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Researchers and Clinicians Meet for First International Summit on Rare Gene Disorder
Collaborations emerge from two-day conference on CAMK2

Washington, D.C. — Thirteen scientists, physicians and parents met in November to exchange findings and plan how to work together to address the acute challenges facing patients with CAMK2, an uncurable gene disorder.

The disorder includes at least four gene mutations whose symptoms often are first observed in infancy. Among the more than 300 individuals worldwide diagnosed to date, symptoms include developmental delay, abnormal motor coordination, severe agitation and behavioral abnormalities, and, depending upon the affected gene, seizures, gastrointestinal issues, neuropsychiatric symptoms and significant heart disease.

"I was deeply moved by the collective dedication evident throughout the event aimed at supporting our children," said Dr. Bonnie Dwyer, executive director of conference host CAMK2 Therapeutics Network, a nonprofit first organized informally in 2019 by parents whose children have the rare disorder.

"The summit turnout — seven physicians and scientists and six parent board members — was gratifying and encouraging for our children's future and the future of the network."
— Bonnie Dwyer, CAMK2 Executive Director

The Network's goals are to develop pharmacologic treatment for symptoms and gene therapies for cure. Families and scientists had met several times before the November meeting, but this was the first summit of scientists, physicians and parents.

Participants were based in the Netherlands, Italy, Ireland, Israel, Portugal and, from the United States, California, Massachusetts, Texas and Washington, D.C. Their two days together resulted in these collaborations:

- A partnership to gather data for a CAMK2 Natural History Study that correlates specific mutations with symptoms.
- A continuing study of how specific CAMK2 gene mutations affect the activity of the protein in the brain that regulates a variety of body functions.
- A collaboration to deepen understanding of how the altered protein affects stem cell signaling.
- A joint effort to encourage trials using stem cells and drug repurposing.

CAMKII is the molecule discovered by Dr. Howard Schulman and Nobel Laureate Paul Greegard 40 years ago, but it wasn't until 2017 that a team of scientists led by Dr. Geeske van Woerden and Sebastian Kury defined the neurodevelopmental syndrome linked to CAMK2 gene disorders.

CAMK2 appears randomly. Vigorous exploration on the condition will contribute to the growing body of knowledge about successful therapies for this and other genetic disorders.

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