Maternal and Postnatal Outcomes: A worldwide decentralized observational registry to evaluate the safety in women with Fabry disease and their infants after exposure to Elfabrio (pegunigalsidase alfa-iwxj/pegunigalsidase alfa) during pregnancy and/or lactation

First published: 02/06/2026

Last updated: 02/06/2026

Study Ongoing

Administrative details

EU PAS number

EUPAS1000000957

Study ID

1000000957

DARWIN EU® study

No

Study countries

Germany

Italy

Spain

United Kingdom

United States

Study description

This is a worldwide, decentralized, single arm, prospective and retrospective, observational registry in women with Fabry disease exposed to pegunigalsidase alfa during pregnancy (within 30 days prior to the date of conception [DOC] and/or during pregnancy) and/or lactation, and their infants.

The objective of this registry is to evaluate pregnancy and clinical outcomes in women with Fabry disease and their infants after exposure to pegunigalsidase alfa at any time during pregnancy and/or lactation, and their infants.

The registry is planned to enrol over a period of 10 years.

The registry will allow physicians to enrol eligible patients and will also allow eligible patients to self-enrol, in accordance with local regulations. Patient enrolment and data collection will be coordinated through a centralized web-based platform. The web-based platform will include information about the registry and will allow the patient or the physician to create an online account to participate in the registry and independently and remotely record patient's data through electronic case report forms (eCRFs).

After enrolment in the registry, pregnancy and clinical outcomes will be collected throughout pregnancy and up to the infant's 12 months of age. Pregnancy information will be collected until deliver or within 42 days of termination of pregnancy. Patients can be enrolled in the registry at any time and depending on when enrolment occurs, patient and infant data will be collected retrospectively and/or prospectively. Information on pregnancy and infant outcomes provided by self-enrolled patients will be confirmed with their

primary care or other attending physician.
Congenital malformations will be described according to established criteria (e.g., Metropolitan Atlanta Congenital Defects Program [MACDP] criteria, European Surveillance of Congenital Anomalies [EUROCAT] criteria). Reported congenital malformations will be adjudicated by an independent Scientific Advisory Committee

Study status

Ongoing

Research institutions and networks

Institutions

Chiesi Farmaceutici

First published: 01/02/2024

Last updated: 01/02/2024

Institution

ICON Commercialisation & Outcomes

Germany

Ireland

First published: 19/03/2010

Last updated: 05/07/2024

Institution | Non-Pharmaceutical company | ENCePP partner

Contact details

Study institution contact

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Study contact

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Primary lead investigator

Joana Almeida

Primary lead investigator

Study timelines

Date when funding contract was signed

Actual: 18/12/2024

Study start date

Actual: 23/05/2025

Data analysis start date

Planned: 30/03/2035

Date of final study report

Planned: 30/10/2035

Sources of funding

- Pharmaceutical company and other private sector

More details on funding

Chiesi Farmaceutici

Study protocol

MOS CLI-06657AA1-06 Protocol v3.0_approved_Redacted.pdf (977.06 KB)

Regulatory

Was the study required by a regulatory body?

Yes

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

Non-EU RMP only

Other study registration identification numbers and links

CLI-0667AA1-06

Maternal and Postnatal Outcomes (MOS)

NCT06941025

Methodological aspects

Study type

Study type list

Study topic:

Disease /health condition

Human medicinal product

Study type:

Non-interventional study

Scope of the study:

Safety study (incl. comparative)

Data collection methods:

Primary data collection

Study design:

This is a worldwide, decentralized, single-arm, prospective and retrospective, observational registry study in women with Fabry disease exposed to pegunigalsidase alfa during pregnancy (within 30 days prior to DOC) and/or during pregnancy) and/or lactation, and their infants.

Main study objective:

The objective of the registry is to evaluate pregnancy and clinical outcomes in women with Fabry disease and their infants after exposure to pegunigalsidase alfa at any time during pregnancy and/or lactation.

Study Design

Non-interventional study design

Case-only

Cohort

Study drug and medical condition

Medicinal product name

ELFABRIO

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

PEGUNIGALSIDASE ALFA

Anatomical Therapeutic Chemical (ATC) code

(A16AB20) pegunigalsidase alfa

pegunigalsidase alfa

Medical condition to be studied

Fabry's disease

Population studied

Short description of the study population

The target population is all women with Fabry disease exposed to Elfabrio (pegunigalsidase alfa) during pregnancy and/or lactation and their fetuses, neonates and infants up to 12 months of age in the countries the study is launched in.

As Fabry disease is a rare disease with X-linked inheritance, heterozygous female subjects show a variable expression with a broad range of disease severity, ranging from asymptomatic to classical phenotype, which in turn infuences timing of treatment initiation, often postponed compared with male patients. Moreover, there are currently different therapies approved for Fabry disease, including enzyme replacement treatments in the US and EU. Therefore, the number of pregnant women with Fabry disease treated with pegunigalsidase alfa cannot be accurately estimated. It is anticipated that very

few pregnant women will be exposed to pegunigalsidase alfa every year; therefore a sample size of 10 women is expected for the duration of this study.

Patients must meet all of the following inclusion criteria to be eligible for enrolment into the registry:
- Female patients with Fabry disease who have been exposed to at least 1 dose of pegunigalsidase alfa at any time during pregnancy (defined as having received pegunigalsidase alfa within 30 days prior to the date of conception [DOC] and/or during pregnancy) and/or during lactation. (DOC, defined as 2 0/7 gestational weeks, will be calculated from last menstrual period [LMP] or ultrasound)
- Patient or parent/legally authorized representative must be able to understand and provide consent through an Institutional Review Board/Independent Ethics Committee (IRB/IEC)-approved Informed Consent Form (ICF).

There are no exclusion criteria.

Age groups

- **In utero**
- Neonate
  - Preterm newborn infants (0 – 27 days)
  - Term newborn infants (0 – 27 days)
- Infants and toddlers (28 days – 23 months)
- Adults (18 to < 65 years)
  - Adults (18 to < 46 years)
  - Adults (46 to < 65 years)

Special population of interest

Nursing women

Pregnant women

Special population of interest, other

Pediatric up to 12 months of age

Estimated number of subjects

10

Study design details

Setting

Physicians of and women with Fabry disease residing in the countries the registry is launched in who are exposed to pegunigalsidase alfa during pregnancy (within 30 days prior to DOC) and/or during pregnancy) and/or lactation will be recruited to participate.

Participation is decentralized and physicians and/or patients will participate in the registry and independently and remotely record patient's data through electronic case report forms (eCRFs) on the web-based platform.

The registry is planned to enrol over a 10-year period.

Pregnancy information will be collected until delivery or within 42 days of termination of pregnancy. Infant information will be collected up to 12 months of age.

Comparators

Not applicable.

Outcomes

Primary Outcome Measures:

1. Pregnancy Outcome:

- Number of live births
- Number of preterm birth
- Number of pregnancy losses (spontaneous abortions, pregnancy terminations, fetal deaths or stillbirths)

Secondary Outcome Measures:

- Number of neonates/infants with major congenital malformations (MCMs)
- Number of ectopic or molar pregnancies
- Number of women with obstetric and delivery complications
- Number of women with complications of preeclampsia or eclampsia
- Number of women with complications of preterm prelabour rupture of membrane
- Number of neonates/infants with minor congenital malformations
- Number of infants with developmental deficiency
- Number of hospitalizations in infants
- Mortality in infants, including neonatal death and infant death
- Head circumference in infants
- Weight in infants
- Length in infants
- Number of infants born as small for gestational age (SGA)
- Number of infants with postnatal growth deficiency or failure to thrive (FTT)
- Duration of breastfeeding, number of exclusively breastfeeding women and number of breastfeeding women supplemented with formula
- Number of adverse events in infants exposed to pegunigalsidase alfa during breastfeeding

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Data analysis plan

Details about the planned statistical analysis will be described in the Statistical Analysis Plan (SAP) which will be finalized before database lock.

Analyses will be descriptive, with data listings, frequency tabulations, and summary statistics as appropriate. No formal hypothesis testing will be performed.

Descriptive statistics will comprise of the number of observations (n), mean, standard deviation (SD), median, minimum, and maximum for continuous variables, and n and percent for categorical variables. The 95% confidence intervals (CI) will also be presented for selected pregnancy outcomes. There will be no imputation for missing data.

Demographic and other clinical characteristics will be summarized descriptively.

Primary analyses:
The number and proportions with their 95% CI will be reported for each of the pregnancy outcomes including live births, preterm births, and pregnancy losses.

Secondary analyses:
The number and proportion of patients/infants with the following birth and pregnancy outcomes will be calculated and reported.
- Major and minor congenital malformations
- Ectopic or molar pregnancies
- Obstetric and delivery complications
- Preeclampsia or eclampsia
- Preterm prelabor rupture of membrane
- SGA
- Postnatal growth deficiency or FTT
- Infant developmental deficiency

The number and proportion of infant hospitalizations and infant mortality, including neonatal and infant death, will be reported.

Infant head circumference, weight and length at birth and during follow-up will be summarized descriptively.

Breastfeeding patterns will be summarized, including duration of breastfeeding, number of women exclusively breastfeeding and number of women who supplemented with formula.

In addition, maternal deaths will be reported.

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.

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