (OP), test type and results. We compared data collected during the initial 4.5 years of the program (**Group 1**: December 2013-May 2018) with data collected in the most recent 4.5 years (**Group 2**: June 2018-November 2022). Both datasets cover 53 months.

Results

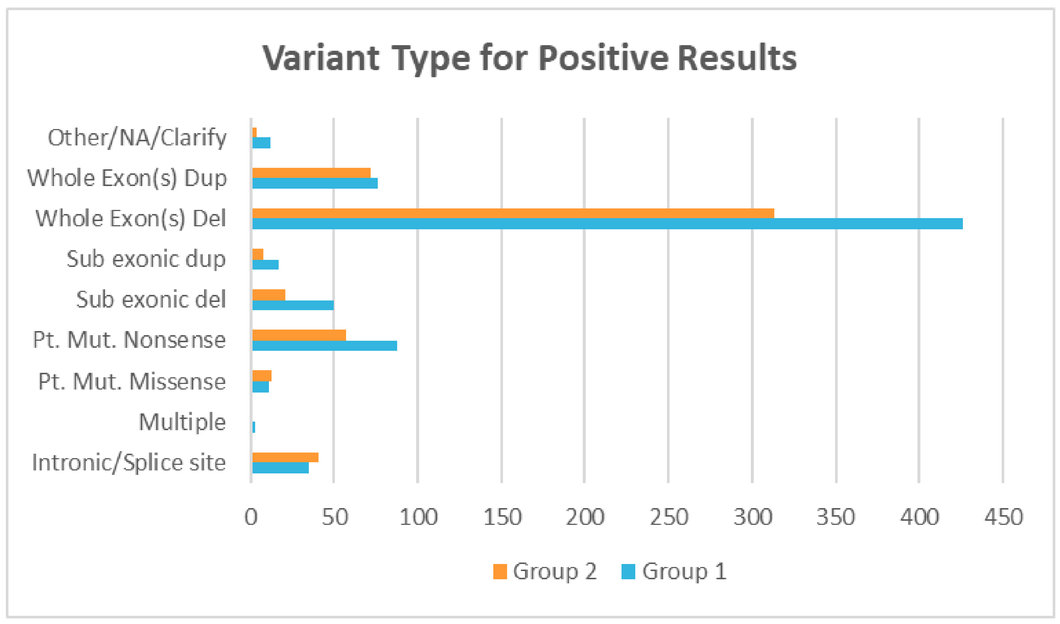
A total of 2,439 tests have been performed through DD, including 1,534 diagnostic and 905 carrier tests. All analyses on this poster reflect diagnostic testing only. The age at diagnostic testing has shown a downward trend in recent years, with an average age of 15 years in Group 1 versus 13.9 years in Group 2, and a median age of 11.4 years in Group 1 versus 8.2 years in Group 2.

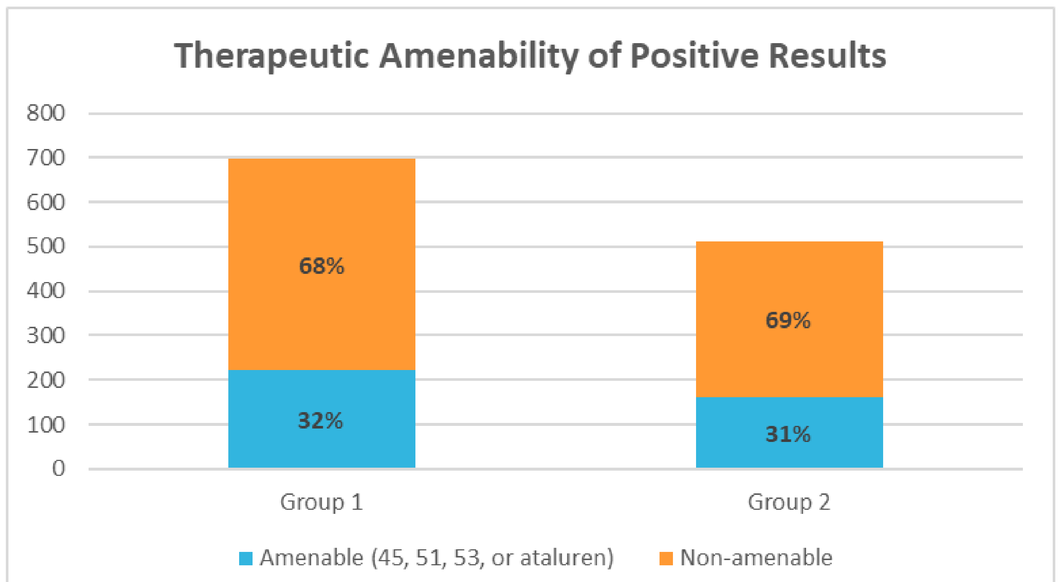
	Time Period	Average Age	Median Age
Group 1	Dec '13 – May '18	15.1	11.4
Group 2	Jun '18 – Nov '22	13.9	8.2

The type of OP has remained consistent, with most tests ordered by specialist physicians. The type of testing has shifted almost entirely to *DMD* sequencing using NGS, which detects both sequence and copy number variants, versus the two-step testing approach used in initial years. Detection of out-of-frame (OOF) vs in-frame (IF) variants has shifted from 79% OOF for Group 1 to 69% for Group 2. The average age of OOF variants in Group 1 was 15 years versus 12.5 years in Group 2.



The variant type and the percent amenable to approved therapeutics have remained consistent in both datasets. Approved therapeutics include exon skipping for exons 45, 51 and 53, and nonsense mutation readthrough (ataluren). A decrease in diagnostic tests was observed in recent years.





Discussion

With the increase in variant-specific therapeutic approaches in clinical trials and in approved therapeutics, DD is an extremely valuable program in the DMD/BMD community despite a decrease in testing in recent years (likely due to the pandemic and the availability of other sponsored testing programs). DD has adapted to the needs of the community and now includes carrier testing, neuromuscular panel testing, and RNA sequencing. RNA sequencing is important to facilitate classification of variants of uncertain significance and testing for individuals with no molecular diagnosis.

Acknowledgements

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