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## FDA reviews potential Niemann-Pick drug

**By Pam Chickering Wilson**  
**Union staff writer**

A Fort Atkinson mother who lost a child to the as-yet-incurable Niemann-Pick Disease C recently testified before a Food and Drug Administration advisory committee, advocating for a potential treatment for this terminal illness.

Barb Vorpahl, whose daughter, Stacey, died of the illness in 2004 at age 19, said that if approved, the drug Zavesca would be the first approved treatment for Niemann-Pick Type C.

Niemann-Pick is a rare disorder that causes progressive deterioration of the nervous system by blocking the movement of cholesterol within the body's cells. Building up in vital organs, it eventually threatens the most basic bodily functions, such as breathing and swallowing.

Zavesca is used to treat Gaucher's disease, the most common of 40 lysosomal storage disorders that include Niemann-Pick.

It has been available for Niemann-Pick patients "off label," but currently is not approved as a treatment for this disease in the United States, explained Vorpahl, board chair of the National Niemann-Pick Disease Foundation. It has, however, been approved for this use in Europe and in four other countries.

Vorpahl and her husband, Gary, joined with other Niemann-Pick families to form the foundation after Stacey was diagnosed at the age of 2. The foundation has now been around for 18 years, and Vorpahl has continued as a board member throughout, serving as chairperson for seven of those years.

Vorpahl explained that Niemann-Pick is a rare and complex disease, so few doctors are familiar with it. The only treatment they have been able to offer in the past has addressed the symptoms, not the underlying cause of the disease.

The Niemann-Pick Disease Foundation first heard of Zavesca a decade ago, Vorpahl said. In the 10 years since, 146 children and adults that the foundation knows of have died from Niemann-Pick Type C.

"This may not sound like many when compared to other well-known diseases, but in our small Niemann-Pick community, with only about 343 known cases, that number is staggering," Vorpahl told

the FDA advisory panel.

Since then, a number of Niemann-Pick patients have had access to Zavesca through clinical trials and have reported very hopeful results. Vorpahl said that although these patients did not experience a cure, Zavesca did appear to slow the "relentless progression" of the disease, giving them months, even years, to enjoy a higher quality of life.

Zavesca now is undergoing review by the FDA committee, which is made up of doctors, specialists and patient advocates. It will report back to the greater body of the FDA on March 10.

"At that time, it could be approved for treatment or there could be an extension if they feel there is not enough data," said Vorpahl.

Vorpahl testified before the advisory committee on Jan. 12 during a full-day review process. The review started with a presentation by the president of the biopharmaceutical company Actelion Pharmaceuticals Ltd., who outlined the findings of the clinical trials. Then FDA representatives had the opportunity to ask questions and express their concerns. Finally, in the afternoon, one hour was allowed for public comment, with each speaker allowed seven minutes.

"I came as a representative of our foundation," Vorpahl said, referring to the National Niemann-Pick Disease Foundation, based in Fort Atkinson.

Two other families also spoke.

"The testimonials from families were so important to show how these children are doing in comparison to families whose children are not on the drug," Vorpahl said.

"We had children in the audience who had seen no decline in three to four years," she said. "Zavesca is greatly increasing their quality of life."

Vorpahl explained that Niemann-Pick follows a different progression with each child, so it is particularly challenging to demonstrate the kind of concrete evidence the FDA seeks in order to approve a treatment.

"Generally, the committee would look for improvements, but in Niemann-Pick, 'no decline' is an improvement," she said.

In these families' experience, she said, Zavesca is holding off the progression of the disease, and patients are even recording some small gains in swallowing and walking.

"You can't imagine our excitement when we started hearing from parents that they were actually seeing improvements in the symptoms of this disease," Vorpahl told the committee Jan. 12. "We were overjoyed to hear that in siblings and others being treated with Zavesca early enough it was actually holding off the symptoms ... This drug is making a real difference."

She contrasted that experience with those of patients like Stacey, for whom it was too late to benefit from this promising drug.

"With the diagnosis of NPC, you are thrown into a world of the unknown," Vorpahl said. "A world of wheelchairs, braces, feeding tubes and trachs; constant battles with insurance companies, medical specialists, school systems and social services."

She described "watching your child choke on each bite they take, sleepless nights suctioning your child so they won't suffocate in their own secretions, holding them close as you wait for another seizure to subside, seeing them stiffen again in pain but not being able to tell you what hurts.

"As a parent, I know the feeling of helplessness watching your child deteriorate before your eyes (and knowing there is nothing you can do to stop it," she said.

"The hardest part for me is having families call the National Niemann-Pick Disease Foundation after they've received a diagnosis, saying, 'What do we do?' and not being able to offer them a cure or even a treatment.," Vorpahl said. "Now, we may have something to tell them that would offer hope."

Each year, Vorpahl said, she has had to steel herself to witness Niemann-Pick children losing ground and to be there to comfort the families whose children have died.

"In the past few years, this is starting to change, thanks to Zavesca," Vorpahl said. "Many of the children and adults that have access to Zavesca are now at least 'holding their own' against NPC ... We believe Zavesca is making that difference."

The drug costs approximately \$159,000 per year, Vorpahl said, noting that FDA approval could lead to insurance coverage that would enable the average family to be able to afford this drug. Without this approval, this treatment would likely remain out-of-reach.

Vorpahl said she's "cautiously optimistic" for positive results come March and she has great hope for what Zavesca can offer Niemann-Pick families in the future, especially in combination with other drugs.