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EMA/COMP/424419/2012
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Human apotransferrin for the treatment of congenital hypotransferrinaemia

On 17 July 2012, orphan designation (EU/3/12/1027) was granted by the European Commission to Sanquin Blood Supply Foundation, the Netherlands, for human apotransferrin for the treatment of congenital hypotransferrinaemia.

What is congenital hypotransferrinaemia?

Congenital hypotransferrinaemia is a genetic disease characterised by abnormally low levels of the protein transferrin in blood. Transferrin attaches to iron in the blood and delivers it to where it is needed, such as the bone marrow, where it is used for the production of haemoglobin (the protein found in red blood cells that carries oxygen around the body). Severely reduced levels of transferrin cause anaemia (low red blood cell counts), which may lead to heart problems. It also leads to free iron accumulating in tissues and organs, where it can cause damage and increase the likelihood of infections.

Congenital hypotransferrinaemia is a life-threatening condition due to severe anaemia and accumulation of iron in tissues which can cause heart problems and infections.

What is the estimated number of patients affected by the condition?

At the time of designation, congenital hypotransferrinaemia affected approximately 0.00012 in 10,000 people in the European Union (EU)*. This is equivalent to a total of around 6 people, and is below the ceiling for orphan designation, which is 5 people in 10,000. This is based on the information provided by the sponsor and the knowledge of the Committee for Orphan Medicinal Products (COMP).

What treatments are available?

At the time of orphan designation, no satisfactory treatments were authorised in the EU for this condition. Treatments included blood transfusions to manage anaemia and iron chelation therapy to reduce the accumulation of free iron in tissues and organs.

*Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed on the basis of data from the European Union (EU 27), Norway, Iceland and Liechtenstein. This represents a population of 506,300,000 (Eurostat 2011).

7 Westferry Circus ● Canary Wharf ● London E14 4HB ● United Kingdom
Telephone +44 (0)20 7418 8400 **Facsimile** +44 (0)20 7523 7040
E-mail info@ema.europa.eu **Website** www.ema.europa.eu
An agency of the European Union

How is this medicine expected to work?

This medicine is made of the transferrin protein extracted from human plasma (the liquid component of the blood) and depleted of the iron attached to it. This medicine is expected to work by replacing the missing protein, which can attach to the iron in the blood. This is expected to improve the symptoms of the disease.

What is the stage of development of this medicine?

The effects of human apotransferrin have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with human apotransferrin in patients with congenital hypotransferrinaemia were ongoing.

At the time of submission, human apotransferrin was not authorised anywhere in the EU for congenital hypotransferrinaemia or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 13 June 2012 recommending the granting of this designation.

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Opinions on orphan medicinal product designations are based on the following three criteria:

- the seriousness of the condition;
- the existence of alternative methods of diagnosis, prevention or treatment;
- either the rarity of the condition (affecting not more than 5 in 10,000 people in the EU) or insufficient returns on investment.

Designated orphan medicinal products are products that are still under investigation and are considered for orphan designation on the basis of potential activity. An orphan designation is not a marketing authorisation. As a consequence, demonstration of quality, safety and efficacy is necessary before a product can be granted a marketing authorisation.

For more information

Sponsor's contact details:

Sanquin Blood Supply Foundation
Plesmanlaan 125,
1066 CX Amsterdam
The Netherlands
Telephone: +31 20 512 3612
Telefax: +31 20 512 3750
E-mail: e.destree@sanquin.nl

For contact details of patients' organisations whose activities are targeted at rare diseases see:

- Orphanet, a database containing information on rare diseases which includes a directory of patients' organisations registered in Europe.
- European Organisation for Rare Diseases (EURORDIS), a non-governmental alliance of patient organisations and individuals active in the field of rare diseases.

Translations of the active ingredient and indication in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Human apotransferrin	Treatment of congenital hypotransferrinaemia
Bulgarian	Човешки апотрансферин	Лечение на вродена хипотрансферинемия
Czech	Lidský apotransferin	Léčba kongenitální hypotransferinémie
Danish	Humant apotransferrin	Behandling af congenital hypotransferrinæmi
Dutch	Humane apotransferrine	Behandeling van congenitale hypotransferrinemie
Estonian	Inimese apotransferriin	Kaasasündinud hüpotransferrineemia ravi
Finnish	Ihmisen apotransferriini	Synnynnäisen atransferrinemian hoito
French	Apotransferrine humaine	Traitement de l'hypotransferrinémie congénitale
German	Humanes Apotransferrin	Behandlung der angeborenen Hypotransferrinämie
Greek	Ανθρώπινη Αποτρανσφερίνη	Θεραπεία της συγγενούς υποτρανσφεριναιμίας
Hungarian	Humán apotranszferrin	Veleszületett hypotransferrinaemia kezelése
Italian	Apotransferrina umana	Trattamento dell'ipotransferrinemia congenita
Latvian	Cilvēka apotransferīns	Iedzimtas hipertransferinēmijas ārstēšana
Lithuanian	Žmogaus apotransferinas	Įgimtos hipotransferinemijos gydymas
Maltese	Apotransferrin uman	Kura tal-ipotransferrinemija konġenitali
Polish	Apotransferyna ludzka	Leczenie wrodzonej hypotransferynemii
Portuguese	Apotransferrina humana	Tratamento da hipotransferrinémia congénita
Romanian	Apotransferină umană	Tratamentul hipotransferinemiei congenitale
Slovak	Ľudský apotransferín	Liečba vrodenej hypotransferinémie
Slovenian	Humani apotransferin	Zdravljenje vrojene hipotransferinemije
Spanish	Apotransferrina humana	Tratamiento de la hipotransferrinemia congénita
Swedish	Humant apotransferrin	Behandling av medfödd hypotransferrinemi
Norwegian	Humant apotransferrin	Behandling av medfødt hypotransferrinemi.
Icelandic	Apótransferrín úr mönnum	Meðferð á meðfæddri hýpótransferrínemíu´

[1] At the time of designation