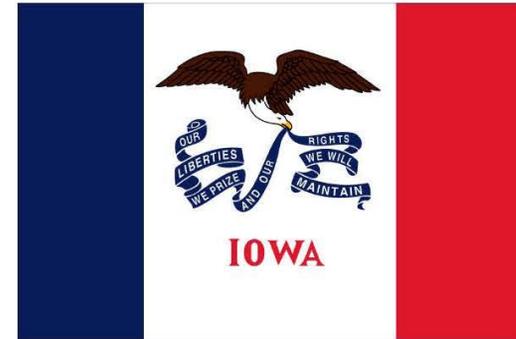
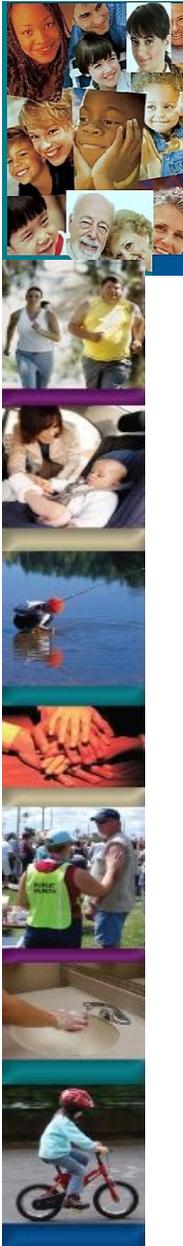


Iowa Neuromuscular & Related Disease Program

Contract 5880MD01 Five year Report
July 1, 2008 through June 30, 2013





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)

Iowa Department of Public Health
Advancing Health Through the Generations



Terry Branstad - Governor

Kim Reynolds - Lt. Governor

Mariannette Miller-Meeks, B.S.N., M.Ed., M.D. - Director

Authorized State Official for this contract:

Julie McMahon, Director, Division of Health Promotion and Chronic Disease Prevention (515) 281-3104

Gretchen Hageman, Chief, Bureau of Family Health (515) 281-4911

University of Iowa Department of Pediatrics (319) 356-1851

Katherine Mathews, MD

Program Director

Christina Trout, RN, MSN

Care Coordinator

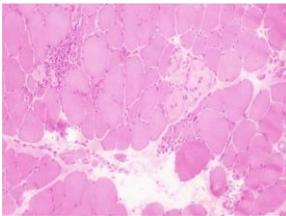
Linda Boehmer, RN, BSN,

Staff Nurse

What is the Iowa Neuromuscular and Related Disease Program?

Iowa Administrative Code, Chapter 4:641-4.6 (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 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) which 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.
- While disabling and life-threatening, many NM symptoms and individual quality of life can be improved with appropriate medical monitoring, early intervention, supportive care in the home and community.

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:
 - World wide 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 (<http://www.cdc.gov/ncbddd/duchenne/index.htm>), 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:

Physician: Katherine Mathews, MD, staffs INMP .25 FTE

- Dr. Mathews is the only Pediatric Neurologist in Iowa with advanced training in genetics & neuromuscular disorders
- She serves as a consultant 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: Christina Trout, RN, MSN, 1 FTE
Linda Boehmer, RN, BSN, .4 FTE

- 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.

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, 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)
- 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.

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.

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

- 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 **210%**.
- 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 51 clinics in FY 11, 54 in FY 12, and 56 in FY13

TOTAL VISITS FY09		New	Return	Total
Community	Davenport	4	25	29
	Dubuque	2	13	15
	Des Moines	20	79	99
	Mason City	1	27	28
	Sioux city	6	41	47
	Waterloo	5	29	34
Total Community		38	214	194
UIHC		83	200	283
TOTAL VISITS FY09		121	414	535

TOTAL VISITS FY10		New	Return	Total
Community	Davenport	5	25	30
	Dubuque		16	16
	Des Moines	16	63	79
	Mason City	1	17	18
	Sioux city		19	19
	Waterloo	3	29	32
Total Community		25	169	194
UIHC		97	188	285
TOTAL VISITS FY10		122	357	479

TOTAL VISITS FY11		New	Return	Total
Community	Davenport	7	22	29
	Dubuque	5	13	18
	Des Moines	18	36	54
	Mason City	2	7	9
	Sioux city	0	13	13
	Waterloo	4	18	22
Total Community		36	109	145
UIHC		97	188	285
TOTAL VISITS FY11		122	357	479

TOTAL VISITS FY12		New	Return	Total
Community	Davenport	11	33	44
	Dubuque	4	10	14
	Des Moines	17	55	72
	Mason City	6	16	22
	Sioux city	2	18	20
	Waterloo	10	29	39
Total Community		50	161	211
UIHC		81	378	406
TOTAL VISITS FY12		131	539	670

TOTAL VISITS FY13		New	Return	Total
Community	Davenport	3	27	30
	Dubuque	0	18	18
	Des Moines	16	125	141
	Mason City	2	16	18
	Sioux city	0	22	22
	Waterloo	3	34	37
Total Community		24	242	266
UIHC		135	337	472
TOTAL VISITS FY13		159	579	738

- See charts following this section, Iowa Neuromuscular Program Utilization, 17 year data.

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.

Advisory Committee Participation & Statewide Collaboration

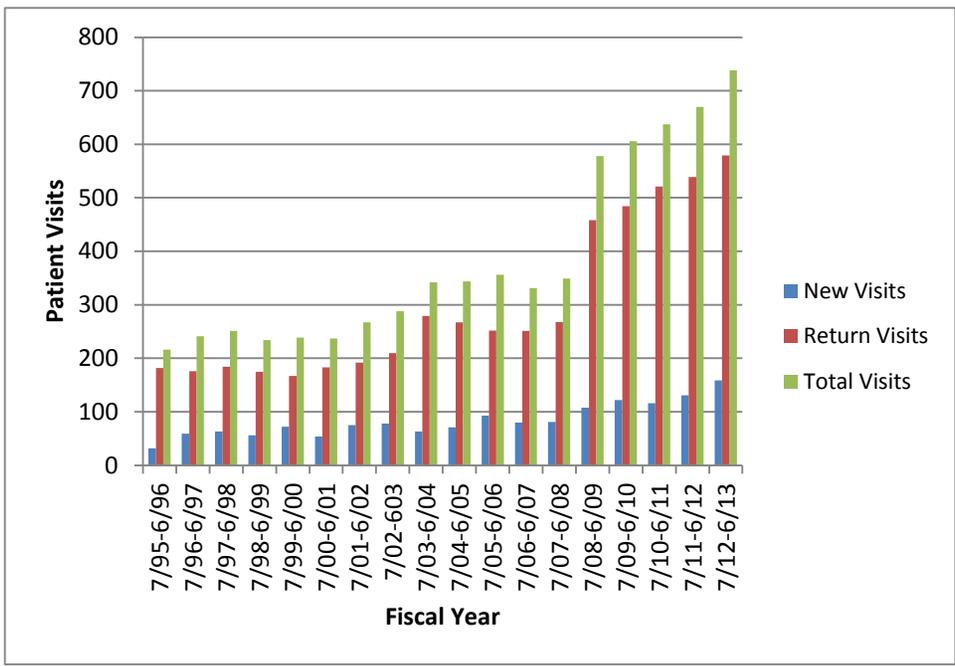
- The INMP participated in CCID Meetings over the past year (quarterly meetings). The INMP nurse, Christina Trout, was the CCID Advisory Committee chairperson previously and represented the clinical genetics programs, as well as the INMP.
- The INMP refers to and accepts referrals from CHSC, RGCS, AEA 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.

Observations & Challenges

- 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.

Iowa Neuromuscular Program Utilization

Iowa Neuromuscular Program Patient Visits, UIHC and Outreach Clinics, New & Return Visits, 18 Year Data



Number of Patient Visits

	Total Neuromuscular Program Patient Visits, UIHC and Outreach; 18 Year Data																	
Fiscal year	7/95-6/96	7/96-6/97	7/97-6/98	7/98-6/99	7/99-6/00	7/00-6/01	7/01-6/02	7/02-6/03	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
Clinics	50	43	45	40	40	44	40	41	48	55	49	51	45	49	54	51	55	56
New Visits	32	59	63	56	72	54	75	78	63	71	93	80	81	108	122	116	131	159
Return Visits	182	176	184	175	167	183	192	210	279	267	252	251	268	458	484	521	539	579
Total Visits	216	241	251	234	239	237	267	288	342	344	356	331	349	578	606	637	670	738

The INMP Helps Iowa to Meet National Performance Measures

The INMP performs many activities that help the State of Iowa meet or exceed national performance measures. These activities are described in this report under heading *Services Provided by the Iowa Neuromuscular & Related Disease Program*. The Iowa Department of Public Health has chosen to prioritize the following national measures in Iowa's Family Health Plan 2009. Each national measure is followed by a reference to the services provided by the INMP to meet these measures.

- ✚ **Assure families of children with special health care needs are partners at all levels of decision making and are satisfied with services received.** **National Measure #2**
 - Patients and families are faced with a variety of decisions for many years once diagnosed with a NM disorder. The INMP strives to provide families with the knowledge needed to make informed decisions, whether acute or chronic, benign or life-threatening.
 - INMP services that help Iowa to meet National Performance Measure #2 are described in services 2b, 2c, 2d, 2e, 3a, 3d, 5a, 5b and 5c.

- ✚ **Assure coordinated on-going comprehensive care within a medical home for children with special health care needs.** **National Measure #3**
 - For children with complex medical care, the medical home often includes primary care, several specialists, the school, and community agencies. Care coordination is integral in maintaining fluent and efficient healthcare to maintain optimal health. The INMP provides care coordination between each of these stakeholders.
 - INMP services that help Iowa to meet National Performance Measure #3 are described in services 2c, 3a, 4a, 4b, 4c, 4d, 4e, 6a and 6b.

- ✚ **Assure families of children with special health care needs have adequate private and/or public health insurance.** **National Measure #4**
 - The evaluation and management of rare and chronic disorders can be costly for families. The INMP provides guidance and support in accessing private, state and federal resources to reduce the barriers to care that finances may create.
 - INMP services that help Iowa to meet National Performance Measure #4 are described in services 3c, 3d, 6b and 6d.

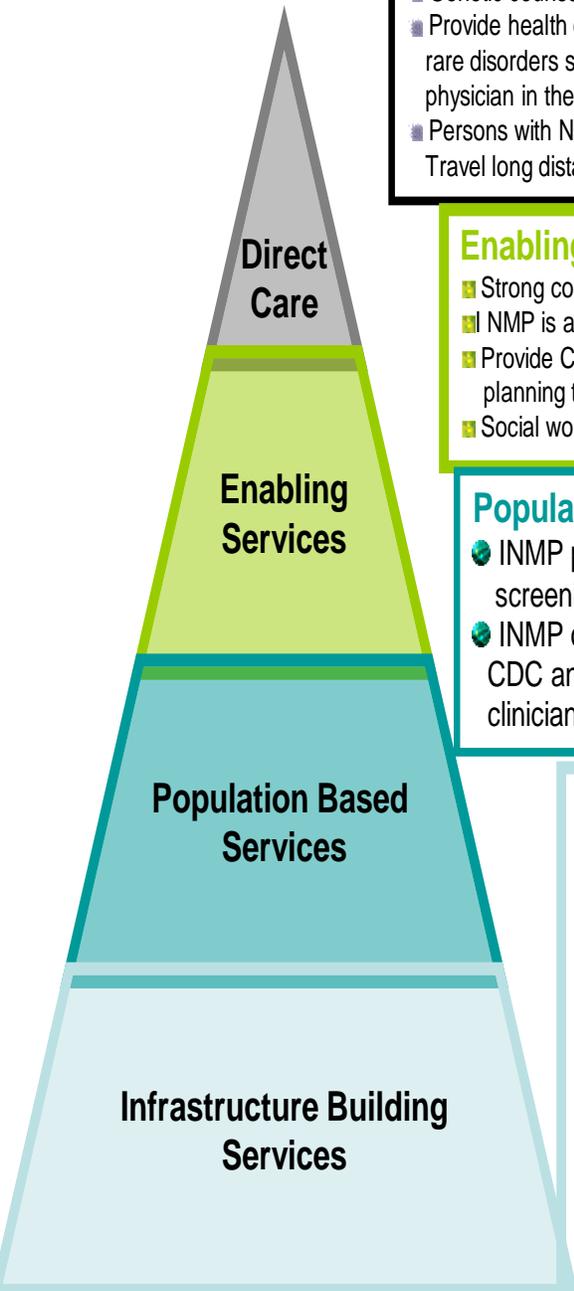
- ✚ **Assure families of children with special health care needs have access to community-based services that are organized for easy use.** **National Measure #5**
 - Geographic distance and community size should not be a limiting factor to appropriate healthcare. The INMP brings specialists closer to many families across Iowa and helps patients and families to identify and access community-based resources to meet their ongoing needs, as well as acting as a resource to the community for the family.
 - INMP services that help Iowa to meet National Performance Measure #5 are described in services 3a, 4a, 6c, 6d, 6e, 6f, 6g and 6h.

✚ Assist youth with special health care needs receive services necessary for successful transition to all aspects of adult life, including health care, work, and independence.

National Measure #6

- Particularly in childhood onset muscular dystrophy, improvements in healthcare are allowing longer life spans. Many of these patients are now considering education beyond high school and thinking about workforce entry. Teens with milder disorders need guidance for vocations and future planning around their disabilities.
- INMP services that help Iowa to meet National Performance Measure #6 are described in services 3b and 5.

The INMP Services Align with Maternal Child Health Priorities



Direct Health Care Services (gap filling) provided by INM program:

- Genetic counseling for family planning
- Provide health care services for underserved special health care needs population (Neuromuscular disorders as rare disorders spread across rural and populated areas of Iowa. There is only one Pediatric Neuromuscular physician in the state)
- Persons with NM disorders are at risk to be underserved due to physical barriers to care because of disability. Travel long distances is difficult with physical disability. INMP provides services in communities across Iowa.

Enabling Services provided by INM Program:

- Strong collaboration with Early ACCESS, AEA, CHSC, health education, family support services, etc.
- INMP is a resource to these agencies as they provide service to patients with neuromuscular disorders
- Provide Care Coordination from time of referral to INMP throughout lifespan. Community and program planning through community-based clinics and communication
- Social work services to ensure access to community, state and federal resources

Population Based Services provided by INM Program:

- INMP provides feedback to the Iowa Newborn Screening Program about future screening for muscular dystrophies. There are national pilots looking at this issue.
- INMP organized & participated in national conferences with patient advocacy groups, the CDC and other national stakeholders. Conferences were held for researchers, clinicians, consumers, & included annual meetings: FSHD, PPMD, MDA & CMD

Infrastructure Building Services provided by INM Program:

- Standards Development: Participation in development of National Guidelines for Duchenne Muscular Dystrophy & Congenital Muscular Dystrophy
- Provide patients, clinicians and researchers access to each other (clinical trials, registries, etc)
- Information Systems: Participation in MD STARnet activity -- a population based monitoring of muscular dystrophy
- Applied research: participation in Wellstone Center Tissue Repository and Database & Neuropathology Conference
- Training: Future providers (MD, NP, PT, RN) & others (law & science students)
- Policy Development & Quality Assurance: National Task Force for Early Identification of Childhood Neuromuscular Disease – Dr. Mathews chaired CDC contract & parent advocacy organization
- Development of materials: Participation in MDA's National Task Force on Adolescent Transition to Adult Independence, Roadmap to Independence.
- INMP highlighted as a model of care: publications & presentations

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, there is management that prolongs life and improves quality of life, with curative treatments being tested for some neuromuscular conditions.

Advances in genetics have promoted understanding of how muscles get weak, improved diagnostic testing, and provided new ideas about how to correct the disease processes. 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 entire family.

Management: In the past few years, expert guidelines have been published regarding the care of patients with these rare 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.

For some forms of muscular dystrophy (Duchenne muscular dystrophy, & possibly dystroglycanopathies), steroids are demonstrated to prolong walking, delay heart disease, decrease need for spine surgery, and improve breathing.

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

Experimental treatments: This is an exciting time to be involved in neuromuscular diseases, because there are so many treatments under investigation. Most are currently in the animal testing stage, but some are in human trials. Current treatment trials include an experimental drug (PTC 124) to “read through” a specific genetic mutation (premature stop), and form of gene therapy to “skip over” the abnormal part of the gene and make a short, but functional protein (exon skipping). Both of these trials are being studied in individuals with Duchenne muscular dystrophy.

Additional trials in other diseases are under development.

In Summary: The INMP personnel are committed to insuring 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). A PDF copy of the articles and a video about the guideline are available through the CDC link, <http://www.cdc.gov/ncbddd/duchenne/index.htm>.

Research:

Research for neuromuscular and related disorders is important. It is not just an academic endeavor, but steps toward improvements in medical understanding and management of disorders, as well as developing potential cures. 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.
- 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 Fascioscapulothoracic 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.

A Clinical Evaluation of FKRP Muscular Dystrophy

- 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 dystrophy caused by changes in the fukutin-related protein (FKRP) gene.
- Knowledge gained from this study will improve the health care recommendations for people with FKRP mutations, and provide a baseline for further study, including potential treatment options.
- A family-focused conference for patients with dystroglycanopathy is held annually and draws people from throughout the state and country.

PTC 124 Clinical Trials in Duchenne Muscular Dystrophy

- Funded by PTC Therapeutics, a private biotech company as a multi-center clinical trial
- PTC is the first investigational new drug designed to enable the formation of a functioning protein in patients with Duchenne muscular dystrophy who have a specific type of mutation (stop codons).
- The INMP had patients enrolled in phase II and phase IIb stages of the study.

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.