Climbing 'Jacob's Ladder' -- Couple Crusades to Save Their Son

Determined to Succeed Where Science Has Failed, Parents Close in on a Treatment

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Feb. 27, 2007 — - Andrew Trossman and Marcy White were watching a few little mice very intently.

"We are counting on them, and Jacob is counting on them," Trossman said.

Jacob is the Toronto couple's 4-year-old son, and since birth he lived with strange symptoms that no one seemed able to identify. Instead of crying, he shrieked like a donkey, and his eyes would twitch uncontrollably.

It was only through an extraordinary coincidence that Trossman and White were set on the path to discovering that their son suffered from a rare genetic brain disorder, and that he had, at most, 20 years left to live.

What followed is straight out of the film "Lorenzo's Oil." Trossman and White were not experts in biomedical engineering, but the doctors they consulted seemed to have given up, so they set out themselves to find a treatment for whatever it was that ailed their son. And just this month, promising results from those very important mice led them to believe they might have had a breakthrough.

Marcy White holds her son Jacob. Jacob has Pelizaeus-Merzbacher disease, or PMD, a rare genetic brain disorder. The disease affects one in 300,000 children worldwide, mainly boys. (ABC News)

The Child Who Could Not Cry

From the first days after Jacob's birth, it was clear he would not have an ordinary life.

"The only thing that really, really still stands in my mind is just hearing somebody say, 'I've never seen this before,'" White recalled. "There is this little baby … with tubes [in] him, in the first, what is it, half hour of his life."

Jacob never cried like a normal baby cries. Not once, because he couldn't. His sound was something else.

"They call it a strider, but it kind of sounds like a donkey," Trossman said. "A very striking sound."

Both of Jacob's vocal cords were paralyzed. He was in and out of hospitals, and for 10 months, doctors could not diagnose what was wrong.

"He just had everybody stumped," White said.

White and Trossman said no one was willing to try to solve Jacob's problem, and they were told they would never know -- an "unacceptable" possibility, they said.

"We had to find out what was wrong," Trossman said. "Because if you don't know what is wrong, you can't fix it."

Earning a 'Ph.D. in Jacob'

So, White and Trossman began trying to figure it out themselves, reading everything they could find. They talked to any expert who would listen, until, in a way, they became the experts. White even audited medical classes at the University of Toronto. She and Trossman said they've earned an unofficial degree -- they call it a "Ph.D. in Jacob."

After months of research, finally a break: White uncovered that her family had a history of childhood illness going back generations. She dug up medical records of her mother's brothers -- uncles she had never met because both died before their second birthday.

"I got shivers when I saw it," she recalled. "It talked about paralyzed vocal cords … recurrent vomiting, nostagnis, which is when the eyeballs shake. … It was a perfect match."

But the clincher was an article buried in a medical journal from almost 25 years ago. It described in great detail cases similar to Jacob's.

"They handed me an article and said, 'You know, we came across this article, we think it is about your family,'" said White.

Of course, White thought "your family" meant a family like hers; but in fact, the family was hers.

"We actually saw there was a family tree, and there is White and that was just unbelievable," Trossman said.

A New Place to Start

Using this new research as a guide, doctors took another set of blood tests. The tests indicated that Jacob had Pelizaeus-Merzbacher disease, or PMD, a rare genetic brain disorder; White is a carrier. Worldwide, the disease affects one in 300,000 children, mainly boys.

PMD is degenerative, and there is no treatment or cure. And in severe cases like Jacob's, children have only 15 to 20 years to live.

"We have a huge deadline," Trossman said. "We wanted to get something in him before he turned 5, because then we would have the benefit of any additional neurological development to carry him for the rest of his life, and it has to."

So, again, White and Trossman hit the books. They traveled to medical conferences, and even visited a lab in Germany in search of answers. They heard about an experimental drug that acts as a myelin sheath replacement, but it wasn't being considered for PMD patients.

Help in the Laboratory

The experimental drug was being tested in clinical settings for patients with spinal injuries, but Trossman and White thought it might work for PMD and it might help their son. They enlisted the help of Dr. Jim Garbern from Wayne State University in Detroit.

"Marcy and Andrew are some of the sharpest people I have met in life, period," Garbern said. "They are crackerjack smart about the latest finding in this specific disease and biology, and are self-taught, though this isn't their area of education."

The couple convinced a drug company to allow Garbern to test the drug on mice with a genetic disorder similar to PMD. The affected mice shake violently, like the eyeballs of those who have PMD.

Going into the mice test, White and Trossman were optimistic.

"It's really exciting. I'm thrilled to be here," White said at the time. "But a little bit nervous, because we've got lots riding on this."

But there were no miracles that first day. The mice kept shaking, just like before.

'The Miracle That We Were Hoping for'

Months went by, and Garbern continued his research. In the meantime, Andrea Bocelli -- Jacob's favorite singer -- was in the Toronto area for a concert. With the help of ABC News,

the family reached out to the concert director and he agreed to let Jacob come to a rehearsal. They sang all the way there, and even got to meet Bocelli backstage.

It was a joyous moment, but through the holidays they received little news from Garbern.

Then a few weeks ago, Garbern sent "Nightline" a DVD with some results of the trials. We brought the DVD to Toronto and showed it to White for the first time.

Garbern had upped the dosage of the drug and injected the mice sooner after birth. It seems to have had a dramatic effect on one of the mice. Once shaking violently, the mouse was now walking, even running. Trossman and White were overwhelmed.

"We just need to get that drug into Jacob," White said.

"I think it doesn't take a brain surgeon to see an improvement," her husband added. "This is the miracle that we were hoping for that first day."

'Pushing as Hard as We Can'

The trials are still in the very early stages; rigorous clinical testing must take place before Jacob can get access to the drug. For Trossman and White, there is hope, but no time for complacency.

"We are pushing as hard as we can," Trossman said.

"As hard as we know how to do," his wife added.

And White insisted that exhaustion is not an issue.

"It doesn't matter," she said. "Until we can get that drug in Jacob, we can't stop. It is not an issue; that is what we are doing. If we can find a doctor here who can help us, here in Canada, [to] get access to this drug and get this drug in Jacob, that is when we can feel like, 'Yes, we have done something' … unless we do something that actually does help him, then we haven't done anything, or we haven't done enough."