

# Center for Special Children

21ST CENTURY MEDICINE HELPS AMISH  
DEAL WITH RARE, INHERITED ILLNESSES

**T**here is no car in the driveway, neither phone nor electricity in the house. Handmade clothes dry on the line.

It's fall 2018, and La Farge physician James DeLine, MD (PG '83), has brought us to talk with Barbara and Daniel Hochstetler, part of the large Amish community in Wisconsin's Driftless Region.

Six of their 11 children live with sitosterolemia, an extremely rare disease that can cause joint damage, stroke or heart attack, due to accumulations of a plant-based fat akin to cholesterol.

DeLine has practiced family medicine in La Farge since 1983, when he completed his residency in the University of Wisconsin School of Medicine and Public Health's (SMPH) Department of Family Medicine and Community Health. In 2014, he founded the Center for Special Children to care for Wisconsin's large concentration of Amish or Old Order Mennonite people.

Rural doctors pride themselves on being able to treat a wide range of conditions in their patients, but DeLine's practice brings him face to face with many rare genetic

conditions that were present when the Amish and Mennonites immigrated from Europe to America and then Wisconsin.

And that, in turn, has brought DeLine into a close collaboration with specialists at UW-Madison who have developed tests and suggested treatments for some of those conditions, including sitosterolemia.

Quietly, DeLine and the Hochstetler parents recounted how they learned that the family carried a gene for the rare disease. Years earlier, one of their sons had been seen at the La Farge clinic with painful arthritis and large lumps in his limbs.

"Later, when we discovered that a relative of his mother had sitosterolemia, we thought back to this young man, and with some searching, we found him, had gene testing done at UW-Madison, and discovered that he too had the disease," he says.

DeLine's goal in starting the Center for Special Children was to attend to the particular health needs of the Amish and Old Order Mennonite families in Wisconsin. The program exists within the La Farge Medical Clinic, also started by DeLine, which now is part of Vernon Memorial Health Care.



*James DeLine, MD (PG '83), near the clinic he founded to care for the Plain community.*

After beginning medication and changing his diet, the Hochstetlers' son's elbow lumps began melting away, notes DeLine, who adds, "He has had no further arthritis, and his exercise tolerance has improved."

Eventually, with genetic testing at UW-Madison, sitosterolemia was diagnosed in six of the 11 Hochstetler children. Only then did Daniel Hochstetler volunteer that he had heart pain (likely just "age catching up



*The characteristic homemade clothes of an Amish family hang just inside the back door of the Hochstetlers' home.*

with me,” he says) during heavy exertion. It actually was caused by a buildup of plaque in his heart arteries. After starting the same drug as his children, he has improved, though he can “still feel it once in a while if I exert myself,” he says.

### **In the Tradition of Family Doctoring**

DeLine has become an expert in the culture, family relationships and medical needs of the Amish and Old Order Mennonites, sometimes called the “Plain” people.

Amish and Mennonite families avoid technologies that they feel would endanger the social cohesion that is key to their survival. Thus, they do not own motor vehicles or use telephones or electricity in the home.

Although their acceptance of technology is highly constricted by culture and religion, the Plain benefit from DeLine’s hybrid of 19th century rural doctoring with 21st century genetic medicine.

The genetic work has relied on clinicians from the SMPH and on testing at the UW-Madison State Laboratory of Hygiene. The lab has developed fast, low-cost diagnostic tests for more than 30 conditions afflicting Plain populations in Wisconsin.

Vanessa Horner, PhD, director of cytogenetic services and molecular genetics at the State Lab, says that once a test has been developed and validated, it becomes a “clinical assay” that must be performed in a certified laboratory such as hers. “It’s a highly regulated, rigorous testing environment.”

Funding for these tests and related activities came from grants totaling \$800,000 from the SMPH’s Wisconsin Partnership Program.

“Addressing the health care needs of Wisconsin communities is a priority for the Wisconsin Partnership Program,” says Richard Moss, PhD, who chairs the program’s Partnership Education and Research Committee.

“This team’s innovative and successful community-engaged research has resulted in increased newborn screenings and affordable genetic testing that have the potential to spare our state’s Plain families

from fatal medical conditions and costly hospitalizations,” adds Moss.

One newborn screening test created at UW-Madison, for example, detects maple-syrup urine disease, which prevents the normal breakdown of certain amino acids from food. Left untreated, the disease causes toxic byproducts that attack the brain and other organs immediately after birth.

According to Mei Baker, MD, co-director of newborn screening at the State Laboratory of Hygiene, which developed the test, “We make special arrangements for lab testing beyond regular working hours. The midwife collects a blood sample, and a hired driver delivers it immediately to our lab. Six or eight hours after birth, we have results, and clinicians at Waisman Center advise the parents on an appropriate formula to avoid symptoms. This service is free of charge, and you cannot do any better than that.”

Genetic diseases among the Plain arise from “founder mutations” that were present in the Amish and Old Order Mennonites who immigrated to America in the 19th century. A second “genetic bottleneck” occurred among smaller groups that moved to Wisconsin, starting about a century ago.

Most of the genetic diseases DeLine sees can be treated if not cured, he says.

### **Mixing Old and New**

DeLine’s long and deep experience with many Amish families, and his anthropological knowledge of family relationships are part of his doctor’s toolkit, as are home visits.

Christine Seroogy, MD, professor, SMPH Department of Pediatrics, is a longtime collaborator in the effort to bring 21st century health care to Wisconsin’s Plain populations.

“Dr. DeLine talks about how helpful it is to see a child in the home, surrounded by siblings, grandparents, parents,” says Seroogy, who is one of several UW-Madison colleagues who provide outreach clinical services with the Center for Special Children. “It’s been quite an experience, and an honor, to take part in those home visits.”

She adds, “Home visits were not part of my medical training, but it’s how doctors used to practice, and Dr. DeLine still does.”



CLINT THAYER/DEPARTMENT OF MEDICINE

*Christine Seroogy, MD*

When Seroogy began working with DeLine in 2007, one focus was severe combined immune deficiency (SCID, or “bubble boy”) disease. Though fatal, SCID can be detected with newborn screening and treated with a bone marrow transplant. Over the years, she has worked closely with DeLine, newborn screening experts at the State Laboratory, and Plain families to improve SCID diagnosis and treatment.

Whereas with other conditions, a genetic diagnosis can keep patients out of hospitals and help avoid unnecessary, costly tests.

DeLine says, “When we must deliver news about a child with a lethal genetic disorder, if the family knows what’s going on—sad though it is—it’s a gift to the family to take the child home and care for them surrounded by their community and their family.”

He concludes, “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.”



**School of Medicine  
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# Wisconsin Partnership Program Helps Improve Health Care for Amish Infants

As a pediatric immunologist, Christine Seroogy, MD, a professor in the Department of Pediatrics at the University of Wisconsin School of Medicine and Public Health (SMPH), saw tragic and serious medical conditions that faced Wisconsin's Amish and Old Order Mennonite people, collectively called the Plain communities.

Members of the Plain population have an increased frequency of inherited genetic diseases. Such conditions can be fatal or result in permanent disability and costly hospitalizations, but many can be diagnosed and treated if they are detected early through newborn screening. However, not all Plain infants undergo newborn screening. In fact, it was estimated that potentially several hundred Amish babies did not undergo newborn screening in Wisconsin each year, putting them at risk for death or serious disability.

This realization inspired Seroogy and a team of collaborators at the SMPH to work toward improving health care for Wisconsin's Plain families.

Since 2013, the SMPH's Wisconsin Partnership Program has supported Seroogy's efforts. Funding has bolstered the team's work to establish a strong community-academic partnership, enhance trust within the Plain communities, expand newborn screening tests and further the knowledge regarding genetic disorders present in the state's Plain people.

Seroogy initially received a grant from the Wisconsin Partnership Program to improve access to approachable, culturally appropriate, high-quality and affordable health care for Plain children. The project's goals were to increase understanding of perspectives regarding newborn screening, improve community outreach, and implement assays to define genetic disorders and inform the development of low-cost genetic testing.



*The Wisconsin Plain Community Project, led by Christine Seroogy, MD, received a 2017 Community-University Partnership Award from UW-Madison Chancellor Rebecca Blank. Pictured at the awards ceremony are, front row (left to right): Chancellor Rebecca Blank, Mei Baker, MD, Gretchen Spicer, CPM, Ellen Wald, MD, James DeLine, MD (PG '83), Ashley Kuhl, CGC, Leslie Orrantia; back row: Murray Katcher, MD '75, PhD, Christine Seroogy, MD, Greg Rice, MD '00 (PG '08), Jessica Scott Schwoerer, MD '05, Jennifer Laffin, PhD, Kyle Bakkum.*

A second Partnership Program grant supported efforts to improve health care delivery and utilization through community engagement, as well as training of health care workers in western Wisconsin.

These grant activities have resulted in improved newborn screening rates and increased knowledge and identification of genetic disorders in the Plain population. Also, specific variant tests are available for affordable testing for 30 genetic diseases.

"Progress and success are based upon the relationships and trust we've built in this community," says Seroogy.

"When a baby is born with a critical heart condition, and we are able to make a genetic diagnosis in a matter of days, we give the family and doctor a definitive diagnosis that leads to valuable information about life-saving treatments," she explains.

An integral collaborator is James DeLine, MD (PG '83), who founded the Center for Special Children as a dedicated space for the care of children with genetic conditions (see main article). The collaboration continues to improve

the health of Plain children in Wisconsin, and it also has translated to biomedical research projects, informed approaches to medical care for all children and improved educational experiences for many UW-Madison students.

"Funding from the Wisconsin Partnership Program has been invaluable in helping us improve health care delivery for Amish infants," says Seroogy. "What we are learning about genetic disorders and how to treat them gives us knowledge that transcends culture and translates into potential health care improvements for all children in Wisconsin."

Seroogy now is using a three-year Partnership Program grant to expand the Wisconsin Infant Study Cohort, the only farm-based birth cohort study in the United States. This award expands the study to include Amish infants. It will help determine the interaction between the environment and immune function related to allergic diseases, and it may provide a model for early detection and prevention of allergic diseases.