Kara Berasi, Board Member

MACPAC April 2021

MACPAC: Kara, we have unmuted your line, so you can make your comment.

MS. BERASI: Thank you. My name is Kara Berasi. I am a board member of CDG Care, a pharmacist, and also the mother of a child with CDG, or congenital disorder of glycosylation. Glycosylation is a process of adding sugar building blocks called "glycans" to proteins, and all body systems require glycosylation to work normally. Over 400 genes play a role in glycosylation, and over 130 of those genes are known to cause CDG.

There are currently 149 known CDG types with additional types being discovered each year, and to give an idea of how rare these disorders are, the most common form, which is PMM2-CDG, has only about 250 diagnosed patients in the United States.

CDG is usually apparent from infancy. It can be associated with a broad variety of symptoms and can vary in severity from mild to severe and also can be disabling and even life-threatening. In fact, 20 percent of patients die within their first year of life. There are currently no FDA-approved treatments, so supportive therapies like physical, occupational, and speech are the only options for the majority of CDG types. There's only one of 149 types that can be treated with oral mannose, but that has not been effective in the other types.

It can be very difficult to get researchers and manufacturers interested in pioneering innovation for extremely rare disorders like this one, especially when there are so many types with similar mechanisms but different body system impacts. So accelerated approval for us is not a shortcut or a pathway with sub-par evidence. It may be the only way a treatment for CDG could become a reality. So, we do not view the additional rebate as a way to help patients by making treatments more affordable; rather, it's a threat to ever getting treatments developed or tested through to approval.

So, we want to stand with you and find another way to get the post-market studies done that seems to be the apparent reason for considering this proposal. The company has done nothing wrong by using this pathway, and yet they're being punished before they defaulted on commitments. So, I'm sure it's more administratively burdensome to apply these penalties and rebates after they've defaulted, but what isn't burdensome in the world of health care or even more so in the world of ultra-rare that my son and so many others live in every day as we fight for the attention of these big drug companies and a chance for a more typical life?

Thank you all so much for your time and kind consideration today and giving me the opportunity to speak.

CHAIR BELLA: Thank you, Kara.