Did Pompe Disease Get A New Champion In President Trump?

By Jamie Wells, M.D. — March 2, 2017

The squeaky wheel often gets the grease but sometimes wheels get an impassioned champion - and that is just as good. That may have happened with Pompe disease when President Trump, addressing a joint session of Congress, highlighted the presence of 20-year old Notre Dame sophomore Megan Crowley, who is afflicted with it.

Pompe disease results from mutations in the GAA gene which result in the inability of the body to break down the complex sugar called glycogen. That resulting buildup, especially in muscles, prevents them from functioning normally.

It's an inherited disease and relatively rare. According to the National Institutes of Health [2], Pompe disease affects about 1 in 40,000 people in the United States while all glycogen storage diseases affect 1 in 20,000. Compare that to the 40,000 women [3] who will die from breast cancer and you can see why it doesn't get as much attention.

These smaller population "orphan Diseases" [4], defined as those that afflict less than 200,000 people across the nation, include conditions like Cystic Fibrosis, ALS, acromegaly (aka gigantism) and Tourette's Syndrome. But they collectively impact 25 million Americans [5], a substantial public health issue. It doesn't take a president. Sometimes national attention comes from other parts of culture. Though Parkinson's disease was long known, it got a highly visible champion when a young Michael J. Fox, in the prime of his career, was diagnosed with it. Since 2000, his foundation has raised and spent $700 million [6], an amount only superseded by the government and private sector.

Perhaps President Trump's mention is a sign that he truly cares about science and health.
research. Not just the government-funded kind, but making treatments easier to get across the spectrum. The big reason why rarer diseases get fewer treatments is because we have a regulatory process that forces business decisions to go for the "home run." With a dozen years and $1.2 billion, almost double what it was 20 years ago due to increased bureaucracy, a drug that has a small market has little chance, even if it works after thorough testing. Even Hepatitis C, which has a huge market, has to have a high price tag because up to 70 percent of the patent period is now tied up getting approval.

There is already a faster route for "orphan" drugs, which helps to bypass the financial obstacles of getting ordinary approval. President Reagan signed the Orphan Drug Act in 1983 in an attempt to incentivize discovery. And to some extent, it has worked. According to the U.S. Food and Drug Administration (FDA) more than 250 drugs “available to treat a potential patient population of more than 13 million Americans” have become available in the three decades since it became law. But is about 9 a year really good? Some have argued that the regulatory process is still crippling discovery because of the government hurdles before human trials can begin. Patient advocacy groups have emerged— especially in these decades— and served to increase awareness while directly advancing the research.

For Pompe patient groups, there may be no better champion than President Trump. But Megan Crowley and her brother (also living with Pompe Disease) had one much closer to home - their father. John F. Crowley’s extraordinary tale of love intermingled with persistence is an inspiration. At the time of their diagnosis, he left his job and singlehandedly started a company in an effort to find a cure. His work was instrumental in the development of a necessary enzyme replacement therapy for this, as well as strides in other rare diseases. Their story was captured in national media and in the film Extraordinary Measures, starring Harrison Ford.

With President Trump and Harrison Ford, you’d think things would be easy. But since I used a sports term earlier I will use one again. You still have to play the game. High profile champions are great for awareness and funding but the slow, thorough business of research continues after the media buzz fades.

That said, an internationally-televised address about the disease is a big step for patients and their loved ones. This attention brings hope about progress toward cures or life-sustaining treatments.

I didn't want to make this too long, so in the next installment --Pompe Disease, Newborn Screening and Inborn Errors of Metabolism-- I will get into the nuts and bolts of glycogen storage diseases like Pompe Disease, and the research hurdles that will need to be overcome.

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