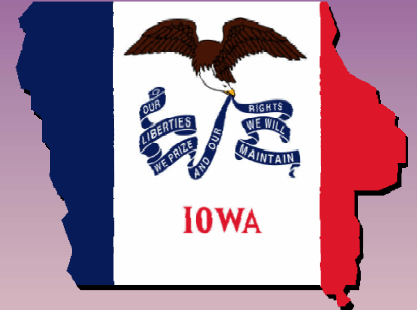
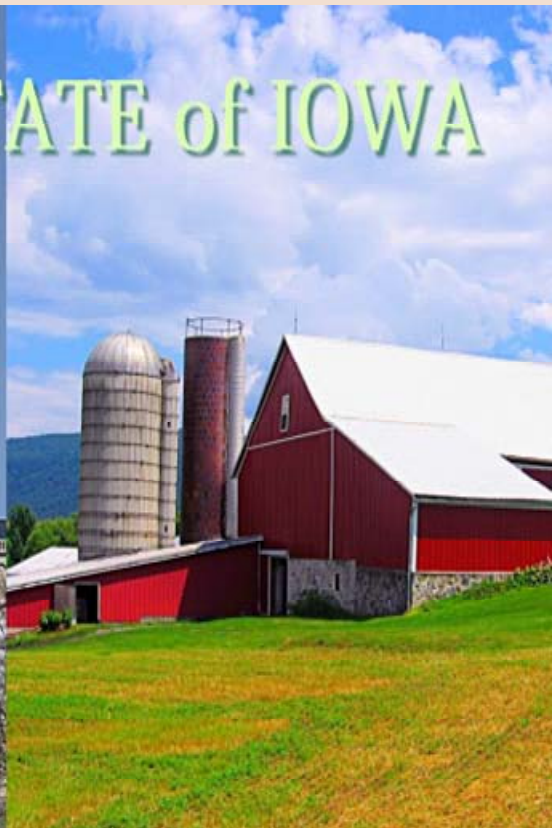
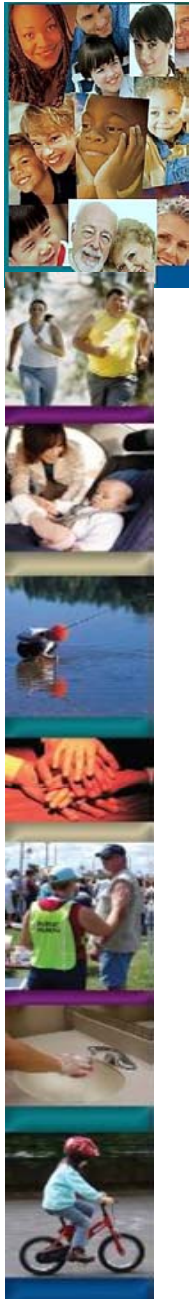


Iowa Neuromuscular & Related Disease Program



Contract 5880MD01 Annual Report
July 1, 2008 through June 30, 2009





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



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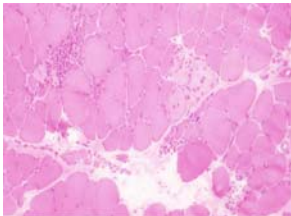
University of Iowa Department of Pediatrics (319) 356-1851

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.

~See family stories about the personal impact of neuromuscular disorders in the next sections of this report.

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), myotonic disorders, 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 July 2008 is 3,002,555.
 - This would mean around approximately 924 Iowans are affected.

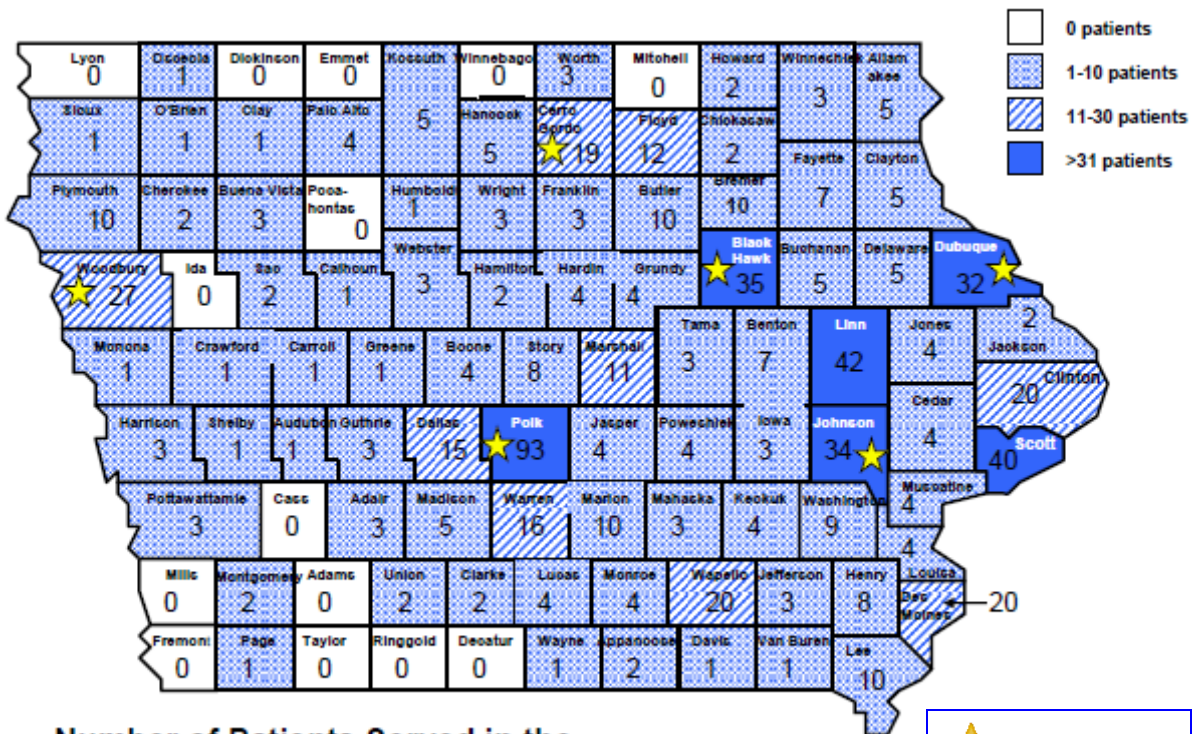
Prevalence of Selected Neuromuscular Disorders in Iowa (1999 data)

Disorder	Prevalence (x10⁶)	Estimated number Of affected Iowans*	# of patients followed through the INMP (% of predicted patients in state)^
Onset of symptoms in childhood			
Congenital Myopathies	10	29	35 (120%)
Duchenne dystrophy	32	92	90 (98%)
SMA	12	34	25 (74%)
Onset of symptoms in childhood or adulthood			
Becker dystrophy	12-24	50	17 (34%)
FSHD	20	57	38 (67%)
LGMD	40	115	30 (26%)
Myotonic dystrophy	50	143	69 (48%)
HSMN	100-200	286-572	43 (15%)

*based on Iowa's 1998 population of 2,862,447

^number of individual patients with the diagnosis seen anytime 1995-1999

Number of Patients Served by Iowa County 2006-2009



Number of Patients Served in the Neuromuscular Program by County, 2006-2009

★ Clinic location

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 (not paid for by INMP budget)

- 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 & Kris Baldwin, 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 recently published in the Neuromuscular Disorders (an international medical journal devoted to Neuromuscular Disorders). Poysky J, Kinnett K. Facilitating family adjustment to a diagnosis of Duchenne muscular dystrophy: April 24-25, 2008, Miami, Florida. Neuromuscular Disorders (2009), doi:10.1016/j.nmd.2009.07.011

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
 - o Information on prevention & early intervention for comorbid risks
 - o Education of school personnel, employers, childcare providers and others
 - o Updates on research for patients, families and healthcare providers
 - o Assistance in planning for transitions from adolescents to adult life
 - o 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:
 - o letters of medical necessity for insurance coverage equipment
 - o FMLA documentation completed to care for children with NM disorder
 - o calls to educators to explain healthcare needs while at school
 - o complete prior approval authorization forms for diagnostic testing
 - o 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 **107%**.
- 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 57 scheduled clinics last year.
- See figure 2, Iowa Neuromuscular Program Utilization, 14 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 figure 1 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.

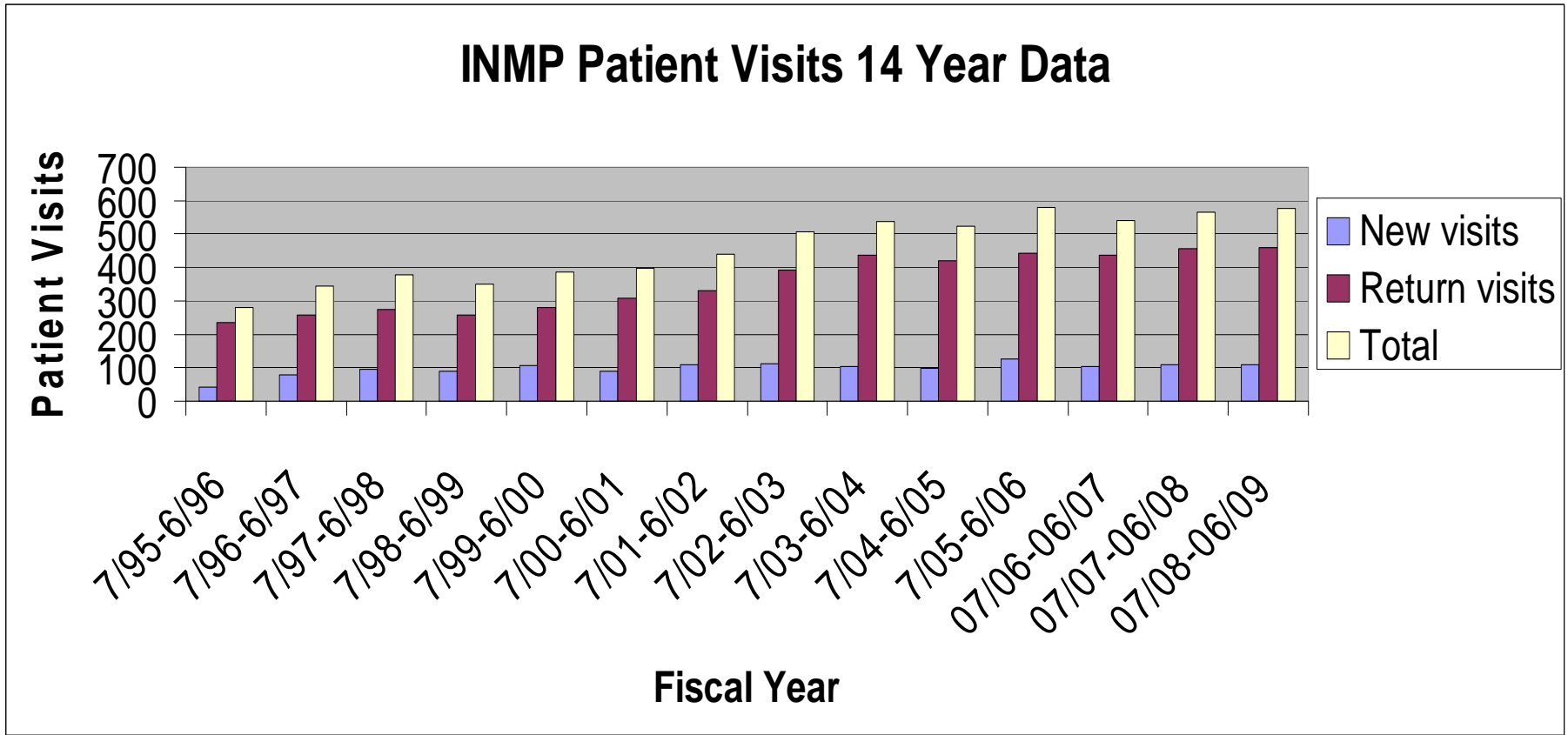
Advisory Committee Participation & Statewide Collaboration

- The INMP participated in CCID Meetings over the past year (quarterly meetings).
The INMP nurse, Christina Trout, was the CCID Advisory Committee chairperson for the past 2 years and represented the clinical genetics programs, as well as the INMP. She also served on the CCID bylaws development committee.
- 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



Number of Patient Visits

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	07/06-06/07	07/07-06/08	07/08-06/09
New visits	42	78	94	90	107	90	109	113	103	99	127	103	109	108
Return visits	235	259	275	258	279	309	330	393	436	420	442	438	456	458
Total	279	345	377	351	386	399	439	506	539	525	581	541	565	578

Patient Stories from the Iowa Neuromuscular and Related Disease Program



This story is presented by Nick's mom....

Nick was diagnosed with Duchenne Muscular Dystrophy (DMD) in January, 2008 at the age of 4. Prior to that time Nick had been a clumsy toddler. He fell frequently and struggled to climb stairs. In November of 2007 Nick was seen by his primary care physician as a follow up to an ear infection appointment. At that time I had asked the doctor to look at how Nick ran. Nick had a wiggle in his run that although as a toddler was cute, I was concerned that Nick wasn't able to run like kids his age. After watching Nick run in the hall, the doctor didn't know for sure. He then asked Nick to sit on the floor and stand up. After watching Nick do this, the doctor looked at me and said that he thought this was Gowers' sign. I had no idea what that was, however by the look on his face I knew it wasn't good. He explained that he thought Nick had Muscular Dystrophy. Nick's primary physician currently had another patient with DMD, who had been working with Dr. Mathews in Iowa City. Nick's primary physician assured us that Dr. Mathews was top notch and scheduled us an appointment with her office.

Within a few weeks we were in Iowa City receiving the confirmation. They had the DNA test to run, however given the symptoms that Nick was displaying, she was confident this would be the final outcome. The wait for the confirmation diagnosis was very agonizing. The blood draw took place in November, 2008; however we didn't get the test results back until January 2009. We were told that that lab was backed up and the Thanksgiving and Christmas holidays caused further delays. We visited Dr. Mathews Iowa City office without Nick in January, 2009 to receive the final diagnosis. Dr. Mathews laid the information out in an understandable way and allowed plenty of time for my husband and me to ask questions. After Dr. Mathews left, Christina Trout came in and provided additional information. It was all overwhelming, however Dr. Mathews and Christina were very patient with our questions, didn't rush us through the information, and assured us that although Nick had this diagnosis, he was the same little boy he was months ago. We went home and tried to make sense of it all. It was explained to us that the breakdown was 2/3rds of the time DMD is hereditary, and 1/3rd of the time it was a "oops" in the genetic code during conception. To our knowledge I did not have any family history of DMD on my mother's side of the family. I have a brother and many male cousins on that side that are perfectly healthy. My youngest sister had a son who too was perfectly fine.

Dr. Mathews had suggested we start Nick on Steroids. After a lengthy discussion my husband and I decided to place Nick on Deflazacort (Calcort). Prednisone was not an option we wanted to go with. Although Prednisone is the FDA approved, insurance covered drug, we also knew that weight gain and DMD were not a good combination. Deflazacort has been used for years outside of the USA to treat this disease. The side effects are minimal when compared to Prednisone. After investigating the cost on line, I placed the order. Nick has been on steroids since the early part of 2009. Within this year we have seen a notable difference in Nick's ability to stand up, run, and climb stairs. He is not doing these activities similar to his peers, however is doing these activities quicker and with a lot less strain than a year ago.

Nick has been wearing AFO (night splints) since May of 2009. Nick doesn't fully know why he is wearing them, but does know his "Magic Boots" as he calls them, gives him power in his feet for the next day. Shelley Mockler is the PT (with the NM team) we have been working with. She coached my husband and me on how to complete Nick's stretches. When she learned we wanted to take Nick to the pool as much as possible, she provided us some additional water stretches. Nick calls it his spider man crawl. In addition, Nick has been taking therapeutic horseback riding sessions at barn where we live. Shelley had explained to us that hippotherapy was another excellent activity for Nick to participate in. As a horse person my entire life, I was thrilled to learn that this was an activity Nick should do. We are very fortunate in our community to have both an indoor pool and a barn with a trained individual on hippotherapy. Shelley has been great resources to us ensuring Nick's PT needs are met.

Our family is very new into this journey of DMD. We have hope and pray constantly that within Nick's lifetime a cure will be found. We have been blessed by having the resources within reach that we do. We leverage the West Des Moines outreach clinic for Nick's 6 month check appointments. Des Moines is much closer for us than Iowa City. My husband and I have been very impressed by the entire staff. Dr. Mathews and Christina are extremely responsive to our needs. They are both very knowledgeable concerning the latest in research. DMD is a disease that no parent would want their child to be diagnosed with. Currently no cure, but yet a community of families with hope that one day a cure will be found. The support of the NM program via Iowa City and the Des Moines outreach clinic has provided us the foundation needed to educate ourselves on this disease and give us guidance for our future challenges.

Comments from the INMP on Nick's story...

Nick's story has some very common and uncommon experiences for children diagnosed with Duchenne muscular dystrophy. It is common for DMD to be diagnosed in boys around age 4 as a result of parents' observations that their child has motor skills that are lagging behind their peers or "look different". It is uncommon to have a primary care provider who has cared for someone with DMD before. Having a primary care provider recognize and refer for diagnosis so quickly provides an efficient and smooth diagnostic process. When the referral was accepted by the INMP staff, calls were immediately placed to the family to arrange an appointment for evaluation and provide the family with anticipatory guidance about what was to come for that appointment. Once the genetic testing confirmed Nick's diagnosis, an hour of time was provided to the family to discuss concerns and learn about the treatment options that would slow the disorder and manage symptoms. This process is considered an important step in establishing trust with families and ensuring adjustment to a serious diagnosis can be made by a supportive healthcare community. Adjustment is ongoing for families as they face many years of physical disabilities, chronic grief and social challenges. Children who are diagnosed with DMD are typically followed by the INMP every 6 months for their lifetime.

EG's story was compiled by review of 20+ years of medical records and discussions with him and his family...

For many families, interaction with the NM Program lasts a lifetime. In 1986, a young couple asked their pediatrician about their son's development. They were concerned that his speech, language, gross motor and social development were slower than his sister's. Examination at that time identified hypotonia and delayed gross motor skills. He had sat independently at age 16 months and was not walking at age 21 months. At his local doctor in Waterloo and parents' insistence, a referral was made to the diagnostic clinics at the University of Iowa Hospitals and Clinics. This boy's initials are EG and this is his medical story. Following this medical review, EG and his mother provide comments on their experiences with the NM Program and living with muscular dystrophy.

Medical summary

In June of 1987, the UIHC pediatric diagnostic clinic discovered abnormal laboratory findings most likely associated with muscle disease, when considered in the context of EG's developmental history. He was 28 months old and had persistent low tone, constipation, developmental delay in many areas, decreased

strength, poor growth and large calves on exam. There was no family history of similar problems, including no one with muscular dystrophy. Referrals were made to a dietitian for poor growth, a gastroenterologist for constipation and the Neuromuscular Clinic for possible muscular dystrophy.

The diagnosis of Duchenne muscular dystrophy (DMD) was confirmed by muscle biopsy. Physical therapy recommendations were made to prevent or postpone joint contractures of the Achilles tendons and hamstrings. EG was registered with the local MDA (Muscular Dystrophy Association). His family was presented with detailed information about the complications of muscular dystrophy that he would experience throughout his shortened lifespan. The NM Program offered medical evaluation and guidance, physical therapy, occupational therapy, speech and educational services and nursing care. EG's health in relation to muscular dystrophy was monitored every 3-6 months in the NM clinic for the next 22 years.

The NM Program services work in coordination with many local and state services. EG was followed by the AEA (Area Education Agency) for delayed speech, language and social development. (Approximately 30% of boys with DMD have mild to moderate cognitive impairment.) EG continued to receive general health care through his local doctor's office, with the NM Program providing guidance and support when the process of muscular dystrophy complicated routine health problems of childhood. For example, EG was considered at higher risk for complications of malignant hyperthermia and respiratory problems when he had general anesthesia for a tonsillectomy. The NM Program coordinated care with the pediatric urologist and gastroenterologists for problems with enuresis and constipation.

Commercial genetic testing had just recently become available as a diagnostic tool in 1988. Genetic counseling and testing was provided to the family and it was determined that EG's mother and sister were not carriers. Therefore, the likelihood of EG's mother or sister having children with muscular dystrophy was low. Most likely, EG had muscular dystrophy as a result of a new genetic change that occurred at the time of his conception, as this happens in one third of all males with DMD. (DMD affects one in 3500 live male births.)

In 1989 (age 4), it became evident that EG was becoming quite weak and falling several times per day. Consent was obtained to participate in a new research trial through the NM clinic, involving oral steroids to slow the disease. While the medication (prednisone) decreased the number of falls and increased his energy level, the side effects of the medication were not tolerated. EG gained weight to the point of obesity and had extreme problems with hyperactivity and irritability. The steroids did not work for EG, but remain a common and often helpful treatment for others with DMD.

EG was still able to walk in 1991, but had difficulty with stairs. He had been prescribed night braces to help control the contractures of his ankles, but they were too uncomfortable. By October 1992 (age 7), EG was using a wheelchair 75% of the time and he lost ambulation by Christmas of that year. The NM clinic provided recommendations to treat leg pain, a prescription for a power wheelchair and a mechanical lift for transferring him from the chair to bed. Just 3 years later, there were increasing concerns about respiratory illnesses as a major threat to EG's health. (Respiratory failure and pneumonia are the most common reasons for death among boys with DMD. Back in the 1980s-1990's, 90% of boys died before age 20 of respiratory or cardiac manifestations of the disease.) He was taught breathing exercises and provided an incentive spirometer.

After losing the ability to walk, contractures worsened and EG developed scoliosis. He was referred to orthopedics and had surgical fusion of his spine to correct the abnormal curvature. While this surgery provided many aesthetic and health benefits, the increased height caused a new problem for EG in feeding himself. When the spine was surgically fused, EG became taller and unable to lower his head by curling the spine. EG couldn't lift his arms to his mouth due to weakness. He also could not bend down to see his homework and write. Thus, following the spine surgery and with progression of the disease, EG lost independence. He required help to feed himself, bath, toilet, dress and for all personal hygiene activities. He was becoming totally dependent on others for most activities of daily living. EG lives with his mother,

whom is a single parent working at least 14 hours per day outside the home. Referrals were made for home health services through a local nursing service.

With the addition of services in the home, financial concerns developed. This was complicated by concerns that Medicaid coverage may not be available each month due to the family income. A referral was made to social services to explore funding options and medical coverage. Before the 1995 school year, a referral was made to Hospital Schools (now known as Center for Development and Disabilities) to further evaluate EG's social, educational and homecare needs and make ongoing wheelchair adjustments. EG had a full time aide at school, an aide to assist in bus transfers to and from school, aides at home to assist with ADLs, and physical & occupational therapy.

In 1997, EG's mother had the first of many long and detailed discussions with the NM Clinic personnel about his future. The risk for respiratory failure was increasing each month. EG's forced vital capacity (a measure of breathing strength) was 43% of the normal value. Soon EG and his family would be faced with decisions about ventilator support and advanced directives. They were given medical advice and psychosocial support throughout the many follow up clinic and phone conversations on this topic. Treatment options included palliative care, mechanical support of breathing without surgery using a ventilator known as bipap or the surgical placement of a tracheostomy for full time mechanical breathing support. With each appointment for two years, the monitoring of EG's breathing showed the disease was progressing. In 1999, EG was hospitalized for the first time with pneumonia. EG and his mother wanted to prolong life, but to avoid use of a machine unless it was the last option.

In January 2000, phone discussions with the NM nursing staff became very frequent. Erik was experiencing chest pain, insomnia, headaches, trouble coughing up secretions, problems with his wheelchair not fitting, poor appetite, urinary hesitancy and constant discomfort. EG was crying several times per week and asking, "What's happening to me?" Counseling was recommended. EG did not think he could physically tolerate a hospitalization for evaluation and treatment, but without it, medical recommendations could not be made. Without treatment, EG's health was in serious jeopardy. Arrangements were made working with local nursing and medical caregivers to do a home study of his breathing while sleeping. This study showed that he was not getting enough oxygen while sleeping at least 50% of the time. EG (age 15) was afraid to go to sleep, in fear that he would stop breathing. EG's pulmonary function testing at the April 2000 clinic showed his forced vital capacity was 11% of normal.

Finally, EG was started on mechanical assistance (bi-pap—bi-phasic positive air pressure). By October 2000 he had better energy, fewer headaches and less pain. He was evaluated for voice activated computer software for writing and playing videogames. He planned to attend a vocational rehabilitation workshop to explore work skills for individuals with disabilities. Although he was feeling better, monitoring of his blood gases revealed ongoing respiratory insufficiency. Appointments over the next year focused on making EG comfortable and discussions of whether to have an elective surgery to place a tracheostomy tube for improved breathing with a ventilator. By February 2003, EG was also developing problems with his heart, which would eventually lead to congestive heart failure. (The heart is also a muscle and works poorly in the later stages of this disease.)

EG was admitted to the University of Iowa Hospitals and Clinics to optimize his bipap treatment and begin treating the heart disease. A plan was established to have the tracheostomy surgery the week after his high school graduation. A tube would also be placed for feeding (gastrostomy tube) in anticipation of his losing the ability to swallow as weakness progressed. EG utilized the care coordinators to facilitate the transition from inpatient care to a higher level nursing care within his community. Care was coordinated by phone and included nurses from the NM program, the hospital Continuity of Care program, EG's local doctor, DHS, the insurance companies & home health health nurses. It was determined that EG would need 8-16 hours of direct care per day by nurses following the surgery, a wheelchair to hold the ventilator and a few other safety features in the home.

In May of 2003, this surgery was cancelled as EG's community did not have adequate nursing support to safely care for him following surgery. His alternatives were to accept placement into a nursing home able to provide care to someone on a ventilator, have his mother quit her job to care for him in the home alone or postpone the surgery indefinitely. The surgery was permanently cancelled with a plan to keep him comfortable and as healthy as possible at home using non-surgical ventilation. For 6 years, EG has been in fragile, but stable condition in his home. His sister is an LPN who assists his mother in caring for him and he has nurses and aides come to the home many hours per day, so that his other can work.

Now, in 2009, EG's home care is now in jeopardy because the home care agencies consider his nearly full-time use and dependence on BiPAP "skilled nursing care" that is beyond the scope of practice for him nursing aides. His private insurance and Medicaid coverage through waivers only provides a certain level of assistance. These healthcare dollars do not go very far if his home care requires RN, rather than non-licensed care providers. EG is no longer able to travel outside of his home town due to dependence on the BiPAP assistance from breathing and physical discomfort. The NM Clinic comes to his community twice per year to provide assessment and management and continues to provide medical and psychosocial support to him and his local providers and agencies by phone and email between actual appointments.

Mother's summary and comments

(The following are the personal and unedited comments from EG's mother. She was provided with the summary of EG's medical history and reflects on her memories and thoughts from the past 16 years.)

7/8/86 I was very disheartened when they couldn't figure out what was wrong with EG. I was told not to compare him to my daughter, regarding the milestones of sitting up, crawling, speaking, etc. I knew something was wrong. No one would believe me.

8/86 I took EG to our family doctor to have a lead poisoning test done. (Still in search of what is wrong with my child). His testing led to a diagnosis of muscular dystrophy. I was glad to finally have a diagnosis, but that later turned into devastation when I started reading about muscular dystrophy. Still, at that time, I had no true realization of how hard my life was to become, trying to raise a child with md and watching him suffer.

8/23/87 This was the first time I came into contact with Dr. Ionasescu and the NM Clinic. Many questions were answered and a muscle biopsy was performed to find which type of md. My greatest fears were realized when the results came back as Duchenne muscular dystrophy. The NM clinic hooked us up with a local EDI (Early Development Interventions services) to teach us physical therapy exercises.

5/25/89 The NM clinic did testing on my daughter, mom, sister and me. It was determined that it was most likely a new mutation in EG. I had a sense of relief that I had not passed this on to EG. I was glad to know my sister and daughter would have children with no risk of passing MD to their children.

3/20/90 EG started on prednisone to slow the disease. He gained a lot of weight, which made it harder to get around. He became very irritable, almost mean-spirited. The year on prednisone was the "year from hell". My petite, quiet child became an overweight bully.

5/3/95 EG had luque rods put in to correct for scoliosis (surgical spine fusion). We would highly recommend the procedure, as he was able to breathe so much better.

9/5/97 Dr. Mathews offered to talk to EG about progression of the disease and whether he would choose to go on a ventilator when the time comes. I finally agreed to let her, as I knew that I could not do it without becoming too emotional. (Thank you Dr. M!)

4/18/00 We were very glad to have the NM Clinic come to Waterloo. Talking to EG in Iowa City clinics was very physically and emotionally draining for both of us.

Early spring 2003 EG lost the ability to play his video games. He was devastated as this was the last independent thing he could do. At this point, I became very depressed. I cried in the shower, at work, in my bed—anywhere that I was away from EG. I didn't want to add to his worries by seeing me fall apart. Dr. Mathews prescribed Wellbutrin for me, which has seemed to help.

In the spring of 2004, I came to the realization that my child was in the end stages of a horrible disease. On the way to Iowa City for a sleep study to monitor his bipap machine, I pulled over and had the talk with EG that I had dreaded for 16 years. I had to tell my beautiful child that he would never reach my age. I told him of his life expectancy with and without the ventilator. I had to tell him that there would be a time he would lose the ability to talk and eat. He was so brave. He was most concerned about losing the ability to speak and then how he would communicate his needs.

At the completion of the sleep study, it was determined that it was time to go on a ventilator and a feeding tube. We scheduled the surgery, but over a 2 month period could not find LPNs or RNs to care for him after the surgery at home. It is not cost effective for the home health agency to pay RNs to do the work and too much of a liability to allow his current aides to do his cares once he has a tracheostomy and ventilator. So, we wait.....

Over the years, we have relied heavily on the NM clinic for information, testing, prescriptions, adaptive technology and for comfort. They have "held our hands" from the beginning to the end stages of EG's DMD. We would have been lost were it not for their constant assistance. I take great comfort knowing that information, advice and comfort are only an email away. The clinic has also helped us find funding sources for equipment and home modifications, though referrals to appropriate agencies.

I have realized over the years that it is very expensive to raise a disabled child. I know now that I will never have all the nice things that others have. EG and I are punished every day by the politicians that continue to cut programs, etc, that can assist us. When he was younger, we got 40 hours a week in aide services. That was cut a few years ago to 28 hours/week and at age 21, he lost even more. Parents of disabled children are forced to pay dearly for home and vehicle modifications, adaptive clothing, special foods, and aides so we can work outside the home and a lot of Tylenol.

Life with EG has been an adventure. We've had to be creative in adapting things for his needs. I've had my house torn apart to widen doorways, install stair lifts, a roll in shower and more. There are a very limited number of things we can do for entertainment. Of course, the things he can do are costly. We've had to improvise, adapt and overcome at every stage of decline with this disease. I personally have no time for "a life". I work, sleep and take care of my son. However, he is the light of my life. I'd rather know him, as he is, then never to have known him at all.

EG's comments

So what does EG think about life with DMD? Now, graduated from high school, EG also gives his comments on living with DMD.

"It sucks! I can't run, swim, party with friends, camp, go boating, etc. I always hurt physically. It takes 3 hours to get up in the a.m. and 2 hours of cares to go to bed at night. I don't have much of a day left after that."

We respectfully thank EG and his mother for remembering their journey through the NM Clinics for the last 22 years. There are many statistics and numbers to document the NM Program's productivity. We hope that the faces and stories behind the statistics will tell more than any numbers, as these are the real reasons for the NM Program.

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**

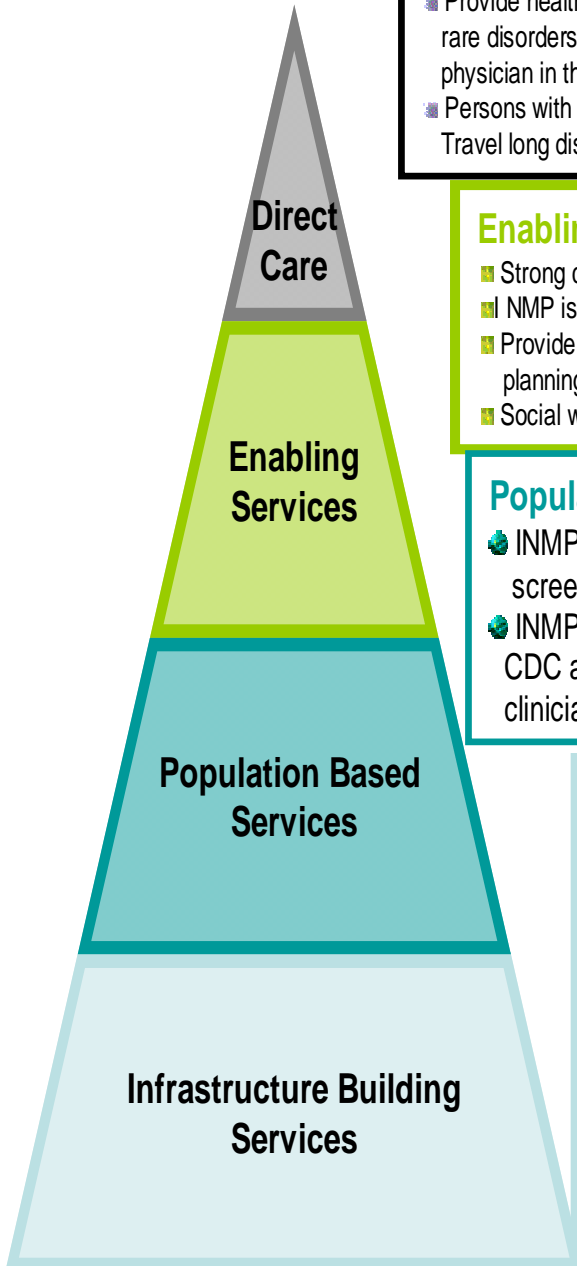
- Geographic distance and community size should not be a limiting factor to appropriate healthcare. The INMP brings specialists closer to many families across Iowa and helps patients and families to identify and access community-based resources to meet their ongoing needs, as well as acting as a resource 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 Duchenne 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 chairs 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. In 2009, there is management that prolongs life and improves quality of life, with curative treatments being tested for some neuromuscular conditions.

Advances in genetics have given us the tools to understand how muscles get weak, improved diagnostic tests, and provided new ideas about how to correct the disease process. Advances in technology allow improved home management of respiratory failure, improved mobility, and decreased 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 in Iowa 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 that are under investigation. Most are currently in the animal testing stage, but some are in human trials. For example, medicines are being tested

in human trials to increase the cell's production of the missing protein in spinal muscular atrophy and several Iowa families are participating in these trials. Iowans with Duchenne muscular dystrophy are taking an experimental drug (PTC 124) to "read through" a specific genetic mutation (premature stop), so that they make a small amount of the protein that would otherwise be missing. Another Duchenne trial starting this year will use a special kind of gene therapy to "skip over" the abnormal part of the gene and make a short, but functional protein (exon skipping).

In animals, drugs are shown to decrease scarring in the muscle, decrease muscle breakdown, decrease cell death, decrease fatigue, and improve blood flow. The drugs that are safest and most promising will be put into clinical trials in the near future.

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 will be 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.

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 national project to assess the epidemiology and clinical course of childhood onset Duchenne and Becker muscular dystrophy.
- Iowa was selected as one of the initial states for this collaborative project which now includes 7 states.
- 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.
- This information derived from this project will be valuable in identifying unmet needs of Iowans with neuromuscular diseases.

United Dystrophinopathy Study

- Ongoing study to compare specific dystrophin mutations (gene changes) with clinical signs and symptoms of DBMD.
- Iowa is one of six neuromuscular centers taking part in this project which started at the University of Utah.
- For DBMD families this is an opportunity for access to genetic research that may better define their family's genetic change and/or allow them to be recruited into future clinical trials based on this information.

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.
- Researchers anticipate that this study could be used in future drug treatment research studies.

A Study of Cardiomyopathy in Friedreich Ataxia

- A retrospective review of medical records from living and deceased persons with Friedreich ataxia.
- The purpose was to gain understanding of the role of cardiac dysfunction in morbidity and mortality in Friedreich ataxia.

Clinical Features of Early-Onset Fascioscapulohumeral Muscular Dystrophy (FSHD)

- Conducted by the University of Iowa with funding from the FSHD Society, a private not-for-profit organization.
- Purpose of this study was to examine and describe the clinical presentation of persons with early onset (infantile) FSHD.
- Of particular interest are the health care needs and experiences of people with this very early onset of this disorder.
- Knowledge gained from this study will improve the health care recommendations for persons with FSHD, and provide a framework for further study into potential new treatment modalities.

A Clinical Evaluation of FKR P 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 FKR P mutations, and provide a baseline for further study, including potential treatment options.

BforSMA (Biomarkers for Spinal Muscular Atrophy)

- Individuals affected with SMA and control subjects were recruited from Iowa to participate by giving blood, urine and completing an exam.
- Identify biomarkers in blood and urine that contribute to variability of onset and severity in spinal muscular atrophy

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 has patients enrolled in phase II and phase IIb stages of the study.
- If proven effective, this drug would be developed as a new medicine to treat or prevent muscle weakness—a major medical breakthrough.

2009 Program Budget

Neuromuscular Program	Contract	Program Income from fees
Income	\$99,799	\$38,400
Reversion of Income	<u>(\$ 1,000)</u>	
Total Income	\$98,799	\$38,400

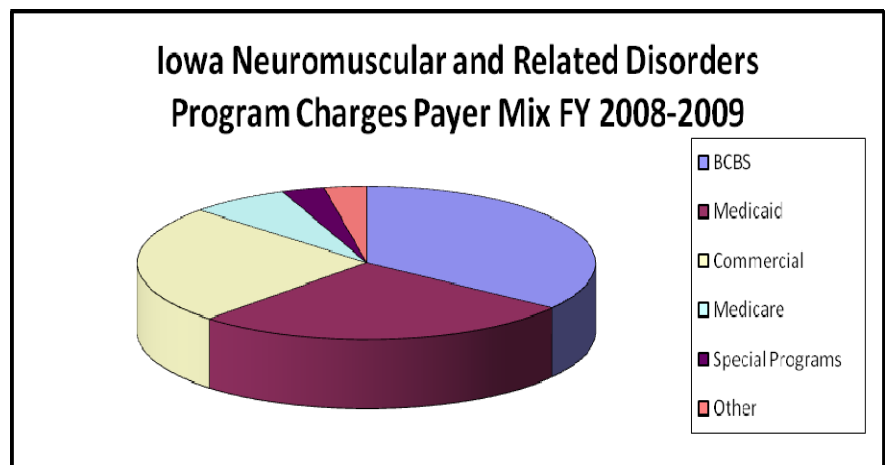
Income from professional charges is linked to the program and used for program specific expenses.

Iowa Neuromuscular Program: Budget

- The program scope is greater than the state dollars and fee income provided.
- State funding is critical to providing special healthcare needs for this vulnerable Iowa population.
- In 2009, state dollars allowed the purchase of specialized physician care and nurse care coordination and support.
- **Without state dollars, the program could not sustain the community based clinics and care coordination necessary.**
- The University of Iowa Hospitals and Clinics Department of Pediatrics provides other needed services in kind, such as social workers, physical therapy, additional nursing support and program expenses.
 - Other expenses include the personnel costs (mentioned above), clinic letters, mailing/postage, billing, supplies/computers/printers, travel to community clinics, rental of community clinic space, overhead for maintenance of buildings and facilities, and other costs associated with maintaining a healthcare program.
 - It is estimated that the Neuromuscular Program (even with state funding) operated with a subsidy of approximately \$54,000 in FY09 from the Department of Pediatrics.
- Payment reform is needed. Nurses cannot bill for care coordination which is such a critical part of the program. Genetic counselors are not able to bill either.
- Improved collaboration with other special health care services is a key.

Charges/Payments and Payer Mix

FY09 Community Clinics- Neuromuscular	
Charges	\$66,438
Payments	\$38,400
Payer Mix	
BCBS	35%
Medicaid	27%
Commercial	25%
Medicare	7%
Special Programs	3%
Other	3%



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.