

# A Prospective, Observational, Post-Authorisation Efficacy Study to Assess Long-term Effectiveness of Risdiplam in Patients with Genetically Confirmed 5q SMA

**First published:** 29/06/2022

**Last updated:** 22/04/2025

Study

Ongoing

## Administrative details

### EU PAS number

EUPAS47916

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### Study ID

48479

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### DARWIN EU® study

No

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### Study countries

 Austria

 Germany

 Sweden

## Study description

This study is a multi-country, non-interventional, longitudinal cohort study utilising data from existing SMA patient registries. The study was initially proposed to be conducted using both primary data collection and secondary data extracted from existing SMA registries. However, given the feasibility assessment results, the prospective primary data collection was removed from the initial version of the plan, after discussion with CHMP. This study is a multi-country, non-interventional, longitudinal cohort study utilising data from existing SMA patient registries. The study was initially proposed to be conducted using both primary data collection and secondary data extracted from existing SMA registries. However, given the feasibility assessment results, the prospective primary data collection was removed from the initial version of the plan, after discussion with CHMP.

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## Study status


Ongoing


# Research institutions and networks

## Institutions

### PPD Evidera

 Sweden

 United Kingdom

 United States

**First published:** 20/11/2013

**Last updated:** 22/09/2025

**Institution**

Laboratory/Research/Testing facility

Non-Pharmaceutical company

ENCePP partner

## Pediatric Neuromuscular Clinical Research (PNCR)

### TREAT-NMD Services Ltd

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

**Last updated:** 01/02/2024

**Institution**

Other

## Contact details

### Study institution contact

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Study contact

[global.clinical\\_trial\\_registry@roche.com](mailto:global.clinical_trial_registry@roche.com)

### Primary lead investigator

Nahila Justo

Primary lead investigator

# Study timelines

## **Date when funding contract was signed**

Actual: 04/06/2021

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## **Study start date**

Planned: 30/06/2022

Actual: 27/06/2022

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## **Date of interim report, if expected**

Planned: 30/09/2025

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## **Date of final study report**

Planned: 30/12/2030

# Sources of funding

- Pharmaceutical company and other private sector

## More details on funding

Hoffmann-La Roche

## Study protocol

[Prot BN43428 risdiplam v1, Published Output-1\\_Redacted.pdf](#) (1.94 MB)

[Prot\\_BN43428\\_risdiplam\\_v2\\_12Dec2024\\_Redacted.pdf](#) (1.37 MB)

## Regulatory

## Was the study required by a regulatory body?

Yes

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

Not applicable

## Other study registration identification numbers and links

BN43428

## Methodological aspects

### Study type

### Study type list

#### **Study type:**

Non-interventional study

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#### **Scope of the study:**

Effectiveness study (incl. comparative)

#### **Main study objective:**

The primary objectives for this study are as follows: 1) To describe the real-world, long-term effectiveness of risdiplam on disease progression and to compare the impact of potential effect modifiers (symptomatic status, SMN2 copy number) on long-term effectiveness, and 2) To compare the

real-world, long-term effectiveness outcomes between a cohort of risdiplam-treated patients and a cohort of DMT-naive patients (untreated with any DMT approved for SMA).

## Study Design

### **Non-interventional study design**

Cohort

## Study drug and medical condition

### **Study drug International non-proprietary name (INN) or common name**

RISDIPLAM

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### **Medical condition to be studied**

Spinal muscular atrophy

## Population studied

### **Age groups**

- 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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### **Special population of interest**

Hepatic impaired

Immunocompromised

Renal impaired

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### **Estimated number of subjects**

600

## **Study design details**

### **Outcomes**

Primary effectiveness outcomes: 1) Time to all-cause mortality (survival) by the end of study participation, 2) Time to prolonged/permanent ventilation, 3) Developmental motor milestone achievement, 4) Motor function assessed using Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND), 5) Hammersmith Functional Motor Scale Expanded (HF MSE) or Revised Upper Limb Module (RULM).

Secondary effectiveness outcomes: 1) Onset of symptoms (within the pre-symptomatic group), 2) Need for nutritional support/tube feeding, 3) Length of stay in hospitalisations and reasons for hospitalisations, 4) Withdrawal of risdiplam treatment and reasons for withdrawal of treatment (risdiplam cohort only). Tertiary effectiveness outcomes: 1) Motor function measure 32 (MFM32), 2) Revised Hammersmith Scale (RHS), 3) Timed function tests (e.g., 6-Minute Walk Test [6MWT], 10-Metre Walk Test [10MWT], Timed Up and Go [TUG] Test), 4) SMA Independence Scale (SMAIS).

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### **Data analysis plan**

The raw data will be extracted from different data sources, followed by homogenization, pooling and then central analysis. All effectiveness outcomes will be summarized descriptively per cohort by an approximate 6-month timepoint. Continuous variables will be described with summary statistics such as n, mean, standard deviation, median, minimum, and maximum values. Also, treatment differences and 95% CIs will be presented. For each categorical variable, odds ratio, or relative risks, 95% CIs, frequency, and percentage will be reported. The time to event for each outcome will be reported in months, summarised descriptively and analysed using Kaplan-Meier method and Cox proportional hazards regression model. Median survival time estimates and survival probabilities at each study period time point will be analysed. The other objective will be achieved by comparing the two cohorts and using statistical techniques like inverse probability weighting and multivariable regression analysis.

## 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 source(s)**

Translational Research in Europe - Assessment and Treatment of  
Neuromuscular Diseases

Longitudinal Data Collection from Patients with Spinal Muscular Atrophy  
(SMARtCARE)

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### **Data source(s), other**

SMA Research and Clinical Hub (REACH UK), French Register of Patients with Spinal Muscular Atrophy (R-SMA France), Neuromuscular Diseases in Sweden – Neuromuskulära Sjukdomar i Sverige (NMiS), The Australian Neuromuscular Disease Registry, The Canadian Neuromuscular Disease Registry, CureSMA: 3 databases: CureSMA Membership Database, Pediatric Neuromuscular Clinical Research (PNCR), and the Clinical Care Network

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### **Data sources (types)**

[Disease registry](#)

[Other](#)

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### **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

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**Check completeness**

Unknown

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**Check stability**

Unknown

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**Check logical consistency**

Unknown

## Data characterisation

**Data characterisation conducted**

No