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Pennsylvania Rare Disease Advisory Council Holds Inaugural Stakeholder Summit

HARRISBURG, PA – The Pennsylvania Rare Disease Advisory Council (PARDAC) today brought together rare patients, caregivers, advocates, policymakers and health care professionals at its inaugural Patient Stakeholder Summit. Held at the Harrisburg Hilton, the summit included the unveiling of key initial findings of PARDAC's ongoing Pennsylvania Rare Disease Needs Assessment survey, gathering Pennsylvania-specific insights from rare disease patients and caregivers from across the Commonwealth, as well as plenary sessions focused on the topics of gene therapy and newborn screening.

"This extraordinary event brought together, for the first time ever, patients, caregivers, clinicians, lawmakers, corporate, and non-profit partners to explore topics important to Pennsylvania's rare community," said Marie Conley, PARDAC Council Chair and Founder of The Conley Cushing's Disease Fund. "Through this summit, we are excited to offer meaningful information, share new insights, and forge stronger relationships that will benefit the estimated 1.2 million Pennsylvanians living with a rare disease."

The event offered a presentation and panel discussion revealing the initial findings from the ongoing Pennsylvania Rare Disease Needs Assessment. The luncheon program was presented by PARDAC Councilmembers Dr. William C. Welch, Chair, Department of Neurosurgery, Pennsylvania Hospital; April Adley, Vice President of Nursing Services for the Children's Hospital and Women Health at Penn State Health along with special guests Acting Secretary of Human Services Meg Snead and Rare Disease Caucus Co-Chairs, Senator Judy Ward and Rep. Barb Gleim.

The presentation focused on four areas based on survey results to date:
Diagnostic Journey and Care
Access to Medications
Impact on their ability to work and/or attend school
Travel for Care

Attendees learned about gene therapy through thought-provoking presentations from Krupa Sivamurthy – Sr. Director, Global Medical Affairs, CSL Behring and Brett Logan - Director, U.S. Market Access & State Government Relations, Alliance for Regenerative Medicine (ARM) and Dan Price, patient advocate. The session was moderated by PARDAC member Patrick Collins – Sr. Director, Healthcare Policy & External Affairs for CSL Behring.

The plenary session on newborn screening was equally informative as attendees heard from Jodie Vento, MGC, CGC - Assistant Professor of Human Genetics and the Genetic Counseling Training Program Director at the University of Pittsburgh; Laurie Varlotta, MD - Chairperson, NBS Advisory Board, Pediatric Pulmonologist and Director of the Cystic Fibrosis Care Center and the Pediatric Pulmonology Fellowship Program at St. Christopher's Hospital for Children, Professor and Department of Pediatrics Drexel University College of Medicine; Nicole Engelhardt, MS LCGC - Genetic Counselor, Children Hospital of Philadelphia; Claire Ellis - Policy Fellow, Everylife Foundation; and Lesa Brackbill - wife, mother, author, and advocate.

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About the PA Rare Disease Advisory Council:

The PA Rare Disease Advisory Council (PARDAC) was created in 2017 through legislation to improve the lives of individuals living with rare diseases and their families throughout Pennsylvania. It is comprised of rare disease patients, caregivers, doctors, nurses, researchers, insurers and representatives from government agencies. The Council aims to be a national leader in transforming the lives of those affected by rare diseases through collaboration, support, education and advocacy. For more information about PARDAC, please visit pardac.org.

About the survey:

The survey is open to rare disease patients residing in Pennsylvania and takes approximately 10 minutes to complete If the patient with the rare disease is under the age of 18, or over the age of 18 in need of assistance, a parent, legal guardian or advocate may complete the survey on behalf of the person with a rare disease. Individuals who want their voices heard can fill out the survey at PARDAC.org. Final results of the survey are expected to be released in early 2023.

The suggested hashtags for the event are #raredisease and #PARareDisease.