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# Her struggle with 'Extraordinary Measures' disease

## Fight with Pompe began long before diagnosis

By [Peter Rowe](#), UNION-TRIBUNE STAFF WRITER

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Earnie Grafton / UNION-TRIBUNE

Alicia Blackington gives the movie “Extraordinary Measures” high marks but wishes it had focused more on the day-to-day struggles of families battling this disease.



Photo by Earnie Grafton - Union-Tribune

Alicia Blackington is one of only six confirmed cases in San Diego County of Pompe disease, the disease at the center of the new Harrison Ford-Brendan Fraser movie, “Extraordinary Measures.” She works two days a week at Aquatic Adventures in Mission Bay.



Brendan Fraser stars as John Crowley and Sam Hall plays John Crowley Jr. in CBS Films’ “Extraordinary Measures.”

Alicia Blackington is a rare individual. That hasn’t always been a blessing.

For years, the Coronado native suffered from mysterious ailments that left her with weak limbs and breathless lungs. Twice, she was hospitalized and put on life support. Once, doctors told loved ones that she was on the brink of death. Always, her condition baffled the experts.

Finally, she was diagnosed with Pompe disease, a gradual and often fatal weakening of the muscles controlling movement and breathing.

So far, so bad. But she caught a break.

“Don’t worry,” said a doctor at the Mayo Clinic in Minnesota. “There is a treatment coming up in the world.”

That treatment, and the breathtaking road to its discovery, is the subject of “Extraordinary Measures,” the Harrison Ford movie that opened Friday. In the film, John Crowley (Brendan Fraser) frantically seeks a cure

to the disease that is killing two of his children. This ailment is so rare, pharmaceutical companies were not pursuing a treatment. Refusing to accept defeat, Crowley stakes everything on a bold move. The Harvard Business School graduate quits his job, raises more than \$100 million, takes over a biotech company and hires a maverick doctor (Ford) to stop this killer.

On the film and in life, they devised a medication — and an adult version of this drug, Myozyme, has helped to save Blackington's life.

“It's not foolproof, not the final answer,” Blackington, 44, said on a warm January afternoon near the University Heights home she shares with her husband, Andy Wagle. “But I don't have the fatigue I used to have, I don't have the need for a nap all the time.”

In 1932, Dutch pathologist Johan Pompe described a condition characterized by an inability to process glycogen, a form of sugar that is normally stored in the body until needed. If it builds up in the body and is not consumed, though, glycogen gradually destroys muscles, including the heart.

While this devastating disease has been documented for decades, until recently many of Pompe's victims suffered without knowing why. The neuromuscular ailment has been, and still is, diagnosed as a form of muscular dystrophy (in fact, the Muscular Dystrophy Association is a major backer of studies of the disease). Often, though, Pompe slipped by undiagnosed, even as its victims' muscles withered and their chances for recovery vanished.

The disease was so unusual — specialists estimate that only one out of 30,000 people develop Pompe — and treatment so elusive, few researchers wanted to talk about Pompe, let alone delve into its intricacies.

“The treatment is very new,” said Jan Panyard-Davis, a clinical nurse coordinator at UCSD Medical Center, where Blackington is one of only five Pompe patients. “And when there is no treatment for a disease or disorder, there's a lot less interest in it.”

Doctors diagnosed Pompe in Megan and Patrick Crowley when they were, respectively, 15 months and 5 months old. Blackington, though, cruised through childhood without serious complaints. As a teen and young adult, she was a competitive equestrian. Later, she and Wagle ran a company that made kayak accessories.

By her early 20s, though, she found it difficult to rise out of a chair or couch. Climbing a flight of stairs was a major task. In 1994, a cold led to a lung infection and three days on a ventilator. Two years later, another cold left her comatose for three days.

“When I woke up, I was a complete invalid,” she said. “I couldn't move my arms or legs.”

She gradually recovered the use of her limbs, but her walk was permanently changed. She now moves gingerly, her legs swinging outward before moving forward. Years later, she discovered that this is called “the Pompe waddle.”

To Blackington and a succession of doctors, nurses and specialists, it was all very mysterious — and dismaying. “It's very frustrating for patients to know they have something seriously wrong with them and not be able to put a name to it,” Panyard-Davis noted.

So when the Mayo Clinic in Rochester, Minn., finally attached a name to Blackington's ailment in 2001, she was relieved. And staggered by the challenges still ahead.

Thanks to the extraordinary measures taken by the Crowley family, cold season is no longer a potential death threat to Blackington, who literally breathes easier. With renewed energy, the 5-foot-2, 95-pound

dynamo works as a real estate agent; works two days a week at Aqua Adventures on Mission Bay; advises United Pompe Foundation; and assists other patient-support groups.

Still, Blackington needs a breathing machine to sleep; has had to adjust to a halting, sidwinding gait — “I don’t know if I’ll ever ride a bike again, but that’s OK” — and eats a protein-rich diet, limiting the sugars her body can’t consume.

Life is better, though, and future Pompe patients will see even more improvements. Blackington’s diagnosis, for instance, came after a series of muscle biopsies. Today, the disease can be detected through simple blood tests.

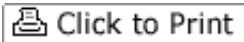
And Pompe is less adept at hiding in the shadows, thanks to “Extraordinary Measures.” Two weeks before the CBS Films production opened, one of the largest online storehouses of information about this disease, [pompe.com](http://pompe.com), experienced a fourfold increase in hits.

While Blackington gives the movie high marks, she wishes it had focused more on the day-to-day struggles of families battling this disease and a little less on the medical teams chasing a cure. But that’s one person’s review.

“My nurses loved this,” Blackington said of the movie. “It shows the dedication and passion of the scientists.”

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