# A Two-Part, International, Real-World, Observational Registry of Participants Diagnosed with Aromatic L-Amino Acid Decarboxylase Deficiency (AADC-d) With or Without Treatment With Eladocagene Exuparvovec (PTC-AADC-MA-406)

**First published:** 16/06/2023

**Last updated:** 25/06/2025

Study Ongoing

## Administrative details

**EU PAS number**

EUPAS105422

**Study ID**

105423

**DARWIN EU® study**

No

**Study countries**

- Brazil
- France
- Germany
- Hong Kong
- Italy
- Saudi Arabia
- Spain
- Türkiye
- United Kingdom
- United States

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**Study description**

This is a 2-part, international, multicenter, longitudinal, real- world, observational registry of participants diagnosed with AADC-d. Part A of the registry is to observe the natural history of AADC-d in participants who receive standard of care (SoC), with the intention of gathering data about the AADC-d diagnosis, disease progression among other aspects registry that seeks to describe the natural history of AADC-d (Part A). Part B of the registry aims to evaluate the long-term effectiveness and safety of eladocagene exuparvovec in participants treated for AADC-d. By involving sites who manage AADC-d from different regions of the world which reflect different medical practices and diverse patient populations, this registry will provide a deeper and broader knowledge about the natural history and management of this condition. Furthermore, clinical outcomes resulting from this natural history registry may be compared with outcomes from future registries/studies with AADC-d gene therapy to be implemented by PTC. Part B of the registry will provide long- term follow-up data from participants treated with eladocagene exuparvovec to extend the safety profile of the product and to help characterize the long-term effectiveness of the gene therapy.

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**Study status**

Ongoing

# Research institutions and networks

## Institutions

### PTC Therapeutics

**First published:** 01/02/2024

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**Institution**

## Contact details

**Study institution contact**

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**Study contact**

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**Primary lead investigator**

Paul Lupo

**Primary lead investigator**

## Study timelines

**Date when funding contract was signed**

Actual: 31/03/2020

**Study start date**
Actual: 26/02/2021

**Date of final study report**
Planned: 29/10/2038

## Sources of funding

- Pharmaceutical company and other private sector

## More details on funding

PTC THERAPEUTICS, INC.

## Regulatory

**Was the study required by a regulatory body?**

Yes

**Is the study required by a Risk Management Plan (RMP)?**

EU RMP category 2 (specific obligation of marketing authorisation)

**Regulatory procedure number**

EMEA/H/C/005352/SOB/002

## Other study registration identification numbers and links

SOB002.1

# Methodological aspects

## Study type

**Study type:**

Non-interventional study

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**Scope of the study:**

Disease epidemiology

Other

**If ‘other’, further details on the scope of the study**

This is a 2-part registry that describes natural history of AADC-d in SoC patients (Part A). Part B aims to evaluate the long-term effectiveness and safety of eladocagene exuparvovec in participants treated for AADC-d

**Study design:**

This is an international, multicenter, longitudinal, real-world, observational registry of untreated (Part A) and untreated (Part B) participants with AADC-d
with eladocagene exuparvovec (Part B). For Part B, participants will be followed for long-term safety and
efficacy outcomes for 10 years

**Main study objective:**

Primary Objectives: Part A: • To describe the natural history of AADC-d in participants on SoC for a minimum of 5 years. Part B: • To assess the long-term effectiveness and safety of treatment in motor function over time with eladocagene exuparvovec in participants with AADC-d for a minimum of 10 years.

## Study Design

**Non-interventional study design**

Other

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**Non-interventional study design, other**

A Two-Part, International, Real-World, Observational Registry

# Study drug and medical condition

**Medical condition to be studied**

Aromatic L-amino acid decarboxylase deficiency

# Population studied

**Age groups**

- Term newborn infants (0 – 27 days)
- Infants and toddlers (28 days – 23 months)
- Children (2 to < 12 years)
- Adolescents (12 to < 18 years)
- Adults (18 to < 46 years)
- Adults (46 to < 65 years)
- Adults (65 to < 75 years)
- Adults (75 to < 85 years)
- Adults (85 years and over)

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**Estimated number of subjects**

65

# Study design details

**Outcomes**

Primary Objectives Part A: • To describe the natural history of AADC-d in participants on SoC. Part B: • To assess the long-term effectiveness and safety of treatment in motor function over time with eladocagene exuparvovec in participants with AADC-d for a minimum of 10 years. Secondary Objectives Part A Only: • To assess the achievement of motor milestones and changes in motor function over time. • To assess changes in Patient-Reported Outcomes (PRO, quality of life and health status) over time.

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**Data analysis plan**

All participants who meet the eligibility criteria will be enrolled in the registry. This is a study of an ultra-rare disease with no a priori statistical hypothesis. The sample size is not driven by any specific hypothesis to be tested or desired precision estimates but is based on anticipated prevalence of AADC-d. The analysis population includes all subjects enrolled in the registry. Data will be summarized using descriptive statistics. Numerical variables will be summarized with mean, median, standard deviation, minimum, and maximum. Categorical variables will be summarized with frequency counts and percentage. Other descriptive statistics may be reported when appropriate. Data visualization may be used to illustrate trends over time. Interim analyses for publication are expected to be conducted, at a minimum, on a yearly basis and will be included in the annual progress report for Part A and performed as part of the ongoing evaluation of effectiveness and safety for Part B.

## Data management

## ENCePP Seal

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

## Data sources

**Data sources (types)**

Other

**Data sources (types), other**

Prospective patient-based data collection

## Use of a Common Data Model (CDM)

**CDM mapping**

No

## Data quality specifications

**Check conformance**

Unknown

**Check completeness**

Unknown

**Check stability**

Unknown

**Check logical consistency**

Unknown

# Data characterisation

**Data characterisation conducted**

No