AADCAware™
GLOBAL AADC DEFICIENCY PATIENT REGISTRY

Gaining insights through AADCAware™

AADCAware is an international, real-world, observational study of the natural history of individuals diagnosed with AADC deficiency

Currently, information about the clinical presentation, prognostic factors, treatment patterns, and utilization of healthcare resources of patients with Aromatic L-amino Acid Decarboxylase (AADC) deficiency is limited. In an effort to learn more and help patients in the future, PTC Therapeutics established the AADCAware study to provide a deeper and broader knowledge of the natural history and management of this disorder.

Study objectives:

- Describing the natural history of AADC deficiency in individuals
- Assessing the achievement of motor milestones and changes in motor function over time
- Assessing changes in quality of life and health status over time

Findings may help further explain the natural history of AADC deficiency, allowing the optimization of therapeutic strategies and a physician's decision-making in this context.

Design & duration:

- International, multicenter, longitudinal, real-world, observational study of individuals diagnosed with AADC deficiency
- Approximately 50 individuals diagnosed with AADC deficiency to be enrolled
- Approximately 20 sites from 10 countries expected to participate
- 24-month enrollment period
- Care of participants to follow routine management practice at respective country and clinical sites
- Study will end after enrolled individuals complete a minimum of 5-years follow-up

About AADC deficiency

AADC deficiency is a genetic disease associated with defects in neurotransmitter synthesis that can lead to a manifestation of a broad spectrum of symptoms.[1,2] Mutations in the dopa decarboxylase (*DDC*) gene result in reduced AADC enzyme activity, leading to severe combined deficiency of the neurotransmitters dopamine, serotonin, norepinephrine, and epinephrine.[1-4]

The most common symptoms of this autosomal recessive disease are hypotonia, developmental delay, and movement disorders, especially oculogyric crises.[1,2,5,6]

AADCAware™ participating investigators

Country	Principal investigator	Site
USA	Dr. Timothy Feyma	Gillette Children's Specialty Healthcare, St. Paul, MN
	Dr. Donald Gilbert	Cincinnati Children's Hospital Medical Center, Cincinnati, OH
	Dr. Mered Parnes	Texas Children's Hospital, Houston, TX
	Dr./Prof. Phillip L. Pearl	Boston Children's Hospital, Harvard Medical School
	Dr. Gerald Grant	Lucile Packard Children's Hospital Stanford, Palo Alto, CA
	Dr. Muhammad Zafar	Duke University Hospital, Durham, NC
UK	Dr. Lucinda Carr	Great Ormond Street Hospital
France	Dr. Marine Guichard	Centre Hospitalier Régional Universitaire de Tours
	Dr. Marie Ange Nguyen Morel	CHU de Grenoble
	Dr. Claudia Ravelli	CHU Paris Est - Hôpital Armand Trousseau AP-HP
	Dr. Agathe Roubertie	CHU Montpellier - Hôpital Gui de Chauliac
Italy	Prof. Roberta Battini	Irccs Fondazione Stella Maris, University of Pisa
	Dr. Vincenzo Leuzzi	Sapienza Universitá di Roma
Germany	Dr. Martin Smitka	University Hospital Dresden
	Prof. Regina Trollmann	Erlangen University Hospital
Spain	Dr. Salvador Ibáñez-Micó	University Hospital Virgen de Arrixaca (Murcia)
Israel	Dr. Bruria Gidoni-Ben-Zeev	Sheba Medical Center
Brazil	Dr. Roberto Guigliani	Hospital de Clinicas de Porto Alegre
	Prof. Charles M. Lourenço	Centro Paulista de Diagnóstico e Pesquisa (CPDP)
	Dr. Helio van der Linden	CRER-Centro de Reabilitação e Readaptação
Turkey	Dr. Murat Öktem	Gazi University
Saudi Arabia	Dr. Brahim Tabarki Melaiki	Prince Sultan Military Medical City
	Dr. Musaad Abukhaled	King Faisal Specialist Hospital

To learn more about the AADCAware study and patient eligibility, please email info@aadcaware.com.

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