

## Understanding AADC deficiency through AADCAware™

### The AADCAware study is designed to help the medical and scientific community gain a better understanding of AADC deficiency

Aromatic L-amino Acid Decarboxylase (AADC) deficiency is a rare genetic disorder that affects the brain, causes weak muscle tone, and affects how a child develops.

Because AADC deficiency is rare, there is limited information about this disorder, how it progresses, and how it can be managed. There is also a lack of information about the healthcare resources that patients with this disorder may need.

To learn more about this disorder, PTC Therapeutics has created AADCAware—an international, real-world, observational study of the natural history of children and individuals diagnosed with AADC deficiency.



#### AADCAware objectives

The objectives of AADCAware are to:

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

Learnings from this study may help further explain AADC deficiency and its progression, which could help inform healthcare providers about how to manage and treat children and individuals with this disorder.



#### Participating in AADCAware

AADCAware is seeking to enroll approximately 50 individuals diagnosed with AADC deficiency, observing each participant for a minimum of 5 years. Enrollment will be across about 20 sites and 11 countries.

During this time, participants will receive routine management and care for AADC deficiency in accordance with each site's normal practice. Participants will be examined throughout the duration of the study. While the frequency of exams may vary based on the site's normal practice, participants are expected to be examined approximately every 12 months.

For a healthcare provider to enroll a patient in this study:

- A patient must have a documented diagnosis of AADC deficiency
- A patient's parent or legal guardian or legally authorized representative must be willing to provide written informed consent to participate

Individuals with this disorder who have previously participated in, or are currently participating in, a clinical gene therapy trial for AADC deficiency are not eligible to enroll in AADCAware. Participation in AADCAware does not prevent study participants from entering into a clinical trial or having gene therapy treatment.



## AADCAware™ participating investigators

Country	Principal investigator	Site
<b>USA</b>	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
<b>UK</b>	Dr. Lucinda Carr	Great Ormond Street Hospital
<b>France</b>	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
<b>Italy</b>	Prof. Roberta Battini	Irccs Fondazione Stella Maris, University of Pisa
	Dr. Vincenzo Leuzzi	Sapienza Università di Roma
<b>Germany</b>	Dr. Martin Smitka	University Hospital Dresden
	Prof. Regina Trollmann	Erlangen University Hospital
<b>Spain</b>	Dr. Salvador Ibáñez-Micó	University Hospital Virgen de Arrixaca (Murcia)
<b>Israel</b>	Dr. Bruria Gidoni-Ben-Zeev	Sheba Medical Center
<b>Brazil</b>	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
<b>Taiwan</b>	Whu-Liang Hu, M.D, Ph.D	National Taiwan University Hospital
<b>Turkey</b>	Dr. Murat Öktem	Gazi University
<b>Saudi Arabia</b>	Dr. Brahim Tabarki Melaiki	Prince Sultan Military Medical City
	Dr. MUSAAD Abukhaled	King Faisal Specialist Hospital



Please talk to your child's doctor if you would like to learn more about AADCAware or are interested in having them enroll your child in the study.

