Orphacol® patient surveillance database (Orphabase)

First published: 16/09/2020

Last updated: 01/10/2026

Study Ongoing

Administrative details

EU PAS number

EUPAS37229

Study ID

49962

DARWIN EU® study

No

Study countries

France
Germany
Italy
Spain
Switzerland
United Kingdom

Study description

Multi-centre, multi-national, retrospective and prospective observational study to collect patients' clinical information (treatment, safety and efficacy data) of patients receiving Orphacol® for the treatment of inborn errors in primary bile acid synthesis due to 3β-hydroxy-Δ5-C27-steroid oxidoreductase (3β-HSD) deficiency or Δ4-3-oxosteroid 5β-reductase (Δ4-3-oxoR) deficiency.

Study status

Ongoing

Research institutions and networks

Institutions

THERAVIA

Hannover Medical School (MHH)

First published: 01/02/2024

Last updated: 01/02/2024

Institution | Educational Institution | Hospital/Clinic/Other health care facility

University Medical Centre Hamburg-Eppendorf

Germany

First published: 01/02/2024

Last updated: 01/02/2024

Institution | Educational Institution | Hospital/Clinic/Other health care facility

Heidelberg University Hospital

First published: 01/02/2024

Last updated: 01/02/2024

Institution

Kremlin Bicêtre Hospital, France France, Hôpital La Timone, Marseille France, Hôpital Beaujon, Paris France, Westfällische Wilhelms-Universität Münster Germany, Universitätsklinikum Heidelberg Germany, University Hospital Hamburg-Eppendorf, Hamburg Germany, Medizinische Hochschule Hannover Germany, Ospedale Infantile Regina Margherita, Torino Italy, Fondazione I.R.C.C.S. Policlinico S.Matteo, Pavia Italy, Azienda Ospedaliera Padova Italy

Contact details

Study institution contact

Theravia Medical Affairs Direction Cmontaut@norgine.com

Study contact

Cmontaut@norgine.com

Primary lead investigator

Theravia Medical Affairs Direction

Primary lead investigator

Study timelines

Date when funding contract was signed

Planned: 17/11/2006

Actual: 17/11/2006

Study start date

Planned: 20/11/2006

Actual: 20/11/2006

Date of final study report

Planned: 13/02/2030

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Theravia

Study protocol

Orphacol_Riskmgtsystem_protocol Orphabase_v1.1_180110_clean.pdf (338.12 KB)

Protocole CA_V2.1_23.04.2024.pdf (732.8 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

Initial marketing authorisation procedure (EU/1/13/870/001-006)

Methodological aspects

Study type

Study type list

Study topic:

Human medicinal product

Study type:

Non-interventional study

Scope of the study:

Effectiveness study (incl. comparative)

Safety study (incl. comparative)

Data collection methods:

Combined primary data collection and secondary use of data

Study design:

Multi-centre, multi-national, retrospective and prospective observational study

Main study objective:

Increase the amount of available data on the treatment of inborn errors in primary bile acid synthesis due to 3β-HSD deficiency or Δ4-3-OxoR deficiency with Orphacol® in infants, children, adolescents and adults, and especially data on initial efficacy and safety of treatment with cholic acid.

Study Design

Non-interventional study design

Other

Non-interventional study design, other

Retrospective and prospective observational study

Study drug and medical condition

Medicinal product name

ORPHACOL

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

CHOLIC ACID

Anatomical Therapeutic Chemical (ATC) code

(A05AA03) cholic acid

cholic acid

Medical condition to be studied

Bile acid synthesis disorder

Additional medical condition(s)

inborn errors in primary bile acid synthesis due to 3β-Hydroxy-Δ5-C27-steroid oxidoreductase deficiency or Δ4-3-Oxosteroid-5β-reductase deficiency

Population studied

Short description of the study population

Patients of any age treated with Orphacol® for inborn errors in primary bile acid synthesis due to 3β-Hydroxy-Δ5-C27-steroid oxidoreductase deficiency or Δ4-3-Oxosteroid-5β-reductase deficiency.

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)

Special population of interest

Hepatic impaired

Immunocompromised

Pregnant women

Renal impaired

Estimated number of subjects

100

Study design details

Setting

The selected investigators are hospital-based Health Care Professional (HCP) managing patients of any age suffering from inborn errors in primary bile acid synthesis and treated with Orphacol®.

Outcomes

The primary criteria for efficacy evaluation are measurement of blood biochemistry parameters, in particular the levels of aspartate aminotransferase (ASAT), alanine aminotransferase (ALAT), gamma-glutamyl transferase (GGT), alkaline phosphatase (ALP), total bilirubin and Vitamin E.

Data analysis plan

Descriptive statistics are used to describe the included patients.

Documents

Study report

ORPHAB_2.PDF (2.47 MB)

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

Retrospective and prospective data collected from patient medical records

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