

Pompe Patient Profiles

Megan Assink



Megan was one of the first children in the world to be enrolled in clinical trials for Myozyme[®] (alglucosidase alfa), then an experimental treatment for Pompe disease. Megan was the second child in her family diagnosed with the rare genetic disease. Her older sister Kelsey died at age nine from the progression of Pompe disease. Shortly after Kelsey's death, Megan, still an infant, was diagnosed with the same disease that claimed her sister's life. As with Kelsey, there were no treatments available for Megan at the time. Doctors recommended a special, high protein diet to hopefully help slow further muscle wasting.

In the year following her diagnosis, Megan developed cardiac and respiratory problems and showed signs of delayed development, including difficulty crawling, sitting and standing up. Doctors predicted Megan would spend most of her life in a wheelchair and on a ventilator like her sister Kelsey.

But Megan was fortunate. Megan's parents found out about a clinical trial for Myozyme, then an experimental enzyme replacement therapy, which had begun at a few hospitals around the world. Eager for any treatment, Megan's mother relocated to Florida with Megan and an older sister for six months so Megan could participate in the research effort. Megan gradually began to gain trunk strength and was able to pull herself up to a standing position on her own. After six months, Megan was able to continue to receive the treatment closer to home in Michigan at DeVos Children's Hospital in Grand Rapids. Within a year, Megan was able to walk without any support.

Today, at age five, Megan is an active little girl who loves to play with her sister and brother, Hope and Tyler. She continues to receive an infusion every two weeks, which is supplemented by physical, occupational and speech therapy. Her echocardiogram results, a test to assess whether heart muscle has become too thick, show her heart as normal. "We are amazed and truly blessed by her progress," says her father, Greg Assink.

Lateef Murdock



Lateef Murdock, an African-American musician, songwriter and music entrepreneur from the Detroit area, was diagnosed with the late-onset form of Pompe disease in 1991, when he was 22 years old.

Despite suffering from a rare genetic disease and being confined to a wheelchair, Lateef lives independently, runs his own production company, and is planning a move to Los Angeles to pursue his dream of writing and producing music full time.

Like many people with late-onset Pompe, Lateef's initial symptoms appeared to mirror other more common medical conditions. When Lateef was 19, he felt minor symptoms of muscle weakness and fatigue, which he first chalked up to being out of shape. Thinking he just needed extra time in the gym, he began to work out. About a year later, after exercise failed to improve his symptoms, Lateef sought medical attention. An initial biopsy was inconclusive, and it took over a year and a half of medical tests before his doctors diagnosed Pompe disease.

Over the years, Lateef's symptoms got worse. By the time he reached his early 30s, he had lost the ability to walk up a flight of stairs. In 2001, Lateef had lost a great deal of strength and was forced to admit he needed a wheelchair. He had the chair for a year before ever using it, joking that it "made a nice coat rack." He began falling frequently and visits to the emergency room became so common, the ER doctors were on a first-name basis with him. Eventually he accepted the fact that he needed the wheelchair. Lateef says he moves a little more slowly now but he's as enthusiastic as ever. He is steadfast in his refusal to give up his independence. "I'm big on doing things in my own time and in my own way," he says.

Pompe disease is not widely known within the African-American community, though African-Americans are eight times more likely to develop Pompe than Caucasians.

Brian White



Until he was 37, Brian White led a seemingly normal life in Northern Virginia, playing sports, traveling regularly, working a full-time job and raising two boys with his wife, Sarah. With the onset of symptoms including difficulty breathing and severe fatigue, he began a 2-year ordeal to find out what was wrong. Doctors initially thought he had narcolepsy, but when standard treatments for that condition failed to solve the problem, further tests revealed he had Pompe disease. Doctors told him that his health would continue to decline.

For his family and for himself, Brian realized that he could not just sit and continue to deteriorate. So he sprung into action. Brian spearheaded efforts to bring together the small and diverse patient community through a Pompe Disease Conference Call program. He organized one of the first-ever Pompe patient conferences where nearly 50 families and experts joined to share information and support. And he took his cause directly to the U.S. Food and Drug Administration (FDA), joining with several other patients to meet with key officials to stress the truly devastating impact of the disease.

A true sports fanatic, Brian continues to play and coach sports with his sons, who are both in elementary school. Though he had to give up two favorite activities, swimming and body surfing, due to his Pompe disease, Brian remains as active as possible – shooting hoops or playing touch football with his boys mostly close to their Fairfax, Virginia, home. “I don’t want my sons to think of me as disabled, and I don’t want this disease to impair my ability to do sports with my sons or have an active family life,” says Brian.

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Not all patients respond in the same manner as those described above. Treatment experiences may vary from one person to another. Please see full prescribing information including boxed warning and important safety information at <http://www.myozyme.com/>