Center for Special Children
A program of Vernon Memorial Healthcare

Spotlight ON HEALTH

Special Edition vmh.org | Vernon Memorial Healthcare

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Sharing the Good

Dr. James DeLine started practicing medicine at the La Farge Medical Clinic in La Farge in 1983. Over the years he's made quite an impression on community members and the patients he has cared for.

Dr. DeLine has always understood the need for collaboration in an effort to offer the best care for his patients, which is why the LaFarge Medical Clinic became a part of Vernon Memorial Healthcare in 2004.

In 2014, Dr. DeLine founded the Center for Special Children, a program within La Farge Medical Clinic, to care for Wisconsin's large concentration of Amish and Old Order Mennonite people.

His goal in starting the Center for Special Children was to focus on the particular health needs of Plain community families.

Dr. James DeLine stands next to the base of the Center for Special Children windmill that was donated to the La Farge Medical Clinic shortly after the new clinic was built in 2015.

The Center for Special Children program is able to offer affordable genetic testing that can provide families with answers they may not have had and has the potential to spare families from costly hospitalizations. This is all made possible based on the help and collaboration of many.

The kind of care and study that this incredible program offers, is the future of medicine. It is often called precision medicine, meaning the medicine model being practiced is customized to the individual patient and their genetic make up.

The medical community continues to learn more about how to use genetic testing to help doctors choose the best course of treatment for all patients.

With the findings occurring in the collaborations born from the Center for Special Children program, plans of care are able to be developed for expectant women with genetic disorders and cord blood testing is now easily incorporated into care plans of at risk families. Plus, regular education conferences with birth attendants and midwives are providing opportunities for improved care at the community level.

The work at the La Farge Medical Clinic - Center for Special Children program is making a difference in the history of medicine and is more importantly helping more people and families than Dr. DeLine ever imagined possible.

As more is understood, more can be done. Dr. DeLine said, "It's hard to treat something you don't recognize and understand. Each time a new genetic condition is identified, the search for a cure can begin."

The care and passion Dr. DeLine has shown in his work with the Plain population has recently been highlighted in two articles that have reached far beyond the La Farge Medical Clinic service area.

Authors Mark Johnson from the Milwaukee Journal Sentinel (MJS) and David Tenebaum from UW-Madison wrote separate articles showcasing many of Dr. DeLine's efforts over the last 36 years, as well as the group effort and collaboration currently occurring with UW-Madison Department of Pediatrics, La Farge Medical Clinic, and Vernon Memorial Healthcare.

Johnson's MJS article made such an impact, it became a front page story in the USA TODAY. While Dr. DeLine is not one to seek out the spotlight, he does appreciate the fact that the mission and message is being shared and discussed on a broader scale, simply for the hope that it is able to help families receive the care that they need.

If you would like more information about the La Farge Medical Clinic - Center for Special Children or would like to read the full articles discussed, please visit www.vmh.org.

A Letter of Gratitude...

Dear Friends,

First of all I would like to thank you all for participating in the 2019 Benefit Auctions; was it donating, buying or just joining us for the day. Both auctions were a big success.

If I think of all the support shown in preparing for and the day of the auctions, my thoughts go to all the families receiving care through this program. I am sure it is very humbling to receive all this support but let's remember there are families among us struggling physically and financially. How much worse would this be without program like this?

Many of us may never have had any experiences with genetic disorders and may question why do we need such a program? Remember genetic disorders are inherited. Therefore, some families have on going health care needs from one generation to the next.

Since we know that only a small percentage of the people have genetic disorders, then we also know that all of the money funded to this program would otherwise need to come from the few people that have disorders.

As time continues, so the program's budget increases each year. In the Fall of 2020, the program will welcome Dr. Katie Williams as a full-time pediatrician and geneticist. This will allow the program to serve more children and families, providing genetic evaluations and specialized pediatric care for children with a variety of medical needs. Dr. Williams worked with Dr. Holmes Morton for five years at the Clinic for Special Children and we are excited for the future of the program with her serving children and families here in Wisconsin and our surrounding communities.

Let us give all honor and glory to God and not unto man.

~ Center for Special Children Advisory Board Member

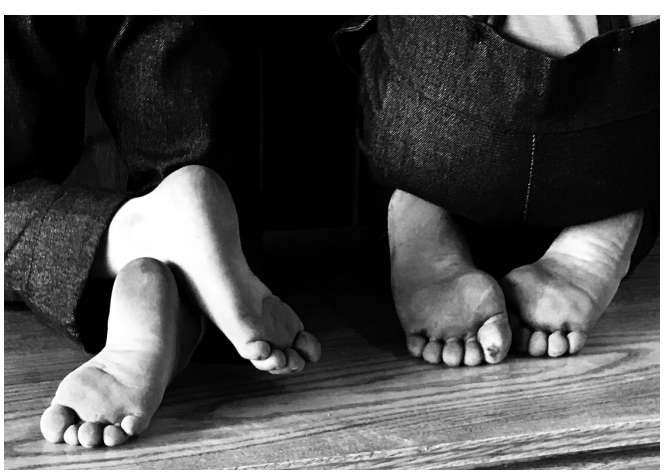
New Screenings & Treatments

Jennifer Kwon, M.D.
Pediatric Neurologist, UW-Madison

Spinal muscular atrophy (SMA) is a genetic disease that causes muscle weakness by affecting the motor nerves (or lower motor neurons) that travel from the spinal cord to the muscle.

SMA is seen in the general population and affects 10,000 to 25,000 children and adults in the United States, and therefore it is one of the most common rare diseases. Depending on the type of SMA, the weakness can be severe or mild.

Children with the most severe weakness are never able to sit without support. This form is called type 1 SMA. Children with type 1 SMA will die by the age of 2 years, without a feeding tube or equipment to help with breathing.



Other children with milder forms of SMA might be able to sit (this is type 2 SMA) or stand (type 3 SMA) for a time, but they generally require wheelchair support and may also need feeding and breathing support as they get older. Though they are weak, children with SMA have normal brains and intelligence.

SMA is caused by changes in a gene, called Survival Motor Neuron 1 or SMN1. We have two copies of SMN1, one we inherit from our father and one from our mother. To have SMA, both copies of the SMN1 gene must be damaged. If only one copy of the SMN1 gene is damaged, then the person is healthy and called a carrier. If both parents are carriers, there is a 1 in 4 chance with each child, that SMA might occur.

Today, SMA can be treated, but the treatments are more effective if they are started very early in infancy. This is especially true with type 1 SMA. If we can start treatment in the first month of life before the weakness is noticeable, then these children may be able to develop normally.

To start treatment this early, we have to diagnose SMA very early. The best way to do this is to screen newborns for this disorder. This is why Wisconsin will be screening newborns for SMA as well as other serious disorders requiring early treatment.

Using newborn screening, we can diagnose infants before they become weak.

Once an infant is diagnosed with SMA, the infant should be started on one of the two new and powerful treatments for SMA.

One treatment, nusinersen (Spinraza) was introduced 3 years ago. Spinraza has to be given into spinal fluid by a spinal tap every 4 months for the patient's entire life.

A newer treatment, introduced in May, 2019, is gene therapy for SMA, called Zolgensma. Zolgensma is the best choice of SMA treatment for most infants, because it only needs to be given once.

Still, there are regular laboratory tests that must be done every week for a month, and then every other week for 2 months, that must be done to look for serious side effects that can occur shortly after gene therapy is given.

Infants from the Plain community have participated in Zolgensma research trials that show that the drug is safe with close monitoring. When given early, children may show no signs of weakness at all!

Gene therapy is very expensive ($2.1 million), but there is a way for members of the Plain community to receive the drug at no cost, though there will still be costs related to the frequent visits and lab tests.

The company that makes gene therapy, Avexis, has a special policy for Plain community members who signed an IRS 4029 form. It does require that the patient's family complete an application to be sent to the company and agree to come for recommended clinical visits and lab tests.

Dr. Kwon visits the Center for Special Children and sees children with neurological disorders to include seizures, headache disorders, developmental disabilities, and disorders of the nervous system.

Therapy Days: Therapeutic Approach to Genetic Conditions

The Center for Special Children has teamed up with Vernon Memorial Healthcare's Therapy Department for over 3 years to improve the quality of life and independence of children with genetic and metabolic disorders.

Each year, Physical Therapists, Occupational Therapists and Speech Language Pathologists travel to the La Farge Medical Clinic to team up with Dr. DeLine to support not only the children, but also families. Over 32 children have benefited from these services to date.

These visits give families the opportunities to ask questions, get recommendations and leave with resources on how to improve their child's independence. Families can receive information on equipment, tools and exercises that could improve development.

Such resources can assist in activities such as dressing, bathing, feeding, speaking, sitting and walking. Treatment plans are individualized based on a child's age, diagnosis, providers' concerns, and the families input

The next Special Children's Clinic Therapy Day is scheduled for Summer 2020. Each session includes a follow-up visit with a medical provider, as well as one-on-one time with a Physical Therapist, Occupational Therapist and Speech Language Pathologist.

We invite those families interested in bringing their child to a Therapy Day to contact Sheri Hammond at 608-625-4039.

Pictured are the VMH Therapy and CSC team that work with families during the Therapy Days at La Farge Medical Clinic. This team consists of VMH Physical Therapy, Occupational Therapy, and Speech Therapy, as well as La Farge Medical Clinic staff.

Parent's Note

When we were blessed with our little "Special Needs Miracle Baby" we wondered in what difficult paths this new little life would take us. Our baby was born 7 ½ weeks pre-maturely and was Down Syndrome! So, she required medical assistance right from the start.

We had no experience with a Special Child so we knew we'd need help and we also knew we'd want her to be taken care of by someone who understands her needs. At first it was as if we were on a roller coaster of up and downs – twists and curves as we tried to understand the needs of a child who is Downs.

There were so many questions we wished we'd have answers for. So, it was with grateful hearts when we were guided to the Center for Special Children. But as it always is with new places, when you walk thru those doors the first time, you wonder what the outcome will be… But we have had some wonderful experiences with Dr. DeLine. He has such a true and caring heart, as we have learned, and he takes time to explain everything in a way we can understand. He no longer is just our doctor, but also a friend whom we feel comfortable with to trust and rely on for our needs.

Dr. DeLine is super in what he does and he has an awesome staff. Between him and Sheri Hammond we have found help in so many ways….my husband with his Parkinson's, our daughter who was just recently diagnosed with a bicuspid aortic valve, our Special daughter with her Special needs and with my on-going health issues. Always trying to provide us with the best of care, but also always seeking ways to keep the costs down, which is greatly appreciated.

It's amazing how God has created our bodies and we have learned a lot of how and why some are born with Down Syndrome. And God provided gifted minds and hands thru our doctors to figure out the different strains of Special Needs' children and how to deal with each different issue. And it is with that knowledge that gives us the insight to reach out and correspond with our Special children more properly.

Without the help of those who know, we as parents would have a struggle to understand their needs and the poor child would struggle along with us! Therefore we are very blessed!

Thank you for being here for us!
~ Elmer and Lizzie Miller

Inherited Eye Diseases in the Plain Community

by Dr. James DeLine, Medical Director, Center for Special Children

Our ability to see well is a gift which we often take for granted. I still remember with wonder placing glasses on for the first time at age 9. I recognized people across the room and saw a squirrel in a tree which were experiences I had never had. I have always been thankful for my glasses and my vision.

For many of us, our eyes work perfectly or can be benefited just with glasses. However others have conditions which affect their vision. Some of these problems are sporadic having nothing to do with our inheritance or genes.

Others occur from accidents or injuries. But some conditions come from inherited disorders. At the Center for Special Children, we see many children whose vision is affected by these conditions.

Some conditions affect only the eyes; others are part of a disorder which affects other parts of the body as well. With the expert assistance of Dr Melanie Schmitt, a children's eye doctor from the University of Wisconsin, we have been able to clarify the underlying cause of vision problems in many children.

Retinal diseases affect the specialized area in the back of the eye. Inherited conditions affecting this part of the eye seen in the plain community include oculocutaneous albinism (OCA), retinitis pigmentosa, Leber's congenital amaurosis (LCA), Jalili syndrome, and Bardet Biedl syndrome. OCA affects the vision and is associated with very light hair and skin and eyes.

Some forms of OCA have some pigment in the skin and hair, so are not completely white. Retinitis pigmentosa and Leber's congenital amaurosis do not have any change in skin color but cause progressive vision loss over time. The course of vision loss varies depending on the condition. In addition, even with the same disorder, each individual may progress differently or have other associated issues within the eye.

Jalili syndrome causes vision loss and light sensitivity associated with very poor teeth. This rare disorder had never been identified in the Amish community or in the US until a family was diagnosed by our research team.

Since then several other families have been identified. Bardet Biedl syndrome causes vision loss along with kidney problems, differences in development, and sometimes extra fingers or toes.

Glaucoma is an eye disease associated with elevated pressure in the eye. If not detected and treated, children and adults with glaucoma gradually lose vision without pain or other symptoms. Pressure within the eye can generally be improved with eye drops. Occasionally specialized surgery is needed to give better control of the pressure within the eye and preserve vision. Like other eye disease, glaucoma may be sporadic or may run in the family as an isolated finding or associated with other eye disease.

Cataracts are common in our older relatives. However occasionally they occur in infants and children. If diagnosed and treated early, these children develop normal vision. In order for normal vision to develop, the eyes must be straight and be able to receive light. Then during the first months and years of life, vision develops normally. If eyes are not straight or light is blocked by cataracts, proper brain development for vision does not occur.

If diagnosis is delayed until a year or two of life, these children never develop normal vision. So early diagnosis is important to maintain vision. All newborn babies should be checked for cataracts. Sometimes cataracts occur from an inherited condition. We have two families in which cataracts have occurred in multiple children. In one family, the children also have congenital heart disease. In the other, the children have challenges in development.

These conditions appear to be new disorders, never before identified. So of course, as we learn from these children, the knowledge gained will help children with similar conditions all across the world and from all backgrounds.

Treatment of eye disease depends on the underlying problem and requires an accurate diagnosis. Lazy eyes are commonly treated with patching the good eye, to force the weaker eye to develop and strengthen.

Special glasses may be used. Sometimes surgery to align the eyes is necessary to optimize vision. Cataracts should be detected early and referred promptly.

If mild, they may be able to be followed, as long as vision is developing normally. If more severe, removal of the cataract may be necessary to prevent permanent vision loss. Glaucoma also needs to be detected and treated in order to prevent loss of vision. Usually eye drops are sufficient but sometimes specialized surgery is necessary to maintain vision.

Treatment of retinal disease remains imperfect but progress is being made. One form of retinal disease, a form of Leber's congenital amaurosis, is now being cured by a type of stem cell therapy. This is the first eye disease in which the missing gene has been replaced and allowed the return of vision. This amazing success in treatment of a previous cause of blindness should give hope that over time, more such conditions will be cured with treatment.

With so many varieties of eye problems, how is a parent to know when their baby or child needs an eye exam? Indicators of eye disease may include eyes that do not align properly ("lazy eye" or strabismus), jiggly eyes (nystagmus), squinting, or a difference in the appearance of the eye (color, cloudy or mis-shapen pupil, etc).

Of course, if an infant doesn't seem to focus on mother or father, that is another indicator of potential eye or brain disease. Early evaluation is wise, as some of the conditions result in vision loss which could be prevented with early treatment.

Optometrists are not medical doctors but have specialized training to check eyes and prescribe glasses. Many communities have such eye doctors. Optometrists may be able to assist with strabismus (lazy eye) and clarifying whether eye disease is present.

Ophthalmologists are medical doctors who specialize in eye disease. They tend to be located mainly in larger hospitals and clinics. Infants and children with eye disease may need to see an ophthalmologist in order to clarify the nature of some rare diseases and direct treatment.

Most families with concerns about their child's eyes or vision will consult their midwife, family doctor, or local optometrist for guidance. They usually will know whether the specialized care of a medical ophthalmologist is necessary.

If you have additional questions, feel free to contact us at the Center for Special Children.

Family Day

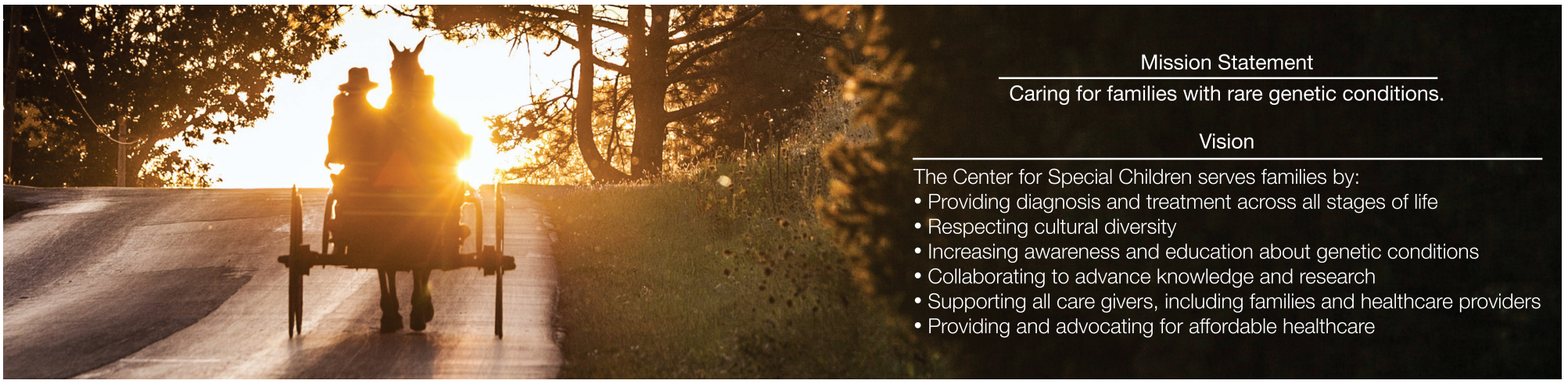
Last spring, the Center for Special Children hosted a Jalili Syndrome and Other Eye Disorders Family Day in La Farge. In addition to inviting families identified with this disorder, we also invited families with low vision problems. The day included a presentation from Dr. Gonzalez, a dentist from the Milwaukee School of Medicine.

Dr. Gonzalez discussed practical care for amelogenesis imperfecta (poor teeth that occurs with Jalili syndrome) and overall good dental care for all children. He was able to do a few brief exams and sent dental supplies (toothbrushes, toothpaste, etc) home with everyone.

Dr. DeLine gave a presentation on Jalili Syndrome and different types of low vision genetic disorders. Our research doctors from Windows of Hope (Exeter, England) were present and were able to meet with families to answer questions. Because of their expertise and families' willingness to participate in the day, we were able to identify diagnoses for a handful of families providing them with answers and ongoing care.

The best part of the day included a presentation from Rhonda Staats of the Wisconsin Council of the Blind and Visually Impaired. Rhonda, who is blind herself, is an advocate for the blind community and an avid braille. She raised two boys, held numerous jobs, and has been on many boards for different organizations that advocate for the blind community.

Rhonda presented alongside two plain community members with varying degrees of low vision. The presenters shared braille writers and other low vision aids that help in their daily lives. The information and aids presented were valuable to those of low vision. We had a great day of learning and fellowship.



Mission Statement

Caring for families with rare genetic conditions.

Vision

The Center for Special Children serves families by:

- Providing diagnosis and treatment across all stages of life
- Respecting cultural diversity
- Increasing awareness and education about genetic conditions
- Collaborating to advance knowledge and research
- Supporting all care givers, including families and healthcare providers
- Providing and advocating for affordable healthcare

Our program is advised by a group of twelve volunteers, who meet every other month to review our mission, activities, and finances. We are honored that the following individuals have dedicated their time and talents to support this program:

Chairman
Raymond Fox
Plain Community Rep.

Vice Chairman
Jerry McGeorge
Executive Vice President of People
Organic Valley

Board Treasurer
Nathan Nolt
Plain Community Rep.

Board Treasurer
Karen Traynor
Chief Financial Officer
Vernon Memorial Healthcare

Ernest Graber	Plain Community Rep.
Joseph Schrock	Plain Community Rep.
Adrian Roberg	English Community Rep.
Crist Hershberger	Plain Community Rep.
Korina Pubanz, RN, CPM, LM	Guild of Midwives
Tammy Raeder, RN, BSN	Nurse - Eau Claire County
Marlin Weaver	Plain Community Rep.
Christine Seroogy, M.D.	University of Wisconsin

In 2020, the Program has an operating budget of $310,000. Our goal is to raise two-thirds of this amount at our two benefit auctions, which will be held in Cashton, Wisconsin on June 20 and Withee, Wisconsin on July 18. Additional support comes from three major, local sponsors:

Vernon Memorial Healthcare
So much care. so close.

Vernon Memorial Healthcare provides continued support for the program and serves on the organizing board. With their continued support, we are providing care for families close to home.

ORGANIC VALLEY

Organic Valley continues to generously support the program. Because of their support in 2015, we were able to begin providing care at the program's inception.

WISCONSIN
UNIVERSITY OF WISCONSIN–MADISON

UW-Madison provides outreach clinics for metabolic disorders, cardiology, opthalmology and neurology. Through UW-Madison's commitment, the program can offer specialty services, genetic testing, community outreach and education.

Donations to the Center for Special Children are used to provide all aspects of care. From offsetting the cost of office visits for families with genetic disorders to supporting innovative research into rare conditions, donations make a difference. Financial gifts provide a 24 hour phone answering service, specialized medical equipment, and access to cutting edge, cost-effective genetic testing. Most importantly, you're making it possible to identify and treat rare diseases at a local level while contributing to a worldwide bank of genomic knowledge that benefits all cultures and communities.

Questions about the Program can be directed to Sheri Hammond at (608) 625-4039
La Farge Medical Clinic, 206 North Mill Street, La Farge, WI 54639