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Needs of caregivers/families affected by Duchenne and Becker Muscular Dystrophy (DBMD): Data from the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARNet)
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Systematically collected information regarding the needs of families of individuals with DBMD is not available.

To develop a profile of needs of caregivers/families of individuals with DBMD.

MD STARNet is a multistate collaboration that conducts population-based surveillance for individuals with DBMD and interviews and surveys with their caregivers. A needs assessment survey of caregivers of individuals with DBMD was conducted between September 2008 and August 2012. The survey included questions about sociodemographic profile of respondents, clinical profile of the individual with DBMD, needs related to home and community, equipment, healthcare services, access to information, psychosocial needs of the individual with DBMD and psychosocial needs of caregiver/family. Two open ended questions asked about the 3 most important met and unmet needs. Response options were coded as needed/not needed and if needed, managed/not managed.

Responses were received from 274 caregivers (70%) of those contacted. The majority of respondents were mothers, white, non-Hispanic, with at least some college education and annual income >$30,000 per year. The age of individuals with DBMD was 14.8 ± 6.9 years, 56% of them were non-ambulatory and 29% required respiratory support. The majority of healthcare services and information related needs were reported as met. Conversely, psychosocial and financial needs were largely unmet, specifically, balancing work/family/caregiving, time and energy for relationships, funding for home and vehicle modifications.

The survey results document a broad range of unmet needs in the areas of psychosocial support and financial resources. These findings help identify gaps in care and services needed and will help guide development of interventions to facilitate optimal health outcomes and quality of life for individuals with DBMD and their families.

http://dx.doi:10.1016/j.nmd.2013.06.487

P.7.7
Health care services received by patients with Duchenne/Becker muscular dystrophy (DBMD): Data from the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARNet)
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Anecdotal reports from families and care providers suggest a wide variation in services received by patients with DBMD.

To document the type of health care services received by patients with DBMD.

MD STARNet is a multistate collaboration that conducts population-based surveillance for individuals with DBMD and interviews and surveys with their care givers. Telephone interviews were conducted between April 2007 and March 2012. In addition to demographic and clinical information, the interview included questions about the type of health care provider seen [primary care, neuromuscular, orthopedist, pulmonologist, cardiologist, physical therapist (PT), occupational therapist (OT), speech therapist (ST), and other] and type of services received [respiratory, dietary, mental health, social work, pastoral, pain management and case management] over the previous 12 months. Frequencies among states were compared with Chi square tests. Statistical significance was set at p < 0.05.

Two hundred and ninety-six care givers (66%) of those contactable, completed the interview. Care givers (%) reported seeing these providers: primary care (88%), neuromuscular (87%), cardiologist (71%), pulmonologist (54%), orthopedist (33%), PT (69%), OT (42%), ST (20%), other (20%). Services received included case management (42%), respiratory (27%), social work (25%), dietary (22%), pastoral (16%), mental health (15%) and pain management (5%). There were significant differences among states in regards to the primary care, cardiac, pulmonary, orthopedic, physical and occupational therapy providers seen and dietary and case management services received.

This study supports and provides a quantitative assessment relative to the anecdotal reports of variation in health care services received by patients with DBMD. Whether these differences affect outcomes needs to be determined.

http://dx.doi:10.1016/j.nmd.2013.06.488

P.7.8
The national Dutch dystrophinopathy patient registry

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Dystrophinopathy patient registries are very useful for improvement of standard care and planning of clinical trials. The Dutch Dystrophinopathy Database contains information of Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) patients in the Netherlands. The historical incidence of DMD is estimated at 20 boys per year and unknown for BMD.

DMD and BMD patients received information through patient organisations, treating physicians, the genetic test centre or a website (www.lumc.nl/duchenne). Included patients gave consent for registration in the national patient registry and the international TREAT-NMD database, permission to contact their physician and filled out a questionnaire about their disease course.

On January 2013 the database contained disease milestones of 420 DMD and 140 BMD patients, including data from 78 deceased DMD and 16 deceased BMD males. 324 DMD and 104 BMD patients have a DNA confirmed diagnosis. The mean yearly incidence for DMD was 18 between 1980 and 2000. Given a mean life expectancy of 27 years this would suggest a prevalence of 486 DMD patients, and thus an inclusion rate in the database of 70%. The mean yearly incidence for BMD was 4 between 1960 and 2000. The mean life expectancy was 55 years suggesting a prevalence of 220 BMD patients, and thus an inclusion rate in the database of 56%.

In the DNA confirmed DMD cohort mean age at diagnosis decreased from 5.6 years before 1970 to 4.0 years more recently. The mean age at loss of ambulation increased to 9.8 years and for scoliosis surgery to
14 years. Age at start of mechanical ventilation ranged from 14 to 28 years.

Close collaboration during five years between patient organisations, physicians and researchers resulted in a patient registry covering three quarters of all Dutch DMD patients. The yearly update is a constant challenge, but the advantage is one national registry representative for the whole population, limiting selection bias to a minimum.

http://dx.doi.org/10.1016/j.nmd.2013.06.489

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Current status of dystrophinopathy patient registry in Japan: Remudy
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We report current status of a national patient registry in Japan: Remudy. Registry of Muscular Dystrophy. Clinical trials for new therapeutic strategies are currently being planned for Duchenne and Becker muscular dystrophies (DMD/BMD); however, many challenges exist in the planning and conduction of a clinical trial for rare diseases. The epidemiological data, the total number of patients, natural history, and clinical outcome measures are mostly unclear. An adequate number of patients is needed to achieve significant results in clinical trials. As solutions to these problems, a patient registry, especially of rare diseases such as DMD/BMD, is an important infrastructure worldwide. Both in and out of Europe, TREAT-NMD, a clinical research network for neuromuscular disorders, has developed a global database for dystrophinopathy patients. We have developed a national registry of Japanese DMD/ BMD patients in collaboration with TREAT-NMD. The database includes clinical and molecular genetic data as well as all items required for the TREAT-NMD global patient registry. As of March 2013, 1078 patients were registered in this database. The purpose of this registry is the effective recruitment of eligible patients for clinical trials, and it may also provide timely information to registants about upcoming trials. This registry data also provides more detailed knowledge about natural history, epidemiology, and clinical care. In recent years, drug development has become dramatically globalized, and global clinical trials (GCTs) are being conducted in Japan as well. It is appropriate, particularly with regard to orphan diseases, to include Japanese patients in GCTs to increase evidence for evaluation, because it would be difficult to conduct such large-scale trials solely within one country. GCTs enable the synchronization of clinical drug development in Japan with those in other countries, minimizing drug approval delays.

http://dx.doi.org/10.1016/j.nmd.2013.06.490

P.7.10

Territory wide study of patients with dystrophinopathy in Hong Kong
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This is a first territory wide study in Hong Kong on Chinese patients with dystrophinopathy on their genetic mutation, motor performance, use of steroid, and the interventions they received.

This study is participated by all the paediatric departments in the eleven hospitals in Hong Kong. Clinical data was systemically collected from medical records of all patients diagnosed dystrophinopathy and were actively followed-up between May 2006 and April 2010.

We found 91 individuals with dystrophinopathy; 82% have Duchenne Muscular Dystrophy (DMD) and 18% have Becker Muscular Dystrophy (BMD). The overall prevalence of dystrophinopathy, for 2010, is 1.03 per 10,000 males aged 0–24 years old. Mutation consists of 47% of large deletion and 10% of large duplication, and 43% of small rearrangement or point mutation, of which 59.4% are nonsense mutations. Exon deletion in the distal hot spot (exons 45–55) represent up to 54% of the identified deletion, while deletion starting in the proximal hot spot (exon 2–20) account for 37%. Only 23% of children had been on steroid and all stopped before or when they lost ambulation. For DMD individuals, the age ranges from 0.5 to 34 years old, with 48% are older than 13 years old, and the mean age of loss of ambulation is 10.5 years old. For those older than 13 years old, 30% have cardiomyopathy, 19% required non-invasive ventilation, 15% had scoliosis surgery and only 5% had gastrostomy. This first territory wide study for individuals with dystrophinopathy in Hong Kong confirms a similar prevalence of such condition with the western countries, but our population has a much higher percentage of point mutation. The findings of infrequent steroid use and the low percentage of gastrostomy among patients with non-invasive ventilation, allows us to compare our current approaches in Hong Kong with the DMD care guidelines to drive improvements in our health care delivery.

http://dx.doi.org/10.1016/j.nmd.2013.06.491

P.7.11

The clinic services resource survey on Duchenne Connect
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DuchenneConnect (DC) is a self-report registry for Duchenne/Becker muscular dystrophy (DBMD). We present the Clinic Services Resource (Clinic Survey), a new addition to the registry in 2011. The goals of the survey include (1) providing families a resource that helps them understand the services offered by clinics caring for DBMD patients, and (2) providing clinics with data they can use to improve their clinical care and trial readiness. Anonymous results are publically available. The Clinic Survey includes data on clinical care in the US, reported by patients/families and clinic representatives. Survey participants are recruited through DC and Parent Project Muscular Dystrophy; participants do not have to be registered on DC. The survey includes ~50 questions covering access, satisfaction, medical services, steroid use, movement, cardiac, pulmonary, support and learning/behavior. Questions are based on the care guidelines. Of registrants living in the US, 12% (207) have completed the survey. An additional 44 non-registrants have participated, for a total of 251 completed surveys as of March 2013. More than 60 clinical sites are represented, and the majority are specialty clinics. 20% of patients report never using steroids. Reasons for not taking steroids are explored. 4% report never having had an echocardiogram and 23% report never having a