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ROLL NUMBER

DESCRIPTION

4034

2007 SENATE HUMAN SERVICES

SCR 4034

2007 SENATE STANDING COMMITTEE MINUTES

Bill/Resolution No. 4034

Senate Human Services Committee

☐ Check here for Conference Committee

Hearing Date: 3-21-07

Recorder Job Number: 5364

Committee Clerk Signature Mary K Monson

Minutes:

Senator Judy Lee called the committee to order.

Roll was taken and all members were present.

Senator Judy Lee opened the public hearing on SCR 4034.

Senator Carolyn Nelson from District 21 introduced the resolution. This resolution is to bring awareness to Smith-Lemli-Opitz Syndrome known as SLOS. See attachment #1. She introduced Mrs. Gretchen Noah.

Gretchen Noah: Her first impression was that this would just be a token bill and not accomplish much. SLOS children typically have profound mental retardation. Their son is mild and can walk and talk. She and her husband have felt it is their job to raise awareness and funds for the foundation. Raising awareness can help families who are seeking a diagnosis for their own child. Studying rare diseases helps to gain insight into related diseases. SLOS is related to autism. Her son's mild case could drive treatment studies for children with autism. If this syndrome is diagnosed early, dcotors can significantly alter the progression of the disease by giving the children cholesterol. The size the children will grow to can be changed, their behaviors can be improved if they diagnose it early. She feels the support groups for families affected by disorders such as SLOS are very important. It communicates to the families that

they are important and their children are important. Spurred on by their son having SLOS, they have raised $40,000.00 for SLOS Foundation. She was frustrated that it took so long because no one knew about the syndrome. It took 2 years to get a diagnosis. She asked her geneticist why pediatricians don't know the warning signs for children with syndromes and ADHD and autism, etc. He said it is that they are not educated in it. She raised $21,000.00 and they are hosting a conference that will train social service professionals, medical professionals, how to recognize the warning signs of the whole gamut of special needs and conditions. The conference is April 23 – 24 in Fargo. The conference is titled "Hearing the Hoofbeats and Thinking Zebras." See attachment A and attachment #2. It has been well received and is long overdue. She made note that if you are not in the state system, you have no idea what programs the state has available. She said her experience with getting on the state program was very favorable. They got on immediately. Some of their friends had long waits in other states. The support her family has received has been much appreciated.

Senator Lee made mention that the rare syndromes need to find a voice. If the families band together they can be heard.

Gretchen Noah: A syndrome is considered rare if fewer than 100,000 people have it. The people who have these rare syndromes do need to be heard.

Senator Heckaman: How does Markie take his medication? Shots? Liquid?

Gretchen: He takes liquid cholesterol, cherry flavored. He was off it for three weeks to be part of a study being done in Oregon and he started showing some self abusive behaviors and tantrums. As soon as he went back on the medication the behaviors leveled out.

Senator Mathern from District 11 testified in support of SCR 4034. See attachment # 3.

Senator Dever asked whether the House Appropriations Committee had acted on SB 2326 yet.

Senator Mathern said SB 2326 had not yet been acted on.

More testimony in favor? None

Opposed? None

Neutral? None

Senator Warner made a do pass motion on SCR 4034.

Senator Dever seconded the motion.

Roll Call Vote: Yes 6 No 0 Absent 0

Carrier: Warner

Date: 3-21-07

Roll Call Vote #: 1

2007 SENATE STANDING COMMITTEE ROLL CALL VOTES

BILL/RESOLUTION NO. SCR 4034

Senate **HUMAN SERVICES** Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number

Action Taken Do Pass

Motion Made By Sen. Warner Seconded By Sen. Dever

Senators	Yes	No	Senators	Yes	No
Senator Judy Lee, Chairman	✓		Senator Joan Heckaman	✓	
Senator Robert Erbele, V. Chair	✓		Senator Jim Pomeroy	✓	
Senator Dick Dever 2	✓		Senator John M. Warner 1	✓	

Total (Yes) 6 No 0

Absent 0

Floor Assignment Senator Warner

If the vote is on an amendment, briefly indicate intent:

REPORT OF STANDING COMMITTEE

SCR 4034: Human Services Committee (Sen. J. Lee, Chairman) recommends **DO PASS** (6 YEAS, 0 NAYS, 0 ABSENT AND NOT VOTING). SCR 4034 was placed on the Eleventh order on the calendar.

2007 HOUSE HUMAN SERVICES

SCR 4034

2007 HOUSE STANDING COMMITTEE MINUTES

Bill/Resolution No. SCR 4034

House Human Services Committee

☐ Check here for Conference Committee

Hearing Date: March 29, 2007

Recorder Job Number: 5613

Committee Clerk Signature Judy Schock

Minutes:

Chairman Price: We will open the hearing on SCR 4034.

Senator Carolyn Nelson, District 21 Fargo: I am sorry I could not bring the main focus for this resolution with me today. He is a cute little boy about three years old. See attached testimony. This is about a disease that years ago we didn't know any thing about. In1980 we probably had 4 kids diagnosed with autism in the Fargo public schools. This disease is one they are just now starting to study because 80% of the pregnancies are carrying fetuses that have this disorder in the miscarriage, or still born. A number of the children who are actually born don't make it past the age of 1 or 2. Markie is now 3. The Mother knew something wasn't right, and went to many doctors, and was persistent. You will see in the material I handed out to you, there are not many in ND. We know of 4 in the state. There could be more, people don't always see doctors. This disease blocks the ability to process cholesterol. They are trying things out with Markie. There is a liquid that looks like a cough syrup that he takes daily for cholesterol. They think there are only about 500 in the US living with this. Many die early. They are looking to what can be done genetically to fix this. That is what we are asking you to look at today. This is a resolution to bring awareness and education.

Chairman Price: Just curious is this disease passed onto males or both genders?

Senator Nelson: It sounded like most of them were males.

Representative Conrad: I move a do pass consent to the calendar, seconded by

Representative Schneider. The vote was taken with 8 yeas, 0 nays, and 4 absent.

Representative Schneider will carry the bill to the floor.

Date: 3/29
Roll Call Vote #: 1

2007 HOUSE STANDING COMMITTEE ROLL CALL VOTES
BILL/RESOLUTION NO. "Click here to type Bill/Resolution No."

House HUMAN SERVICES SCR 4034 Committee

☐ Check here for Conference Committee

Legislative Council Amendment Number

Action Taken Do Pass Consent Calendar

Motion Made By Rep. Conrad Seconded By Rep. Schneider

Representatives	Yes	No	Representatives	Yes	No
Clara Sue Price – Chairman	✓		Kari L Conrad	✓	
Vonnie Pietsch – Vice Chairman	✓		Lee Kaldor		
Chuck Damschen	✓		Louise Potter	✓	
Patrick R. Hatlestad			Jasper Schneider	✓	
Curt Hofstad	✓				
Todd Porter					
Gerry Uglem	✓				
Robin Weisz					

Total (Yes) "Click here to type Yes Vote" 8 No "Click here to type No Vote" 0

Absent 4

Floor Assignment Rep. Schneider

If the vote is on an amendment, briefly indicate intent:

REPORT OF STANDING COMMITTEE

SCR 4034: Human Services Committee (Rep. Price, Chairman) recommends **DO PASS** and **BE PLACED ON THE CONSENT CALENDAR** (8 YEAS, 0 NAYS, 4 ABSENT AND NOT VOTING). SCR 4034 was placed on the Tenth order on the calendar.

2007 TESTIMONY

SCR 4034

#1

SENATE

NORTH DAKOTA LEGISLATIVE ASSEMBLY

SENATOR CAROLYN NELSON
District 21
1 Second Street South, #5-402
Fargo, ND 58103-1959
cnelson@nd.gov

STATE CAPITOL
600 EAST BOULEVARD
BISMARCK, ND 58505-0360

Assistant Minority Leader
COMMITTEES:
Judiciary
Government and Veterans Affairs

SCR 4034

This is a resolution to bring awareness to Smith-Lemli-Opitz Syndrome, known as SLOS. The syndrome was first described by geneticists David Smith, Luc Lemli and John Opitz in 1964 but it wasn't until 30 years later that scientists discovered the cause.

SLOS blocks the body's ability to make or process cholesterol. The disorder affects 1 in about 40,000 births and is most common in whites of European ancestry.

This resolution is intended to raise awareness of the disease and I'm very happy to introduce the mother of an SLOS child, Mrs. Gretchen Noah. Gretchen and Mark Noah are parents of Markie, a sweet, cute, rambunctious 3 year old. Gretchen is here today to talk to you about Markie, his medical situation, and her dreams for him.

SCR 4034

Senator Carolyn Nelson

A concurrent resolution to bring awareness to SLOS Disease (Smith-Lemli-Opitz syndrome).

This resolution was brought about from the family of Markie Noah, a 3 year old boy Fargo boy who was diagnosed with SLOS disease in the spring of 2006.

SLOS blocks the body's ability to make or process cholesterol. It can cause serious birth defects, or death in severe cases. It has no cure.

SLOS is characterized by distinctive physical features - a small head, upturned nose, droopy eyelids, cleft palate, short thumbs, low-set ears, a single palm crease and webbing between the second and third toes - as well as mental retardation or learning disabilities, and behavioral problems.

Infants with SLOS have weak muscle tone, experience feeding difficulties, and grow more slowly than other children. A majority of the patients have autistic characteristics.

The diagnostic rate is 1 in about 50,000 and only about 500 people in the United States are living with the disorder.

Researchers believe that there are no people with SLOS disease who live independently, except for Markie. Markie has a mild case of the disease which is allowing researchers to learn more about him and his disease.

Dr. Forbes Porter, a researcher with the national institute of health, is studying mutations in Markie's genes and how his cells synthesize cholesterol. Treating the learning and behavior problems associated with SLOS could involve trying to change brain chemistry.

Dr. Porter stated that through his research with Markie, the outcome could impact thousands of lives, including SLOS patients as well as other children with more common disorders, such as autism.

According to Dr. Porter, about 80 percent of SLOS pregnancies never survive to a full term. More than half of the children with the syndrome must be tube-fed for several years.

The disorder affects 1 in up to 40,000 births and is most common in whites of European ancestry.

The resolution will raise awareness of the disease so that mothers can get test to see if their unborn child has the disease so that they may receive early treatment. The added awareness will also aid other children so that they may be properly diagnosed with the disease so that they can live a better quality of life.

In August of 2006, Markie's family hosted a walkathon, which raised over $40,000 for SLOS disease awareness.

Nelson, Carolyn C.

From: Jayne Brown [jbrown@medicine.nodak.edu]
Sent: Monday, March 19, 2007 3:54 PM
To: Mathern, Tim; Nelson, Carolyn C.; Fiebiger, Tom D.; Flakoll, Tim; Mathern, Tim; Pomeroy, Jim R.
Subject: SLOS resolution and state genetics plan

Hello,
On behalf of the Division of Medical Genetics, located at the School of Medicine & Health Sciences at UND, I would like to state we have four confirmed cases of Smith-Lemli-Opitz Syndrome diagnosed since 1983. We have three patients with a last known address within North Dakota and one in Minnesota. For your information, John T. Martsolf, MD is the only geneticist in North Dakota, therefore the statistics concerning patients seen in North Dakota by a geneticist would be maintained in our office.
Unfortunately, we cannot account for the number of North Dakotan residents seen for genetic appointments outside of North Dakota.

I would also like to inform you of a conference we are putting on April 23 & 24, 2007 in Fargo. Monday morning we will have Dr. Susan Sparks talk about Smith-Lemli-Opitz Syndrome, following this we will focus on North Dakota newborn screening by utilizing a panel discussion and then the emergency treatment of metabolic disorders with Dr. Cheryl Greenberg. We will follow Tuesday with Dr. Bryan Hall talking about dysmorphic features, Dr. Martsolf will talk about the process of making a referral for genetic services, as well as a parents discuss their process through the diagnostic system. For more information about the conference you may visit our web site at www.medicine.nodak.edu/genetics.

We are currently in the final stages of writing a State Genetics as recommended by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services. The goal of the state genetics plan is to write a working plan to address the genetic needs of the North Dakota citizens.

As genetics is integrated into public health, all public health programs need to be strengthened. This requires a commitment to sharing resources to address and identify gaps and encompass integration of population-based screening, genetic risk assessment, health promotion and disease prevention strategies within chronic disease, cancer, environmental health and other programs. To foster the integration, many states have developed state genetics plans. The _intent_ of a state genetics plan is to enhance genetics into public health and health care practices. The health status of North Dakotans should be optimized by using genetic/birth defect services. Genetic medicine should also improve public health care and social services systems within the state.
Therefore, we would love to have legislators input and invite you to join us April 22 at the Ramada Plaza from 5:00-6:30.

I look forward in hearing from each of you regarding your interest.

Sincerely,
Jayne Brown

--
Jayne Brown
Human Service Information Coordinator
Division of Medical Genetics
UND School of Medicine & Health Services PO Box 9037 Grand Forks, ND 58202-9037
(701) 777-4277

Conference Goals

Upon completion the learner will be able to:

- Describe the signs, symptoms and current issues concerning Smith-Lemli-Opitz Syndrome (SLOS).
- Identify metabolic disorders detected with ND newborn screening.
- Identify and manage emergency care for specific metabolic diseases.
- Facilitate and strengthen relationships between the health care professionals and the family support professionals.
- Identify resources available for families.
- Identify legislative impacts on healthcare related to genetics & newborn screening.
- Recognize families' struggles through the process of finding the diagnosis.
- Identify services available through the North Dakota Genetics Program.
- Recognize the diagnostic approach with a child who has dysmorphic features.

Continuing Medical Education

This activity has been planned and implemented in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education (ACCME) through the joint sponsorship of the University of North Dakota School of Medicine and Health Sciences and Dakota Medical Foundation. The University of North Dakota School of Medicine and Health Sciences is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

The University of North Dakota School of Medicine and Health Sciences designates this education activity for a maximum of 14 Category 1 Credit(s)™ toward the AMA Physician's Recognition Award. Physicians should claim credit commensurate with the extent of their participation in the activity.

This conference has also been approved for 13 contact hours by the North Dakota State Board of Social Work Examiners (www.ndbswe.com).

Conference Schedule

Monday, April 23, 2007

8:00- 8:30	Registration and continental breakfast
8:30- 8:45	Welcome and Opening Remarks
8:45- 9:30	Spectrum of Smith-Lemli-Opitz (SLOS), Dr. Susan Sparks
9:30-10:15	Neurological features and functioning in SLOS. Dr. Susan Sparks
10:15-10:45	Break (in exhibit area)
10:45-11:30	New SLOS Research, Dr. Sparks
11:30-12:30	Lunch with Markie Noah
12:30-2:00	Newborn Screening Panel Discussion, including Cystic Fibrosis.
2:00-2:15	Break (in exhibit area)
2:15-4:00	Continuation of Newborn Screening Panel Discussion
4:00-4:15	Break (in exhibit area)
4:15-5:15	Adventures in Metabolic Nutrition. Dr. Cathy Breedon
5:15-6:15	Social Hour (no host)
6:15-8:30	Dinner with Dr. Cheryl Greenberg (Practical Approach to Emergency Treatment of Metabolic Disorders.)

Tuesday, April 24, 2007

8:00-8:30	Continental Breakfast
8:30-9:30	Diagnostic approach with a child who has dysmorphic features. Dr. Bryan Hall
9:30-9:45	Break (in exhibit area)
9:45-10:45	Referral to a Clinical Geneticist? Now What? Dr. John Martsolf
10:45-11:30	Legislative impacts to genetic healthcare. Donene Feist
11:30-12:15	Family resources for the health care professionals. Panel Discussion
12:15-12:30	Break: Pick up Box Lunch
12:30-2:00	Parent Panel: Parents' perspectives on the diagnostic process.
2:00	Closing Remarks

Sponsored by:

UND School of Medicine & Health Sciences
Division of Medical Genetics &
Dakota Medical Foundation

A

Hearing Hoofbeats and Thinking ZEBRAS

"Look for the ordinary but remember the rare."

Screening, Testing & Management of Children with Genetic Disorders

April 23 & 24, 2007
Ramada Plaza & Suites
& Convention Center
Fargo, ND

The University of North Dakota
School of Medicine
& Health Sciences

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DAKOTA MEDICAL FOUNDATION
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DIVISION OF MEDICAL GENETICS
SCHOOL OF MEDICINE AND HEALTH SCIENCES ROOM 5910
501 North Columbia Road Stop 9037
GRAND FORKS, NORTH DAKOTA 58202-9037

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Grand Forks, ND
58201
PERMIT #10

Hearing Hoofbeats and Thinking ZEBRAS

Screening, Testing & Management of Children with Genetic Disorders

Conference Fees

Early Bird Registration (prior to Feb. 28, 2007)	$70.00
Registration (Deadline March 28, 2007)	$90.00
Student Registration (valid student ID required)	$25.00

Scholarships available for UND Medical Students

Continuing education credit will be given only to those who attend all sessions and complete required documentation.

Payment Method

Please submit payment with your registration.

Hotel Reservation Information

The conference hotel is:

Ramada Plaza & Suites & Conference Center
1635 42 Street SW
Fargo, ND 58103
(701) 277-9000

To reserve a hotel room, please call the Ramada directly and reference the conference to secure the group rate.

Exhibitors

If your organization would like to exhibit at the conference, please contact Jayne Brown at (701) 777-4276 or email her at jbrown@medicine.nodak.edu.

Who should Attend:

Health and Human Services Professionals (ie. MD, PA, NP, nurses, OT/PT, speech/language, social workers)

A special Thank You goes out to our Genetic Advisory Committee for your support and assistance.

Thank you to Cullen Children's Foundation for being a Commercial Sponsor.

Presenters

Cathy Breedon, Ph.D., Clinical Nutrition Specialist at MeritCare Medical Center.

Cheryl Greenberg, M.D., Head of the Department of Pediatrics and Child Health; Medical Director, Child Health Program; Director of the Metabolic Service; clinical geneticist with the Program in Genetics and Metabolism; and Professor in the Departments of Pediatrics and Child Health, Biochemistry, and Medical Genetics at the University of Manitoba.

Bryan Hall, M.D., Former Chief of Genetics and Dysmorphology at the University of Kentucky.

John T. Martsolf, M.D., Professor of Pediatrics and Director of Division of Medical Genetics at UND School of Medicine and Health Sciences.

Susan Sparks, M.D., Ph.D., Pediatrician, Clinical Geneticist, and Clinical Biochemical Geneticist at the Children's National Medical Center (CNMC) in Washington, DC.

Newborn Screening Panel:

Barb Schweitzer, RN, BSN, (moderator), Director of the North Dakota Newborn Screening Program, North Dakota Department of Health.

Sara Copeland, M.D., Assistant Professor, University of Iowa, School of Medicine, Department of Pediatrics. Dr. Copeland is also a consultant for the North Dakota Newborn Screening Program.

Alan Kenien, M.D., Pediatric Endocrinologist at MeritCare Medical Clinic. He also is a consultant for the ND Newborn Screening Program.

Stephen Tinguely, M.D., Chair of Pediatrics University of North Dakota School of Medicine and Health Sciences. Pediatrician, Meritcare Health Systems and co-director of MeritCare Cystic Fibrosis Program.

Family Resource Panel:

Pat Conway, Ph.D., LCSW, (moderator); Director of Family-to-Family, Professor at the Center for Rural Health of the University of North Dakota (UND) School of Medicine and Health Sciences.

Donene Feist, Executive Director and a parent activist of the North Dakota Chapter of Family Voices.

Tammy Gallup-Millner, RN, MPA, Director of the Children's Special Health Services unit, Medical Services Division, Department of Human Services.

Parent Panel:

Gretchen Noah, (moderator), Parent of a child with Smith-Lemli-Opitz Syndrome and lay advocate for Smith-Lemli-Opitz Syndrome Parent Group.

Phil & Dianna Hansen; Parents of three children, one with scoliosis and a chromosome disorder.

Jane & Stephen Nelson, M.D., Dr. Nelson and his wife have four children, one who was diagnosed with Cornelia de Lange Syndrome.

Scott & Robyn Throlson, First-time parents of a child with Primary Carnitine Deficiency.

Janette Venaas-Gilbraith, parent of two children, one with cerbral palsy.

Visit www.medicine.nodak.edu/genetics or www.marknoah.com for conference updates.

Hearing Hoofbeats and Thinking ZEBRAS~
Screening, Testing & Management
of Children with Genetic Disorders
Ramada Inn, Fargo, ND
April 23 & 24, 2007

Registration Type:

- ○ Early Bird Registration (prior to Feb. 28, 2007) $70.00
- ○ Registration (Deadline March 28, 2007) $90.00
- ○ Student Registration (valid student ID required) $25.00

Please check your Monday dinner preference: ○ Greek Lemon Chicken ○ Pork Loin Dijonnaise

Name: _____ Title: _____

Organization: _____

Address: _____ City: _____ State: ____ Zip: ____

Phone: _____ Fax: _____ E-mail: _____

Special Needs/Requests: _____

Name (as you would like it to appear on your badge): _____

Scholarships available to UND Medical Students

Payment Information:

Make checks payable to Division of Medical Genetics. Send the registration form and payment to Attn: Jayne Brown, Pediatrics/Division of Medical Genetics, School of Medicine and Health Sciences Room 5910, 501 N. Columbia Road Stop 9037, Grand Forks, ND 58202-9037

NORTH DAKOTA
MEDICINE
UNIVERSITY OF NORTH DAKOTA SCHOOL OF MEDICINE & HEALTH SCIENCES

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From Tragedy to Triumph

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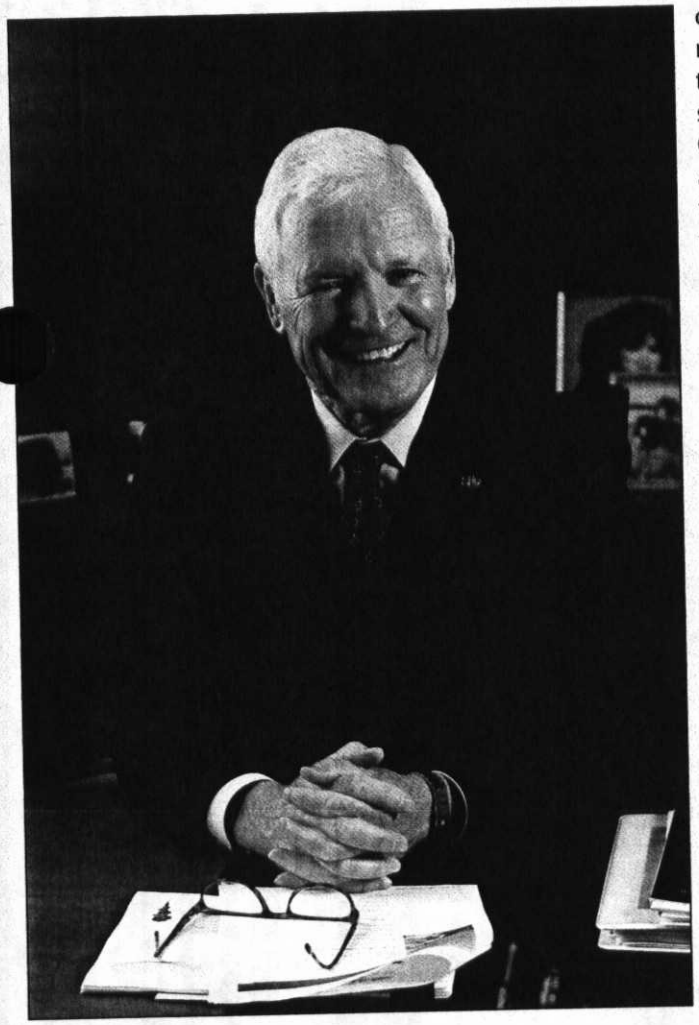
VOLUME 31, NUMBER 5
www.ndmedicine.org

Donor Appreciation Issue

WHEN I MOVED TO NORTH DAKOTA nearly twelve years ago from Kentucky, our executive team set out with distinct and immediate goals for the UND School of Medicine and Health Sciences; one can't help but reflect and marvel at the end of another successful year to appreciate just how far we've come together.

From Dreams to Reality

Our patient-centered learning curriculum continues to be a national role model. Mayo Medical School is revising their curriculum to be like ours and others have taken notice. To be one of only eight medical schools in the entire nation studied by the Carnegie Foundation as an example of how to educate doctors for the 21st century is both an honor and testament to our strong program.



We have been outspoken on the importance of professionalism and offer one of just a few courses on interprofessionalism in the country; others are following our lead.

Our Center for Rural Health is, no doubt, one of the best in the world. We have the only Rural Assistance Center in the nation and over 46 projects underway, connecting resources and knowledge to strengthen the health of people in rural communities.

Progressive research continues in the areas of cancer, diabetes, alcoholism, neurodegenerative disorders, eating disorders, drug addiction, and preventative medicine and behaviors. I am so very proud of our faculty who are not only excellent teachers but al contribute to new knowledge for treatment and prevention of diseases. We have increased outside funding for research from $4-6 million to $16-25 million; attracting excellent faculty and fueling North Dakota's economy.

Moving Forward

Like most deans across the country, a major goal of mine for the next few years is to raise endowment money for the school. Our tuition is now at the national average for public medical schools and student debt for the incoming class upon graduation will average $125,000. We are committed to asking our alumni, friends, foundations and businesses to join us in financial partnerships. **Endowments for professorships, chairs, scholarships and technology will help us retain and attract outstanding students and faculty while minimizing tuition increases.**

So, where does that leave us? With our record of success, we continue t set lofty goals and are striving to be top community-based school in the country. We must assure through private philanthropy that we have the resources to maintain a quality institution and assure future students the outstanding education we continue to deliver. I hope you will join me in this quest.

Best wishes for a happy, healthy holiday season!

H. David Wilson

H. David Wilson, M.D.
Vice President for Health Affairs and Dean

The University of North Dakota School of Medicine and Health Sciences currently provides North Dakota with:

41% of its Physicians
91% of its Physical Therapists
62% of its Occupational Therapists
75% of its Physician Assistants
45% of its Clinical Laboratory Scientists
30% of its Athletic Trainers
88% of its Cytotechnologists

Dornacker, who has been doing locum tenens work at Garrison for the past year got to know the staff at the hospital and found that the "nurses were great, the people were great, the town was great — it was just a great experience."

He and his wife, Kristy, have two children, Alexander, 8, and Cassandra, 6. The family resides in Garrison.

In Valley City, **Jithender Kandimalla, M.D. (Surgery Residency '06)**, a recent graduate of the UND's -year general surgery residency gram has established his practice with MeritCare Clinic Valley City.

Kandimalla earned the medical degree at Osmania Medical College in Hyderabad, India in 1991 and a master's degree in general surgery from Ghandi Medical College in Hyderabad in 1995. He took advanced training in Liverpool, United Kingdom; Glasgow, Scotland; and the Bronx, NY.

Kandimalla specializes in the surgical treatment of a wide range of diseases and conditions which involve the breast, stomach, esophagus, pancreas, gall bladder, lower intestines, hernia and abdomen.

His wife, **Sridevi Gowravaram, M.D.**, a pediatrician, also practices at MeritCare Valley City Clinic. They and their two sons, Akhil and Praval, reside in Valley City.

Essential Element

"We know that our state's medical school has made a huge difference on impact of our entire system," says **Long**, chief executive officer at West River Health System (WRHS) in Hettinger.

The extraordinary medical center, where 15 physicians practice, has benefitted greatly from the infusion of UND medical grads who years ago built it into a premiere rural health care facility.

An important teaching site for UND medical students, the WRHS maximizes its time with students to begin to interest them in a possible future career based in Hettinger.

"It's a good experience for students to see us, and consider us in the future," he says.

"UND is a prime source of recruiting for us. It's a prime source for information and support through the Center for Rural Health."

"We give a lot of credit to the existence of the medical school for our success."

That success has spurred the WRHS to launch a $7 million building and renovation project, which is expected to get under way in earnest next spring.

"The UND School of Medicine and Health Sciences and its graduates are imperative to the future of delivery of health care in the state of North Dakota," says Kim Miller, president and CEO of Mercy Hospital in Williston. People who have grown up here and who know the state are more likely to practice here and to stay here. "It's so critical."

-Pamela D. Knudson

"Its graduates are imperative to the future of delivery of health care in the state of North Dakota,"

Kim Miller, president and CEO of Mercy Hospital in Williston

Needle in a Haystack

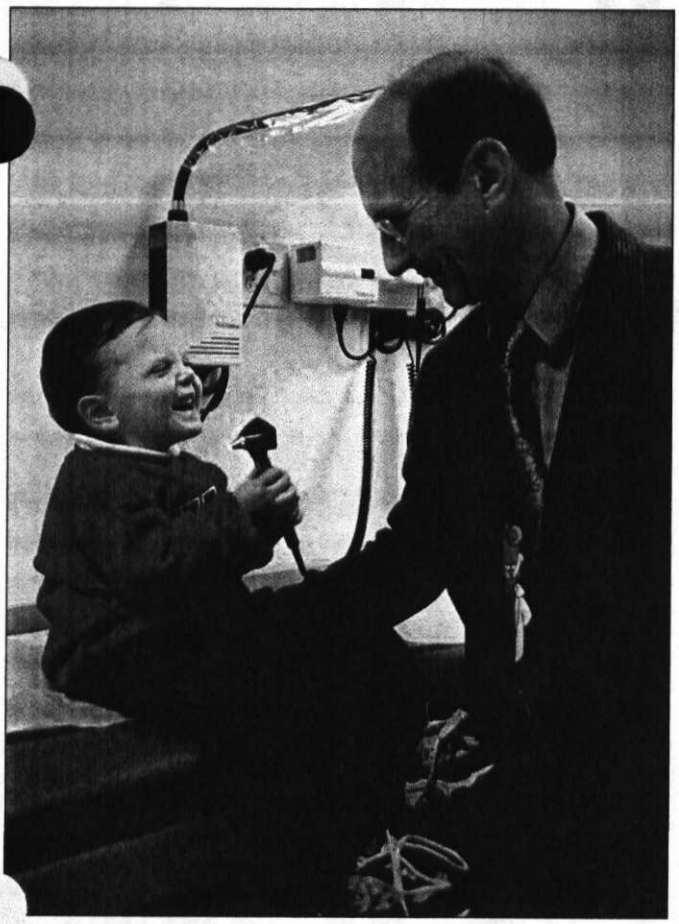
Geneticist Gives Family Answer to Elusive Diagnosis

WHEN HER SON, MARKIE, WAS STILL an infant, **Gretchen Noah** of Fargo knew something was wrong.

"I have three older children and I know what a child does at what stage," she says.

The family consulted physicians in Fargo and Rochester, MN.

"We went through many eyes, and they'd say, 'he's fine; he's just tiny,'" she says. People told her and her husband **Mark Noah, BSPT '87**, 'you've been to Mayo; let it rest.'"



Professor John Martsolf, M.D., made "a very good call" by recommending that Markie Noah (left) be tested for a rare genetic disorder, Smith-Lemli-Opitz Syndrome.

But Gretchen could not be placated.

"It's lonely at undiagnosed. It's frustrating."

Markie was not correctly diagnosed until **John Martsolf, M.D.**, professor of pediatrics, Grand Forks, recommended the child be tested for Smith-Lemli-Opitz Syndrome (SLOS), a genetic disorder that occurs in one of 40,000-60,000 births and results in the body's inability to properly metabolize precursors of cholesterol.

In the ten steps the body uses to produce cholesterol, Markie "does the very last step wrong," Gretchen says, sounding every bit as knowledgeable and well-read as the health professional. Because the body is missing a specific enzyme necessary in the production of cholesterol, the result "is toxic to the brain" and leads to various abnormalities.

"He had a number of the symptoms," she says. "He had global delays (developmental delays spanning many areas), a heart condition, a pushed-up nose, his thumb and first finger are dwarfed... (yet) he's one of only 10 percent who does have a regular cholesterol level," making his a very mild case and therefore difficult to detect.

In the fall 2004, at the age of 11 months, Markie was brought by his family to see Martsolf, North Dakota's only clinical geneticist, to help unravel the mystery.

Martsolf recommended the family seek a second opinion and requested tests be conducted by laboratory scientists at Mayo, where Markie's results showed slightly elevated levels of a particular cholesterol precursor that suggests the existence of SLOS.

Markie's "bloodwork was so mild that they wouldn't positively" diagnose the syndrome, Gretchen says. The family went further to seek advice from the Kennedy Krieger Institute at Johns Hopkins University School of Medicine in Baltimore where more elaborate, sensitive tests were conducted. Eventually they were invited to the clinic of **Forbes Porter, M.D., Ph.D.**, an internationally recognized specialist in this field at the National Institutes of Health in Bethesda, MD, where the SLOS diagnosis was confirmed.

Markie has "one of the mildest cases (the doctors) have ever seen," Gretchen says. "Dr. Porter told us it was 'a great find' for Dr. Martsolf to have spotted it, and that even he, Dr. Porter, would have had a tough time seeing it."

Generally, "pediatricians, ologists and developmental atricians tend not to recognize SLOS in the mild form," Forbes says. "The mild presentations are the hardest to diagnose since they do not have the classical appearance, so this was a very good call by Dr. Martsolf."

"Children with SLOS have autistic features, thus understanding the pathophysiological processes that underlie SLOS could provide insight into a more common disorder such as autism," he explains. "If there is a connection between cholesterol metabolism and autism, understanding the pathophysiological problems in mild cases of SLOS will potentially be very helpful."

Forbes "was just giddy about Markie," Mark recalls, because his case "may help drive the treatment for thousands of kids with autism," the focus of a very large, new initiative. Because symptoms of autism and SLOS are similar, there's speculation that me children with SLOS may be rrectly diagnosed as autistic. Why is it so important to have the correct diagnosis in such cases?

According to Martsolf, "You can't really talk to the parents about what to expect and about the chances that it would occur again in future children until you have the right diagnosis."

"As awful as the syndrome is, it's so much worse to be undiagnosed," Gretchen says. "We slipped through every crack... It took a geneticist to say, 'yes, he has the almond eye, he has the pincher grip.'"

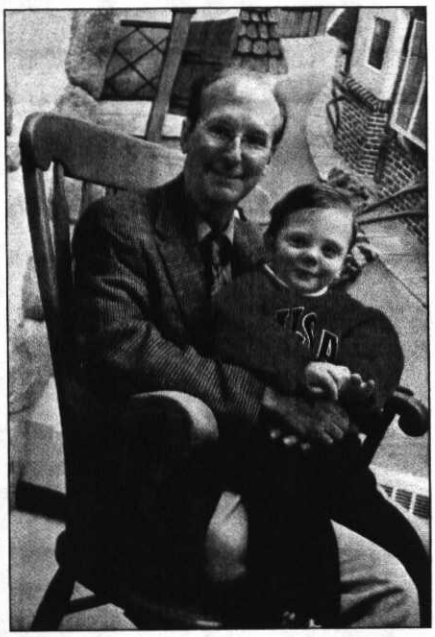
"We're very, very fortunate. When Markie was first diagnosed, he was severely delayed. Now everything's going great. He runs, he talks, he's on to four-word sentences – and we expect great things from him."

"Our son wouldn't have been diagnosed (with SLOS) without Dr. Martsolf," Mark says.

"I'm so unbelievably grateful that Dr. Martsolf picked it up," says Gretchen. "I'm so grateful to him for a number of things *he did right*.

"I'd love for everybody possible to know that we have an excellent geneticist!"

-Pamela D. Knudson



With Dr. Martsolf's (above) help, "Now everything's going great" for Markie, says Markie's mother, Gretchen Noah of Fargo

WEB EXCLUSIVE: visit www.ndmedicine.org to learn more about Markie Noah

Pediatrics Department to Host Genetics Conference

The Department of Pediatrics has received a $10,000 grant from the Dakota Medical Foundation to conduct a conference on genetics and health care next spring in Fargo.

The grant will be used to organize and present the conference, "Hearing Hoofbeats and Thinking Zebras: Screening, Testing and Management of Children with Genetic Disorders," set for April 23-24 at the Fargo Ramada Inn.

Sponsored by the Division of Medical Genetics, part of the pediatrics department at the UND medical school, the event is intended for primary health care providers, especially family physicians, pediatricians, nurse practitioners and physician assistants.

It will focus on North Dakota's newborn screening program as well as the diagnosis, treatment and management of infants who have been identified as having Smith-Lemli-Opitz Syndrome (SLOS) or other metabolic disorders.

By law, every infant born in North Dakota is screened 37 disorders, some of which "are difficult to treat and some t be treated quickly" to avoid sickness or death of the child or potentially serious, lifelong consequences, according to **John Martsolf, M.D.**, professor of pediatrics and director of the Division of Medical Genetics at the medical school.

"It is important that front-line, primary care health providers know what to do if they have a patient who's been detected with a disorder from the newborn screen," he said. "Proper emergency management of children with metabolic disorders is critical."

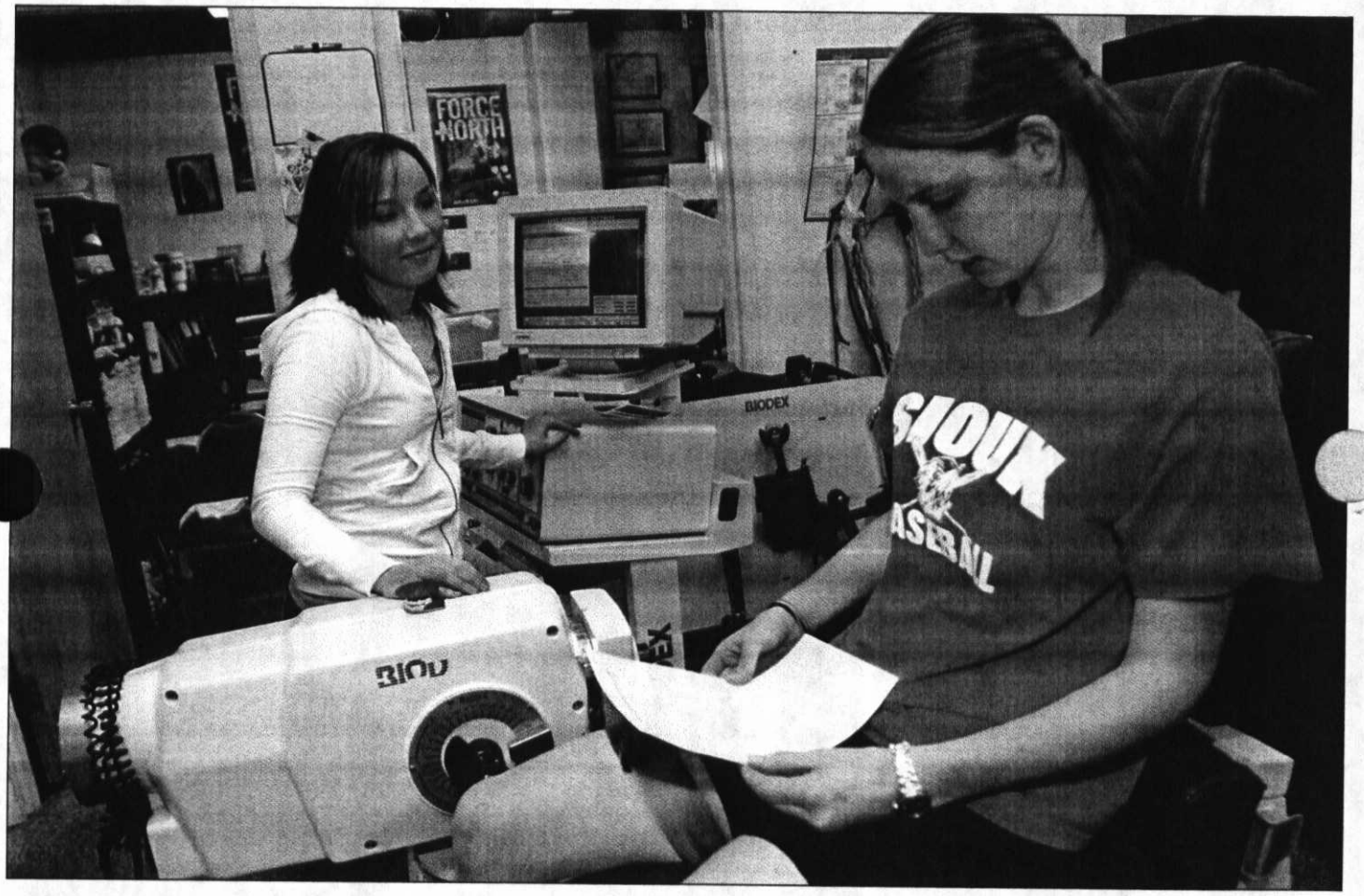
Martsolf, North Dakota's only clinical geneticist, says conference participants will also explore "how the state's newborn screening program is working and how the follow-up has gone," he said. The event also will provide "a forum for discussion of the resources available in North Dakota for these children."

The title of the conference, "Hearing Hoofbeats and Thinking Zebras," refers to the need for health care providers, when presented with common signs and symptoms, to think of the unusual or uncommon possibilities, Martsolf said.

WEB EXCLUSIVE: visit www.ndmedicine.org for more information or to register

From Tragedy to Triumph

Athletic Training Students Experience Continuation of Care through UND Center for Sports Medicine



Athletic training student Brie-Anne Woitas measures the strength of a patient's leg using an isokinetic testing machine, one of the many pieces of rehabilitation equipment students become familiar with while doing rotations at the UND Center for Sports Medicine.

FEW KNOW OR EVEN THINK ABOUT what happens after an athlete is injured during a game. No matter the side, the crowd supplies the requisite applause as the hobbling competitor is helped to the sidelines by the athletic trainer. However, the athletic trainer's work does not end there. In the weeks, or even months, ahead, both the athlete and the trainer work hard to get him or her back in the game.

The Division of Sports Medicine within the Department of Family and Community Medicine at the University of North Dakota (UND) School of Medicine and Health Sciences has been offering a Bachelor of Science in Athletic Training degree since 1991.

According to the National Athletic Trainers' Association, certified athletic trainers are health care professionals who specialize in preventing, recognizing,

#3

Senator Tim Mathern
Human Services 3/21/07
9AM
Red River RM

Hello,
On behalf of the Division of Medical Genetics, located at the School of Medicine & Health Sciences at UND, I would like to state we have four confirmed cases of Smith-Lemli-Opitz Syndrome diagnosed since 1983. We have three patients with a last known address within North Dakota and one in Minnesota. For your information, John T. Martsolf, MD is the only geneticist in North Dakota, therefore the statistics concerning patients seen in North Dakota by a geneticist would be maintained in our office. Unfortunately, we cannot account for the number of North Dakotan residents seen for genetic appointments outside of North Dakota.

I would also like to inform you of a conference we are putting on April 23 & 24, 2007 in Fargo. Monday morning we will have Dr. Susan Sparks talk about Smith-Lemli-Opitz Syndrome, following this we will focus on North Dakota newborn screening by utilizing a panel discussion and then the emergency treatment of metabolic disorders with Dr. Cheryl Greenberg. We will follow Tuesday with Dr. Bryan Hall talking about dysmorphic features, Dr. Martsolf will talk about the process of making a referral for genetic services, as well as a parents discuss their process through the diagnostic system. For more information about the conference you may visit our web site at www.medicine.nodak.edu/genetics.

We are currently in the final stages of writing a State Genetics as recommended by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services. The goal of the state genetics plan is to write a working plan to address the genetic needs of the North Dakota citizens.

As genetics is integrated into public health, all public health programs need to be strengthened. This requires a commitment to sharing resources to address and identify gaps and encompass integration of population-based screening, genetic risk assessment, health promotion and disease prevention strategies within chronic disease, cancer, environmental health and other programs. To foster the integration, many states have developed state genetics plans. The _intent_ of a state genetics plan is to enhance genetics into public health and health care practices. The health status of North Dakotans should be optimized by using genetic/birth defect services. Genetic medicine should also improve public health care and social services systems within the state. Therefore, we would love to have legislators input and invite you to join us April 22 at the Ramada Plaza from 5:00-6:30.

I look forward in hearing from each of you regarding your interest.

Sincerely,
Jayne Brown - Division of Medical Genetics - UND

Markie's journey

Metabolic disorder presents development challenges

By Dave Kolpack, The Forum
Published Sunday, November 19, 2006

Gretchen and Mark Noah have big plans for their son, Markie. In their hearts, they know he will make a difference in the world, that he will do great things.

For now, the Fargo couple is happy to hear him laugh and see him act like a rambunctious 3-year-old, riding around on his bright green tractor with yellow trim or getting into scuffles with his four sisters.

The Noahs knew early on that their boy was different. Special.

He couldn't sit or crawl. He was so sensitive to sounds he covered his ears during his sister's hockey games. He would push the same button on his tractor over and over. It would play the theme from "Green Acres." Over and over. While children his age could say one or two words and understand simple instructions, Markie was lost in silence.

Photo caption: Markie Noah, left, plays with toys as his mother, Gretchen, and sister, Katherine, relax recently in their south Fargo living room. Markie has been diagnosed with a genetic disorder known as Smith-Lemli-Opitz syndrome. Associated Press

Not until last spring was Markie diagnosed with Smith-Lemli-Opitz syndrome (SLOS), an inherited metabolic disorder that blocks the body's ability to produce cholesterol. It's commonly characterized by malformations of the heart, lungs, kidneys and

gastrointestinal tract. Children with SLOS have developmental disabilities, behavioral issues and, in the most severe cases, profound mental retardation.

It has no known cure, but there are treatments to relieve some of the symptoms and disabilities.

"Mark Christopher Noah. Healthy Baby!□6lbs 7oz" Nov. 12, 2003.

For the first year in her son's life, Gretchen kept a diary, recording on a calendar the milestones that marked his progress.

His baptism. His first bath. His first taste of solid foods – green beans. A drink of water from his first sippy cup. And on May 21, 2004, his first tooth, "ta-da!"

But tucked between those signposts were worrisome problems.

A day after Markie was born, the obstetrician discovered he had a heart murmur. It shouldn't cause any serious problems, doctors reassured the Noahs. Markie also had a small head, upturned nose, a droopy left eyelid, and a dwarfed thumb and index finger on both hands.

There was more. He had trouble nursing. He had chronic ear infections.

In his first month, for five days in a row, he couldn't sleep and developed colic – that inconsolable crying spell. He didn't babble like a typical baby. He grunted.

The Noahs attended to each illness, unaware that collectively they were an alarm bell for the mental and physical disabilities of the rare hereditary disorder.

Markie first started to communicate with sign language, learning signs for apple, cookie or candy. Later, he used the word "bah" to refer to most items in the house.

"We didn't know if he would ever talk," Gretchen said.

At about 14 months, the Noahs enrolled him in physical, occupational and speech therapy at Pediatric Therapy Partners, a children's clinic in Fargo. There was no emotional connection between the patient and his therapists. They spent hours teaching Markie how to pinch a Cheerio in his thumb and index finger.

"Everything was a struggle for him," therapist Stephen Olson said.

A geneticist at the University of North Dakota suggested Markie could have SLOS, but the doctors the Noahs saw couldn't confirm it. Gretchen pursued the idea on the Internet after talking to a mother she met at a support group for children with heart defects.

"I worry for you. I never want you to be teased or feel different. I want you to be healthy and happy." Jan. 19, 2004.

Smith-Lemli-Opitz syndrome was first described by geneticists David Smith, Luc Lemli and John Opitz in 1964, but it was not until 30 years later that scientists discovered that children with SLOS were unable to make or process cholesterol.

As a result, they have distinctive physical features, including a cleft palate, low-set ears, a single palm crease, webbing between the second and third toes, and some types of genital malformations. A majority of patients have autistic characteristics – they can't maintain eye contact, they avoid showing affection.

Dr. Forbes Porter, Markie's physician and a researcher with the National Institutes of Health, said about 80 percent of SLOS pregnancies never survive to a full term. More than half of the children with the syndrome must be tube-fed for several years.

The disorder affects 1 in up to 40,000 births and is most common in whites of European ancestry.

Markie's case is rare because it's mild.

Before giving birth to Markie, Gretchen had six miscarriages. The Noahs have since learned that their genetic combination led to the disorder and the miscarriages.

Markie's oldest sisters – Margaret, 9, and Kelly, 8 – have tested negative for the disorder; since 5-year-old Mary and 18-month-old Katherine show no signs or symptoms of the disorder, their parents say they can get tested when they're older to learn whether they are carriers of the altered gene.

"We've gone through six miscarriages, so obviously we're not in charge," said Mark, a physical therapist for a nursing home.

Except for the heart condition, Markie's major organs function normally.

Without his medication, though, he will harm himself – uncontrollably biting his shirt collars and slapping himself in the head with both hands, much like the autistic savant Dustin Hoffman played in "Rain Man." During temper tantrums, he throws himself on the ground. These behaviors are associated with abnormal cholesterol metabolism.

Only about 500 people in the United States are living with the disorder, which is commonly misdiagnosed, Porter said.

For women who suspect they are at risk of carrying the altered gene, a prenatal blood test can measure an enzyme important for the body's production of cholesterol.

One problem caused by a lack of the enzyme: Potentially toxic byproducts from cholesterol production build up in the blood and other tissues, according to the Genetics Home Reference Web site of the National Institutes of Health.

"I just know that you are going to change the world. I tell you all the time that you are going to cure cancer ... or make a difference ... or save a life ... or help the needy. You will do great things! God's work. I'm sure of it."

Gretchen made that journal entry on March 31, 2004, when her son was 4½ months old – a full two years before he was diagnosed with Smith-Lemli-Opitz syndrome. It seems prophetic.

Because Markie's case is so mild, Porter believes he makes an ideal candidate for studying the genetic mutations and characteristics of SLOS. How he responds to medication could affect the lives of other children with more severe forms of the disorder and more common ones such as autism.

Porter is treating the learning and behavior problems by trying to correct the biochemical defect in the brain. For the past six months, Markie has been taking three spoonfuls a day of a cherry-flavored cholesterol serum, a standard treatment.

It has made a vast improvement in Markie's speech and behavior, his mother said.

For one, he doesn't have tantrums anymore.

"Out mama," he tells Gretchen when he wants to be removed from his crib. "Oh, thank you, mama," he says after she lifts him out.

Shown a picture of a parrot, he says, "Tah-rock-er-ah-pirt." That means "tropical bird," his mom says.

Markie is no longer enrolled in physical therapy, but twice a week he works on flexibility and strength in his hands, and is learning to use a pair of scissors. He's no longer oblivious to everyone in the room.

"Now he has blossomed into this little boy who is active and runs and plays and laughs and jokes and all those kinds of things," Olson said.

Porter says Markie will always have some degree of learning and behavioral problems but he should have a normal life expectancy.

Mark and Gretchen have hope, and they plan to enroll Markie in preschool next year.