Driving Regulatory Progress for Inherited Ataxias: CPTA's Impact in Advocacy and Change

By Alexander Diegel and Terina Martinez

Inherited ataxias are a diverse collection of genetic disorders that are characterized by clinical manifestations like impaired coordination of voluntary movements, such as gait and speech. The clinical presentation is typically associated with atrophy of the cerebellum, a structure located at the base of the brain that maintains balance and regulates muscle movements. Each inherited individual ataxia has a unique genetic cause and spectrum of clinical presentation, which may affect different body systems as a result. For example, spinocerebellar ataxias (SCAs) often include dysphagia (difficulty swallowing), stiffness, hyperreflexia (overactive reflexes), spasticity (unusually stiff or rigid muscles), and some cognitive impairment, while autosomal recessive ataxias often include peripheral neuropathy (when sensing nerves in the body don't function properly) and seizures. However, there are also many additional features specific to each SCA subtype.

C-Path's CPTA Executive Director Terina Martinez (top center) participated in the virtual United Against Ataxia Hill Day, where she met with several Massachuesetts Representatives, including Rep. Jim McGovern's Staffer Bella Edo (bottom right).

Although the underlying pathophysiology and concomitant pipeline of new therapies for the ataxias is expanding, there remain large gaps in our understanding of these diverse disorders. In particular, as novel therapies approach clinical evaluation, there is an urgent need for actionable tools to understand the natural history, progression and variance in these diseases, as well as reliably measuring therapeutic effects in clinical trials that are also meaningful to individuals with ataxias.

C-Path's Critical Path to Therapeutics for the Ataxias (CPTA) consortium provides a neutral forum that brings together experts from across different fields of ataxia research, and drug development to generate actionable tools to accelerate drug development for these conditions.

Tangibly demonstrating its important role in the broader ataxia community, CPTA joined forces with the National Ataxia Foundation (NAF, a member of CPTA) and the Friedreich's Ataxia Research Alliance (FARA) and their grassroots members, supporting efforts like an Ataxia Hill Day advocacy event on September 25 to commemorate National Ataxia Awareness Day.

"Tackling rare neurodegenerative diseases like inherited ataxias is extremely complex and difficult; we need to leverage each and every tool, data point, and strategic partnership possible to catalyze progress," explained CPTA Executive Director Terina Martinez. Advocacy efforts galvanize the entire ataxia community and clearly articulate to federal decision makers and drug developers the fundamental importance of investing in ataxia research and clinical development at the individual, family, and community levels. Together, we seek to disrupt the insidious, generational trauma wrought by inherited ataxia and to de-risk drug development in this space."

CPTA's work extends beyond advocacy and awareness, as the group hosts the largest aggregated and publicly accessible spinocerebellar ataxia database on the planet, which is specifically curated for maximal interoperability to inform regulatory decision-making. The collaborative nature of the program also drives advancement in the analysis of existing and new endpoints and biomarkers, developing a biology-based, integrated disease staging system as a foundational framework for its activities, and the aggregation and analyses of existing clinical data across the ataxias to inform the community's understanding of natural history of the different ataxias.

"By convening diverse stakeholders coalesced around shared goals within a neutral forum, CPTA seeks to stock the proverbial ataxia drug development toolbox with innovative, regulatory-grade tools to treat and potentially prevent inherited ataxias," Terina said. "CPTA is proud to work with the broader ataxia community in these important endeavors."

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