

Duchenne and Becker Muscular Dystrophy: Parental Knowledge and Perspectives on Genetic Counseling

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Duchenne and Becker muscular dystrophy are debilitating X-linked recessive neuromuscular disorders resulting from mutations in the *DMD* gene on Xp21.2. Both disorders are considered to be a spectrum of disease referred to as Duchenne/Becker muscular dystrophy (DBMD). Current treatments and therapies have substantially improved muscle strength, and prolonged mobility and lifespan for individuals with DBMD, but there is no curative treatment and the eventual outcome is usually fatal. Genetic counseling is an important communication process between the patient, their family, and a healthcare professional. Genetic counseling also serves as an essential source of support for DBMD families. There is little literature that explores the process of genetic counseling for DBMD families, and assesses parents' knowledge of their child's condition and their feelings regarding the genetic counseling process. Accordingly, a questionnaire was developed to assess parents' knowledge of their child's condition, determine how, where, and when this information was presented to them, and investigate their feelings regarding the genetic counseling process and how this process could be improved. Questionnaires were completed by 128 respondents. The majority of parents of children with DBMD were relatively knowledgeable regarding the natural history and progression, etiology, inheritance, and potential carrier health risks; however, several misconceptions were identified regarding recurrence risks for known carriers. The highest percentage of parents reported receiving genetic counseling from genetic counselors. Parents who reported receiving genetic counseling had significant higher knowledge scores than parents who did not have genetic counseling ($P < 0.047$). There is no significant difference in knowledge among parents who reported being counseled by a genetic counselor, geneticist, or neurologist, although parents receiving counseling from a genetic counselor reported an overall higher level of satisfaction. Our findings indicate that while genetic counseling may be successful in educating parents about DBMD, parents continue to make recommendations to improve content and presentation of the information provided during genetic counseling.