Iowa Neuromuscular & Related Disease Program

Contract 5880MD01 Five Year Report

July 1, 2015 through June 30, 2016

Congenital & Inherited Disorders

Division of Health Promotion & Chronic Disease Prevention
Phone: 1-800-383-3826
www.idph.state.ia.us/genetics/default.asp

Iowa Neuromuscular & Related Disease Program:

a contract established by IDPH in the Division of Health Promotion & Chronic Disease Prevention in mid 1980's by Iowa Administrative Code, Chapter 4:641-4.6 (80 GA, HF2362)

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What is the Iowa Neuromuscular and Related Disease Program?

Iowa Administrative Code, 641-4.6 (80 GA, HF2362).

- **Ensures access to comprehensive health care services** for children, adults, and families with a wide variety of neuromuscular and related disorders throughout the state of Iowa.
- **Addresses complex and difficult needs of one of Iowa's most vulnerable populations**.
- Mandated to provide:
  - Specialized and efficient diagnostic evaluations
  - Care coordination of neuromuscular healthcare concerns
  - Patient and family education
  - Supportive patient and family services for psychosocial concerns
  - Physical therapy evaluation and recommendations
  - Access to research opportunities
- Provide services which promote optimal medical outcome and quality of life, as these disorders affect all aspects of life for the individuals and their families
- Provide education and information about neuromuscular and related disorders to families, health care providers, educators and other interested individuals

What Does The Appropriation Support

- To achieve the mandate of the outline above, the majority of the program requires alternative funding sources (e.g. University of Iowa Hospitals and Clinics, non-profit foundation grants, patient billing).
- The Iowa Neuromuscular program funds .63% of a care coordinator

What are Neuromuscular Disorders and Who Is Affected?

Neuromuscular disorders:

- Affect individuals of all socioeconomic backgrounds and ethnicity across Iowa.
- Are chronic disorders often with onset of symptoms in childhood.
- Have symptoms (muscle weakness, breathing problems, heart failure, etc) that progress throughout childhood and adult life.
- Usually result in life-long physical disability, such as loss of walking or use of arms.
- Rarely have a cure and many forms are life-shortening.
- Usually result from gene changes, which can be inherited or "run in families" or occur as a new genetic change.
- Many NM symptoms and individuals qualities of life can be improved with appropriate medical monitoring, early intervention, supportive care in the home and community.

- Some diseases now have specific treatments that can dramatically improve outcomes, including extension of life.

There are many forms of neuromuscular disease, including:

- Disorders of the muscle (myopathies)
  - Examples include congenital myopathy, myotonic disorders and muscular dystrophies.
  - Muscular dystrophy refers to a group of inherited disorders marked by progressive weakness and degeneration of muscle tissue.
- Disorders of the nerve (neuropathies)
  - The types of neuropathy seen in this program are typically inherited or genetic disorders of the nerve or nerve sheath.
- Other Neuromuscular disorders, such as spinal muscular atrophy (SMA), myasthenia gravis and more.

~Want more information? See Appendix A for further descriptions and links to disorder-specific family organizations

- Current prevalence:
  - Worldwide surveys estimate the prevalence of disabling neuromuscular disease around 1 in 3000 to 3500.
    - Iowa's estimated population for 2012 is 3,074,186.
    - This would mean around approximately 1,025 Iowans are affected.
  - MDSTARnet, the Muscular Dystrophy Surveillance Tracking and Research Network, is a program currently active in four states including Iowa to identify ALL patients in the state with any form of muscular dystrophy. This effort will provide current prevalence numbers for the US.

Services Provided by the Iowa Neuromuscular and Related Disease Program

Iowa Neuromuscular and Related Disease Program (INMP) services are provided with a multi-disciplinary care approach. The INMP team members work together in clinic appointments, as well as between appointments. INMP team members include:

Physicians: Katherine Mathews, MD (not paid for by INMP budget) and Seth J. Perlman, MD, (not paid for by INMP budget)

- Dr. Mathews and Dr. Perlman are the only Pediatric Neurologists in Iowa with advanced training in neuromuscular disorders
- Dr. Perlman is the only physician in Iowa with expertise in pediatric electrophysiology studies.
- Dr. Mathews has additional expertise in genetics
- They serve as consultants to local healthcare providers across Iowa, including primary care providers as well as physicians in other sub-specialties.
- Details of physician activities are described in the INMP services.

Nursing/Care Coordination: Christina Trout, RN, MSN, Linda Boehmer, RN, BSN

- The daily functions of the nursing personnel are described in the INMP activities (Care Coordination, Patient/Family/Community Education and Patient/Family Advocacy).

Physical Therapy: Shelley Mockler, PT and Katie Laubscher, PT (not paid for by INMP budget)

- INMP PTs have expertise in pediatrics, wheelchair & postural support, durable medical equipment, and disorder-specific activity recommendations.
- Examples of PT goals: overcome barriers to physical disability, prevent deformity, maintain maximal level of conditioning for the disorder and improve comfort.
- Recommendations from INMP therapists with expertise in NM disorders are provided to patients, families, schools or community-based PT.
- It is not cost-effective for a therapist to attend the clinics based on billing, however this service has thus far been provided by the Center for Development and Disability.
- Documenting effect of new therapies through several motor function tests.

Food and Nutrition Services: Stephanie Borst, MS, RDN, CSP, LD (not paid for by INMP budget)

- A registered dietitian tailors the medical nutrition therapy provided to neuromuscular patients based on their needs and diagnosis
- Addresses and helps manage nutrition side effects of medications
- Guides and encourages families to consume a diet that will to help maintain or achieve a healthy weight
- Provide medical nutrition therapy for modified diet consistencies when needed due to chewing or swallowing difficulty
- Manages a patients nutrition support for those who are dependent on enteral nutrition

Social Services: Jim Porter, MSW (not paid for by INMP budget)

- The socioeconomic impact of neuromuscular disease can be devastating to families.
- Social service consultation is available at each INMP appointment.
- The primary role for social services in the NM clinics is to guide families to state, federal and community resources, such as Medicaid, Disability and local home health services.
- The social worker & nurses make referrals to community healthcare services, based on the recommendations of the NM team (visiting nurses, respite, hospice, etc)

INMP Services: Evaluation, Healthcare Management & Support

1. Diagnostic Evaluation

a. Detailed neuromuscular examinations
b. Review of family history and medical records
c. Diagnostic testing: biochemical, genetic, electrophysiology, neuro pathological

2. ***Medical Management of Neuromuscular Healthcare Concerns***

a. Medical monitoring to identify complications for early intervention or corrective treatment. Common monitoring:

-EKG, echoes for decreased heart function leading to heart failure

-PFTs for poor pulmonary function leading to respiratory insufficiency

-Evaluation of joint contractures which need orthopedic intervention

b. Referrals to other specialists, as needed (cardiology, pulmonary, orthopedics, GI, endocrinology)

c. Communication with medical home (primary care, home care, school, etc) regarding changes in health and healthcare recommendations by letter and/or phone.

d. Education of the patient and family regarding benefits & limitations of treatment options.

e. Discussions & guidance with difficult decisions, such as surgeries, respiratory ventilation, nutrition, pain control, psychosocial difficulties and advance directives towards the end of life.

f. Provide new disease specific therapies.

3. ***Physical Therapy Management of Neuromuscular Healthcare Concerns***

a. Physical therapy: Instruction for home therapy programs, consultation with local therapists and orthotists

b. Recommendations for bracing, orthotics & equipment (wheelchairs, scooters, lifts) to maintain function and independence in the home, school, work or community.

c. Provide supporting documentation for therapy and durable medical equipment, as required by payers

d. Recommendations on management of ADL's (transfers, mobility, home adaptations, vehicle modifications, etc) and assistance with identifying funding sources.

e. Monitoring motor function during new treatments

4. ***Care Coordination of Neuromuscular Healthcare Concerns***

a. Care coordination is a critical service for patients with complex and rare disorders that also involve many social concerns, including financial barriers to care. The care of these patients is time and labor intensive in the clinic setting, during hospitalizations and in daily life in the community.

b. Designated nurses & a social worker in the INMP coordinate services within the INMP and across the other medical and social disciplines as well as the medical home.

c. Care coordination is available to patients, families, providers and the community by phone, email, fax or mail.

d. The INMP was promoted as a model of care for neuromuscular patients at the PPMD annual meeting, as the INMP is population based and the role of care coordination is supported by public health appropriations.

e. The approach to care coordination at the time of referral and diagnosis was described in the following publication: Poysky J, Kinnett K. Facilitating family adjustment to a diagnosis of Duchenne muscular dystrophy. Neuromuscular Disorders (2009).

5. ***Patient and Family Education***
   a. Written and verbal information specific to disease process, treatment & management
   b. Genetic counseling (inheritance information & genetic risk assessment) to individuals or couples for family planning and prenatal options
   c. Anticipatory guidance regarding prognosis and level of disability
   - Information on prevention & early intervention for comorbid risks
   - Education of school personnel, employers, childcare providers and others
   - Updates on research for patients, families and healthcare providers
   - Assistance in planning for transitions from adolescents to adult life
   - Information about advance directives & living wills, as appropriate

6. ***Coordinated Care, Advocacy & Support Services***
   a. Phone & email triage and direct assistance with daily management of physical, emotional and social aspects of the disorder.
   b. Advocacy in communicating with educators, employers, health insurers and more.
   Examples:
   - letters of medical necessity for insurance coverage equipment
   - FMLA documentation completed to care for children with NM disorder
   - calls to educators to explain healthcare needs while at school
   - complete prior approval authorization forms for diagnostic testing
   - Guide employers in workplace accommodations for persons with disabilities

   c. Referrals to the Muscular Dystrophy Association & other Regional & National disease specific organizations
   d. Assistance in identifying community, state & federal social & financial services
   e. Referrals to home health care, respite and hospice agencies
   f. Maintain communication with home care and hospice agencies
   g. Referrals to patient and family support groups
   h. Access to research opportunities, as desired

IDPH Contract Description of Work and Services

Contract service description:

- Provide a clearly delineated package of services for individuals and families with neuromuscular conditions
- Participate in the activities of the Center for Congenital and Inherited Disorders Advisory Committee for the purposes of providing assistance and technical support to IDPH in the implementation of the Rules and Regulations.
- Coordinate and integrate services with other programs serving similar purposes and populations i.e. CHSC community based clinics, Early ACCESS

Meeting Contract Objectives

INMP Services

- Program services are fully described at the beginning of this report.

Number of Clinics & Patients

- Since FY 1995-1996, the total number of patient visits has increased by **289%**.
- The contractual agreement between the IDPH and the NM Program states that the NM Program must provide at least 15 clinics per year. This number has been met with 15 outreach clinics and 41 UIHC clinics in FY14

Clinic Locations

- Davenport, Des Moines, Dubuque, Mason City, Sioux City, Waterloo, and Iowa City
    - Clinic sites have been determined by geographic location, population density and the availability of rental space. See Iowa county map for locations and patients per county.
    - The community clinics are highly valued by the families served because it is difficult for them to travel any distance.

Process for Counting Annual Patient Numbers:
For many years, administrative personnel have entered demographic information on all patients seen as part of the Iowa Neuromuscular Program onto a spreadsheet. This is the most accurate count, however it is quite labor intensive. To reduce this effort, we have explored generating patient numbers from the electronic medical record (EMR). Please note that the EMR numbers are dependent on accurate designation of a patient as a neuromuscular patient rather than a general neurology patient by schedulers. The accuracy of this designation, particularly for new patients, is imperfect. Therefore, the EMR numbers will be lower than those collected manually and will underestimate the population served by the program. Last year we included both the manual entry numbers and the EMR numbers. The EMR total is ~10% lower than the manual count. We have identified other limitations in using the EMR numbers exclusively. The tables below note which approach was used for counting.

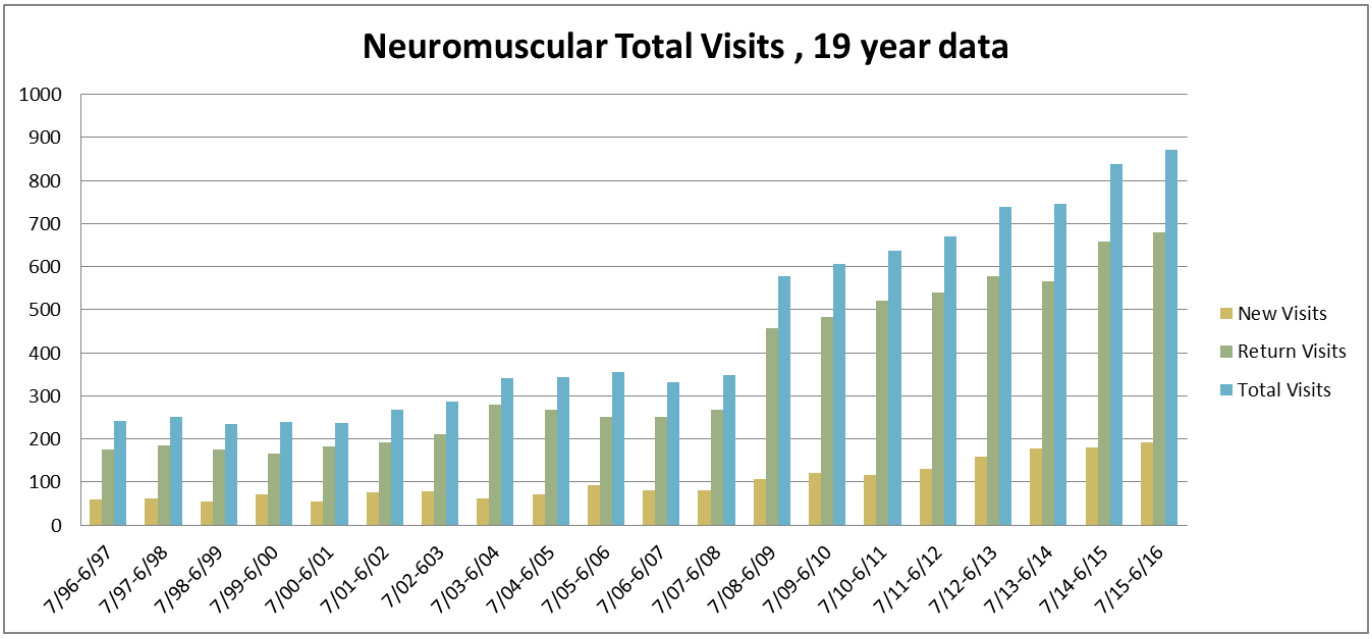
TOTAL VISITS FY16 (EMR numbers)		Total
	Davenport	154
	Dubuque	12
	Des Moines	106
	Mason City	33
	Sioux city	7
	Waterloo	55
Total Community		**372**
UIHC		**467**
TOTAL VISITS FY16		**839**

The chart and table below show longitudinal Iowa Neuromuscular Program Utilization over 19 years, using manual data collection.

Number of Patient Visits (manually entered)

7/96-6/97	7/97-6/98	7/98-6/99	7/99-6/00	7/00-6/01	7/01-6/02	7/02-603	7/03-6/04	7/04-6/05	7/05-6/06	7/06-6/07	7/07-6/08	7/08-6/09	7/09-6/10	7/10-6/11	7/11-6/12	7/12-6/13	7/13-6/14	7/14-6/15	7/15-6/16
43	45	40	40	44	40	41	48	55	49	51	45	49	54	51	55	56	55	63	72
59	63	56	72	54	75	78	63	71	93	80	81	108	122	116	131	159	179	181	191
176	184	175	167	183	192	210	279	267	252	251	268	458	484	521	539	579	566	658	680
241	251	234	239	237	267	288	342	344	356	331	349	578	606	637	670	738	745	839	871

Iowa Neuromuscular Program Patient Visits, UIHC and Outreach Clinics, New & Return Visits, 19 Year Data



Advisory Committee Participation & Statewide Collaboration

- The INMP participated in CCID Meetings over the past year (quarterly meetings). The INMP nurse, Christina Trout, was the CCID Advisory Committee chairperson previously and represented the clinical genetics programs, as well as the INMP.
- The INMP refers to and accepts referrals from CHSC, RGCS, AEA and other state-wide programs. The INMP services are coordinated and integrated services with other programs serving similar purposes and populations.
- Payments received from the receipts of service are used only for the program.
- Involved in CIDAC discussion of newborn screening for neuromuscular disorders.

<u>Observations & Challenges</u>

- The INMP is a gap-filling service, as there are limited providers and care coordinators with expertise in NM disorders
- The number of patients has doubled in the last dozen years, partly due to the fact that patients are living longer by decades.

- Patients require multi-disciplinary services and long-term followup that is often not available in the community.
- Patients are not always physically able to travel short or long distances to access to specialized healthcare, therefore require services brought to their community.
- Funds have continued to decrease even though the demand for INMP services is greater.

Current Updates in Neuromuscular Diseases

Comments from Dr. Katherine Mathews, Director of the Iowa Neuromuscular Program

As recently as 15 years ago, there was limited treatment or management for most neuromuscular diseases. Today, medical management prolongs life and improves quality of life for many people with neuromuscular disease and many other treatments are under investigation. This is a dynamic and exciting time in neuromuscular diseases!

Advances in genetics have led to improved understanding of how muscles get weak, more accurate and less invasive diagnostic testing, and new approaches to treatment. Advances in technology allow improved home management of respiratory failure, mobility, and pain. These advances create a special challenge for a rural state, such as Iowa, as neuromuscular disorders are relatively rare, information is changing rapidly and expertise is limited.

Diagnosis: Diagnosis previously required expensive and invasive tests, such as EMG (using needles in the muscle to measure electrical activity in the muscle), nerve conduction velocity (delivery of electric shocks and measuring time to reach the muscle) and muscle biopsies (surgical procedure).

Genetic testing has virtually eliminated the need for these painful and expensive procedures in many diseases, including Duchenne muscular dystrophy, spinal muscular atrophy, and some forms of limb girdle muscular dystrophy. Diagnostic genetic testing allows a more precise and accurate diagnosis than the previous diagnostic tests, often resulting in more targeted medical monitoring, greater peace of mind for families, and the ability to provide accurate genetic counseling for the extended family.

Management, including new medications: In the past few years, expert guidelines have been published regarding the care of patients with many types of neuromuscular diseases, and additional guidelines are in preparation. INMP personnel have participated in the development of some of these guidelines. Most neuromuscular diseases affect many body systems and require routine monitoring of breathing, heart function, skeletal system and GI function.

Introduction of nighttime ventilation (BiPAP) when the breathing capacity falls has resulted in longer lifespan and has decreased the number of hospitalizations for pneumonia in neuromuscular diseases. This support is provided at home and can be managed by family members. Similarly, mechanically assisted cough can allow home management of illnesses that would previously have resulted in hospitalization.

Improvements in mechanical wheelchairs, standing devices, and braces allow patients greater independence and less pain.

Three exciting new treatments for some patients with neuromuscular diseases were approved by the FDA over the course of about a month in late 2016 and early 2017.

A novel drug, **nusinersen**, was approved for treatment of all patients with spinal muscular atrophy (SMA). This disease is fatal in infancy in its most severe form and results in progressive weakness in all patients. Nusinersen stops disease progression for those severely affected infants, allowing long term survival. In older children, function is stabilized or improved with this treatment. SMA is caused by a loss of the protein survival motor neuron (SMN) encoded by the gene *SMN1*. Loss of SMN results in the death of cells in the spinal cord (motor neurons) required to transmit messages from the brain to the muscles. A closely related gene, *SMN2*, has a single mutation that results in production of an unstable SMN protein that has only minimal function. Nusinersen is an antisense oligonucleotide (ASO) that reverses the effect of the mutation in *SMN2*, so that the *SMN2* gene makes the SMN protein that is critical for the survival of the motor neurons.

For one form of muscular dystrophy (Duchenne muscular dystrophy), steroids prolong walking, delay heart disease, decrease need for spine surgery, and improve breathing. However steroids have significant side effects, one of which is marked weight gain and obesity. **Deflazacort** is a steroid that is less likely to cause weight gain compared to prednisone and is now approved for use in DMD.

Eterplirsen is has provisional approval to treat a subset of boys with DMD with mutations that interrupt the processing of the dystrophin gene at a specific location resulting in absence of the protein. This medication is also an ASO that acts on pre-RNA, in this case to exclude exon 51 in the dystrophin gene, resulting in production of a dystrophin protein that is shorter than normal, but functional. In preliminary studies, this medication slows disease progression.

Experimental treatments: This is an exciting time to be involved in care of patients with neuromuscular diseases, because there are so many treatments under investigation, and as noted above, there has been recent success in demonstrating disease modification. This success impacts the whole field, as investigators, drug companies, and scientists see that treatment is possible and that therapies that affect gene processing can be used safely and effectively in humans. We note that the new therapies are accompanied by a significant increase in work required by the clinical neuromuscular team, as we try to get medications to patients who can benefit, working with insurance companies and other healthcare providers involved in drug delivery.

The INMP team is actively involved in clinical research for neuromuscular diseases. We conduct two natural history studies (dystroglycanopathies and Friedreich ataxia) and have a range of clinical trials to offer. In the past year, we have been a research site for clinical trials in Duchenne muscular dystrophy (6 studies), Friedreich ataxia (1), and facioscapulohumeral muscular dystrophy (1). The opportunity to participate in research directed toward improved treatment is very important to many patients and families in the state.

In Summary: The INMP personnel are committed to ensuring that Iowans have access to the best possible care, including current treatment and access to research. The next section summarizes some of the activities that are outside the scope of the state-funded program, but benefit INMP patients directly and indirectly.

Collaboration, Partnership & Research Opportunities

The State of Iowa is receiving national recognition from the National Institutes of Health and the Centers for Disease Control for its population-based approach to neuromuscular disease.

- Dr. Mathews and Ms. Trout were invited to participate on an expert panel to help develop national comprehensive care considerations for individuals who have DBMD.
- These considerations were developed by professionals from academic institutions and expert clinicians with facilitation of a team from Center for Disease Control. Expert clinicians were selected based on both their relevant clinical and research experience as well as recommendations from acknowledged leaders in the field.
- These DMD care considerations were published in The Lancet Neurology, volume 9, issues 1 and 2, Bushby K, Finkel R, Birnkrant DJ, et al, Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management, and part 2: implementation of multidisciplinary care, pages 77-93 and 177-189, respectively, Copyright Elsevier (2009).
- Updates to the CDC Care Considerations have been submitted to Lancet. Ms Trout chaired a new section on transitioning adolescence with DMD to adult living and healthcare practices. Ms Trout was also asked to chair a supplement article to the journal Pediatrics summarizing these recommendations for their audience. These updates are due to be published later in 2017.

***Research**:*

Research for neuromuscular and related disorders is important. It is not just an academic endeavor, but is required for better medical understanding and management of these rare disorders, always working toward the day that these are managed chronic diseases rather than progressive diseases leading to early death. From a family perspective, this is hope for the future. INMP participation in research allows rapid dissemination of updates to families and direct translation of research into their healthcare.

Muscular Dystrophy Surveillance, Tracking and Research Network: MD STARnet

- MD STARnet is a CDC funded project to assess the epidemiology and clinical course of muscular dystrophies. It initially focused on childhood onset Duchenne and Becker muscular dystrophy, but the infrastructure is now used to examine the epidemiology of all forms of muscular dystrophy.
- Iowa was selected as one of the initial states for this collaborative project and has been continuously funded to carry out statewide surveillance
- It is under the administration of the Congenital and Inherited Disorders Registry in collaboration with the Iowa Department of Public Health and Colleges of Medicine and Public Health.
- The information derived from this project will be valuable in identifying unmet needs of Iowans with neuromuscular diseases.

Clinical Outcome Measures in Friedreich's Ataxia

- Purpose of this study is to identify ways to follow the disease progression in Friedreich's Ataxia and be able to measure changes over a short period of time.
- This natural history and outcome data facilitates design of treatment trials.

Educational booklet on Early-Onset Fascioscapulohumeral Muscular Dystrophy (FSHD)

- Funded by the FSHD Society, a private not-for-profit organization.

- A booklet providing education and suggestions for the schools was created by Iowa Neuromuscular Program personnel, headed by Shelly Mockler, PT.
- This booklet is distributed nationally through the FSHD society and has been very well received.

Clinical trial readiness in the dystroglycanopathies

- Funded through the National Institutes of Health as part of the University of Iowa Wellstone Muscular Dystrophy Cooperative Research Center.
- Researchers examine the clinical presentation of muscular dystrophies caused by changes in the glycosylation of α-dystroglycan.
- Knowledge gained from this study will improve the health care recommendations for patients with this rare disease and allow for improved planning of clinical trials.

Industry sponsored clinical trials (Disease)

- PTC therapeutics (DMD)
- Serepta Therapeutics (DMD)
- Horizon therapeutics (FA)
- aTyr (FSHD)
- Pfizer (DMD)
- Eli Lilly (DMD)
- Fibrogen (DMD)

Patient conferences supported through non-profit partnerships

The following conferences were held in the past year:

- A family-focused conference for patients with dystroglycanopathy is held annually and draws people from throughout the state and country.
- FARA conference for Friedreich ataxia families

Planned for 2017

- MDA DMD Family Conference
- PPMD ESO Duchene

Appendix A

Types of the Neuromuscular Diseases

Brief descriptions of a few of the common disorders seen in the Neuromuscular Program follow.

Myopathies

Duchenne muscular dystrophy (DMD), most common form which affects males.

- Symptoms begin in the first 5 years of life and progress steadily.
- Boys usually lose the ability to walk by age 12 and require the use of power wheelchairs by age 14.
- Although life may be extended with the use of mechanical ventilation, historically, nearly 95% of these individuals die from respiratory insufficiency or heart failure before the age 22.
- This disorder follows x-linked inheritance and women can be carriers without knowing they are at risk for children with DMD.
- 30% of males with DMD also have mental retardation.
- Approximately 1/3 of boys with Duchenne (and Becker) muscular dystrophy did not inherit the disorder, but have the gene alteration as a new genetic mutation. Thus, eradication of the disorder is unlikely.

Becker muscular dystrophy (BMD), less severe form of DMD.

- This is a milder form of Duchenne muscular dystrophy with onset of symptoms ranging from childhood to adulthood
- Life expectancy and level of disability are highly variable.
- Life expectancy is shortened most often related to cardiomyopathy or heart failure

Limb-girdle muscular dystrophies (LGMD)

- Presents in childhood or adulthood with limb and girdle weakness
- Affects males and females equally
- LGMD is caused by mutations in more than 22 different genes, and is characterized by weakness of the shoulder and hip muscles, with progression to the rest of the body.
- The rate of progression and severity of disability is extremely variable.
- Heart and lungs can be affected resulting in heart and respiratory failure

Myotonic dystrophy

- Affects individuals of all ages, but is most debilitating when symptomatic in infancy or childhood.
- A multisystem disease that affects the muscles, central nervous system, heart, eyes and endocrine glands.
- The severe form (congenital myotonic dystrophy) causes profound weakness, difficulty sucking and swallowing, impaired breathing and mental retardation.
- The severity of this disorder often increases with each generation, particularly when passed through mothers.

Neuropathies

Charcot-Marie-Tooth disease

- Group of hereditary motor and sensory neuropathies or peroneal muscular atrophy
- Affect the nerves of the feet, lower legs and hands, resulting in weakness and loss of sensation.
- Vary in severity, this group of disorders usually does not result in loss of ability to walk or shortened life expectancy
- Affecting approximately 4 in 10,000 people, it is a very common genetic problem.

Other: Anterior Horn Cell Disorders

Spinal muscular atrophy (SMA)

- A motor neuronopathy is a disease of the neurons in the spinal cord.
- Wide a spectrum of severity, ranging from a type fatal in early infancy to a type characterized by weakness that is slowly progressive so that patients are able to walk in adulthood.
- Early juvenile form is slower to progress, but leads to loss of ambulation in childhood or young adult life.
- Disorder is inherited in an autosomal recessive pattern, thus affects males and females equally. The prevalence is about 1 in 6,000.