

# Orphacol® patient surveillance database (Orphabase)

**First published:** 16/09/2020

**Last updated:** 23/04/2024

Study

Ongoing

## Administrative details

### EU PAS number

EUPAS37229

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### Study ID

49962

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### DARWIN EU® study

No

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### Study countries

- ☐ France
  - ☐ Germany
  - ☐ Italy
  - ☐ Spain
  - ☐ Switzerland
  - ☐ United Kingdom
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## 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 3 $\beta$ -hydroxy- $\Delta$ 5-C27-steroid oxidoreductase (3 $\beta$ -HSD) deficiency or  $\Delta$ 4-3-oxosteroid 5 $\beta$ -reductase ( $\Delta$ 4-3-oxoR) deficiency.

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## Study status

Ongoing

# Research institutions and networks

## Institutions

### Laboratoires CTRS (Cell Therapies Research & Services)

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

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

Institution

### Heidelberg University Hospital

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

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

Institution

## 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

## Hannover Medical School (MHH)

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

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

**Institution**

Educational Institution

Hospital/Clinic/Other health care facility

Kremlin Bicêtre Hospital, France France, Hôpital La  
Timone, Marseille France, Hôpital Beaujon, Paris  
France, Westfälische 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  
[virginija.bambalaite@theravia.com](mailto:virginija.bambalaite@theravia.com)

Study contact

[virginija.bambalaite@theravia.com](mailto:virginija.bambalaite@theravia.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

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### Study start date

Planned: 20/11/2006

Actual: 20/11/2006

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### Date of final study report

Planned: 30/09/2025

## Sources of funding

- Pharmaceutical company and other private sector

## More details on funding

Theravia

## Regulatory

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

Yes

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

EU RMP category 2 (specific obligation of marketing authorisation)

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### **Regulatory procedure number**

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

## Methodological aspects

### Study type

### Study type list

#### **Study type:**

Non-interventional study

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

Effectiveness study (incl. comparative)

Safety study (incl. comparative)

**Main study objective:**

Increase the amount of available data on the treatment of inborn errors in primary bile acid synthesis due to  $3\beta$ -HSD deficiency or  $\Delta 4$ -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

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**Non-interventional study design, other**

Retrospective and prospective observational study

## Study drug and medical condition

**Name of medicine**

ORPHACOL

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**Medical condition to be studied**

Bile acid synthesis disorder

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**Additional medical condition(s)**

inborn errors in primary bile acid synthesis due to  $3\beta$ -Hydroxy- $\Delta 5$ -C27-steroid oxidoreductase deficiency or  $\Delta 4$ -3-Oxosteroid- $5\beta$ -reductase deficiency

## 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**

Renal impaired

Hepatic impaired

Immunocompromised

Pregnant women

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## **Estimated number of subjects**

100

# Study design details

## **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.

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## **Data analysis plan**

Descriptive statistics are used to describe the included patients.

## Data management

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

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### 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

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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