Kristen Wheeden, Executive Director

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MACPAC: Kristin… you can unmute your line and make your comment.

MS. WHEEDEN: Thank you. Hi. My name is Kristin Wheeden and I serve as Executive Director of the American Porphyria Foundation, and I’m also a mom to a 15-year-old son named Brady, who lives with a type of porphyria called erythropoietic protoporphyria, or EPP for short.

Porphyria is not a single disease but a group of eight inherited genetic disorders that result in an accumulation of porphyrins and porphyrin precursors in the body. Now I’ll not give you an entire lesson or the porphyrias today, because Melanie will be sure to cut me off quickly, but I will share that EPP presents with intolerance to sunlight. Sun and other artificial light causes extreme phototoxicity and burning pain, akin to putting your hand in boiling water. It’s something that no one in their lives should feel. And if treatments all are in place, all should have access.

Patients with extremely rare diseases like the porphyrias may never see a treatment developed without orphan drug act incentives, and pragmatic approaches to FDA approval like the accelerated approval pathway. In fact, the first treatment under the Orphan Drug Act was the 1983 approval for a porphyria treatment called hematin, that is still part of the standard of care to this day.

We are very fortunate to have three approved therapies and one drug approved -- excuse me -- that is currently in a Phase III clinical trial addressing porphyria subtypes. Each of these treatments received one or more FDA designations designed to provide incentives to encourage and aid the development of drugs for debilitating rare diseases, like the porphyrias.

As a parent, I can assure you that we want robust evidence on whether a new treatment is effective, as well as the risks that are associated with it. Accelerated approval is not a shortcut around safety and efficacy. It is essential in its pragmatism for both patients and manufacturers. We agree that manufacturers securing accelerated approval must follow through with FDA-required studies, but disagree that failure to do so in ultra-rare conditions is a problem, much less one justifying a penalty on use of the accelerated pathway.

It is a struggle -- and I will put on my professional hat as well as my mom hat -- it is a struggle to find companies interested in and willing to invest in developing treatments for low-prevalence conditions. Please do not follow through with recommendations that can disincentivize and create another barrier to therapy development for people with rare and ultra-rare diseases like the porphyrias.

Thank you for listening.

MACPAC CHAIR BELLA: Thank you, Kristin.