To Whom it may concern,

Hello, my name is Ryan Colburn and I'm 37 years old. Like everyone else, I have a genetic code that is 3bn letters long. And also, like everyone else, I have some typos in that code, 2 of which are in my GAA gene on chromosome 17 (meaning that I have Pompe). This letter is in support of adding screening for Pompe, and other typos we can do something about in newborns.

I hope the medical information provided is clear about the devastating outcome in the infantile onset form of Pompe disease for without treatment and how paramount early screening is for opening up the possibility of an alternate outcome. For me there is no decision required based on this information – it is obvious without any room for interpretation that by adding the screening you are saving lives and with an emphasis for the department of health to prioritize urgent action, even providing an opportunity for these children and their families to minimize the headwind that having a rare disease can have on their lives; and by not doing the screening you are choosing death for these infants before they ever get to blow out a single birthday candle. The data can be loosely viewed as this: before ERT (Enzyme Replacement Therapy) treatment existed (FDA approved in 2006), the lives of infantile onset Pompe patients were transient, over before they started and with heartbreaking, family destroying results, whereas now, as we celebrate each "birthday" since the initial ERT was available, so too do we celebrate the birthdays of infants growing as children with infantile onset Pompe disease. And those who were fortunate enough to find a quick diagnosis are even breathing with their own lungs and walking with their own legs as they play and grow with their peers. It is clear that immediate diagnosis WILL provide a life changing result, and for that newborn screening is clear and required. On the administrative side, the incremental cost to add the test, amortized and shared by payers anyway, is insignificant in consideration of the costs associated with child birth, and avoiding a cycle of poor health.

I felt compelled to write this letter to provide an additional perspective on the importance of newborn screening beyond this obvious severe infantile case. I was conclusively diagnosed with Pompe at 31 years old. It was in a way a fortunate circumstance that I was diagnosed, as I was doing a second round of genetic testing at the end of a specific 2 year all hands on deck effort to figure out "what was wrong with me." It was lucky in that the lab that did this second round of testing had a standard genetic sequencing practice of including "similar" symptom diseases when requests came in to do targeted sequencing. So 2 years into dedicated investigation with primary care physicians, neurologists and neuromuscular specialists we stumbled on an answer. All that after having seen "specialists" for 20+ years, having had numerous surgeries and invasive procedures to try to understand other abnormalities all without discovery - all of which, with hindsight, were unnecessary as the abnormalities were explained by Pompe disease. I recently converted old family video to DVD and watched it, and can see clear as day my struggles getting up the stairs at kindergarten graduation – which gives me a minimum bound of outward symptoms presenting before I was 5 years old.

Knowing what I know now, I wouldn't have wanted to know (or have my family know) in 1984 that I had Pompe disease. My physical challenges and associated social challenges growing up shaped me in a fundamental way into the person I am today…

All of that is also easy to say, as there wasn't a thing I could do about having Pompe disease then anyway – but things are different now. Now there has been an FDA approved treatment for 15 years, with an additional 2 likely approved this year, along with numerous talented people working on expanding the scope and efficacy of new treatments to slow the disease, prevent progression and eventually stop and reverse its debilitating progression. A person born today with my exact circumstances wouldn't have to see their body deteriorate for 31 years before understanding why. They wouldn't have to struggle through their youth with comments about their capabilities. Ultimately they and their families wouldn't have to be passengers in life, they could chose to steer their own ship - the American way. They would be able to receive treatment at the onset of symptoms and without question be far better off physically than I am today. Just as importantly they could link up with doctors and researchers, and, if they chose, proactively participate in efforts to further improve understand about Pompe. This allowing those who experience rare diseases to start a cycle of increased awareness and appreciation for those working to help them. This in turn catalyzes an increase in attention from talented young adults choosing their focus and careers. This cycle accelerates positive change and growth in our ability to support high functioning and healthy Iowans, Americans and Humans.

I didn't have a choice for 31 years, with new born screening I would have that choice today. **Support of this proposal wins at every level, and its even more important with the critical role that Iowa's NBS lab plays for contracting states (with the added benefit of faster cost amortization). This proposal has immediate positive impact as it saves lives of newborns and infants right now, and it also has long term positive return for this and your contact testing states as it enables the health of late onset patients as well as promotes accelerated growth in our technological and medical competency.**

Sincerely,

Ryan Colburn

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37 years old, same as you, but with 2 typo's on chromosome 17