A Prospective, Long-Term Registry of Patients with a Diagnosis of Spinal Muscular Atrophy (SMA) - (RESTORE)

First published: 27/08/2021

Last updated: 16/07/2026

Study Ongoing

Administrative details

EU PAS number

EUPAS41853

Study ID

41854

DARWIN EU® study

No

Study countries

- Argentina
- Chile
- Greece
- Ireland

Japan
Korea, Democratic People's Republic of
Poland
Portugal
Romania
Russian Federation
Taiwan
United States

Study description

This prospective observational registry will assess long-term outcomes of patients with a diagnosis of SMA, including long term safety and effectiveness of OAV-101.

Study status

Ongoing

Research institutions and networks

Institutions

Novartis Pharmaceuticals

First published: 01/02/2024

Last updated: 01/02/2024

Institution

Networks

iSMAC, French SMA Registry, SMArtCARE, CuidAME, Cure SMA

Contact details

Study institution contact

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Study contact

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Primary lead investigator

Novartis Clinical Disclosure Officer

Primary lead investigator

Study timelines

Date when funding contract was signed

Planned: 01/10/2017

Actual: 12/01/2018

Study start date

Planned: 25/09/2018

Actual: 25/09/2018

Data analysis start date
Planned: 30/06/2038

Actual: 31/12/2019

Date of interim report, if expected
Planned: 12/12/2019

Actual: 02/07/2020

Date of final study report
Planned: 28/10/2038

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Novartis

Study protocol

PRO-1300 RESTORE AVXS-101-RG-001_Redacted.pdf (1.75 MB)

PMA1234_RESTORE Protocol Amendment 3.0_V4.0 final_28Sep2023 - signed_Redacted.pdf (784.1 KB)

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

AVXS-101-RG-001

Other study registration identification numbers and links

AVXS-101-RG-001, NCT04174157, COAV101A12001

Link to ClinicalTrials.gov

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Human medicinal product

Study type:

Non-interventional study

Scope of the study:

Disease epidemiology

Effectiveness study (incl. comparative)

Healthcare resource utilisation

Safety study (incl. comparative)

Data collection methods:

Combined primary data collection and secondary use of data

Study design:

Prospective, multi center, multinational, non-interventional observational registry of patients diagnosed with SMA.

Main study objective:

This registry will assess long-term outcomes of patients with a diagnosis of SMA. It will also characterize and assess long-term safety and effectiveness of OAV-101 in the real-world setting.

Study Design

Non-interventional study design

Cohort

Study drug and medical condition

Medicinal product name, other

OAV-101

Medical condition to be studied

Spinal muscular atrophy

Population studied

Short description of the study population

Inclusion Criteria:

• Patients not treated with OAV-101 with a diagnosis of SMA genetically confirmed on or after 24-May-2018

OR

Patients treated with OAV-101 with a genetically confirmed diagnosis of SMA regardless of

the date of diagnosis.

AND

• Appropriate consent/assent has been obtained for participation in the registry.

Exclusion Criteria:

• Currently enrolled in an interventional clinical trial involving an investigational medicinal product to treat SMA.

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)

Estimated number of subjects

700

Study design details

Setting

Patients will be recruited from the following regions:

o Australia/Asia Pacific

o Europe

o Middle East and Africa

o Latin America

o North America

Centers may be identified from those participating in existing SMA registries including but not limited to the international SMA consortium (iSMAC), SMArtCARE, Cure SMA, French SMA Registry, CUIDAME (Spain) or may be recruited de novo by the RESTORE registry. Patients may be enrolled in either one of the existing SMA registries with their data transferred to this registry database or they may participate in this registry without being enrolled in an existing registry.

Outcomes

To assess the effectiveness of treatments for SMA, characterize motor performance, assess the long-term safety of OAV-101, characterize risk of hepatotoxicity, thrombocytopenia, thrombotic microangiopathy, cardiac AEs and sensory abnormalities suggestive of ganglionopathy in SMA patients treated with OAV-101, assess ventilation-free survival and overall survival of all patients with SMA. To assess healthcare utilization, caregiver burden and patient functional independence. To characterize the natural history/epidemiology of patients with less than 4 copies of the SMN2 gene. To characterize the use of systemic glucocorticosteroids and other systemic immunosuppressive medication used to help manage the humoral immune response to the AAV9 vector.

Data analysis plan

Data will be analyzed per the statistical analysis plan (SAP). The analysis populations will consist of all patients enrolled. The primary analysis will be to

summarize outcomes by the therapy a patient was on at the time of enrollment. Descriptive statistics will be presented for the primary analysis. No formal a priori hypothesis testing will be performed. Continuous variables will be summarized using the number of observations, mean, 95% confidence interval (CI) for the mean, standard deviation (SD), standard error (SE), median, minimum, and maximum. Categorical data will be summarized using counts and percentages. Incidence rates (per person-years) and 95% CIs of AEs will be calculated. Survival will be presented using Kaplan-Meier methods. Further data analysis may be undertaken to meet specific regulatory requests.

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)

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

Data sources (types)

Disease registry

Electronic healthcare records (EHR)

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