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Public summary of opinion on orphan designation

Human apotransferrin for the treatment of beta-thalassaemia intermedia and major

On 19 November 2018, orphan designation (EU/3/18/2093) was granted by the European Commission to Sanquin Plasma Products B.V., the Netherlands, for human apotransferrin for the treatment of beta-thalassaemia intermedia and major.

What is beta-thalassaemia intermedia and major?

Beta thalassaemia is an inherited disease in which patients are unable to make enough haemoglobin, the iron-rich protein found in red blood cells that carries oxygen around the body. Beta thalassaemia major is a severe form of the disease in which patients need frequent blood transfusions, while beta thalassaemia intermedia is a less severe form, which may worsen with age. Both types of beta thalassaemia are caused by defects in the gene responsible for producing beta-globin, one of the components of haemoglobin, which result in low levels of haemoglobin in the blood.

Beta thalassaemia intermedia and major are life-long debilitating diseases. They may be life threatening because of severe anaemia (low red blood cell count due to lack of haemoglobin), the need for repeated blood transfusions and the risk of complications associated with them.

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

At the time of designation, beta thalassaemia intermedia and major affected approximately 1 in 10,000 people in the European Union (EU). This was equivalent to a total of around 52,000 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 designation, the main treatments for beta thalassaemia intermedia and major were blood transfusions and the use of iron chelators (medicines for reducing 'iron overload' - the high iron levels in the body caused by repeated blood transfusions). In some cases, allogeneic haematopoietic

[*]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 28), Norway, Iceland and Liechtenstein. This represents a population of 517,400,000 (Eurostat 2018).

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stem cell transplantation was used to cure the disease. This is a complex procedure where the bone marrow of the patient is cleared of cells and replaced with healthy bone marrow cells from a matched donor, allowing the patient to produce red blood cells with normal haemoglobin.

The sponsor has provided sufficient information to show that human apotransferrin might be of significant benefit for patients with beta-thalassaemia intermedia and major. The medicine works in a different way from existing treatments and early laboratory data indicate that the medicine may improve red-blood cell counts and other signs and symptoms of the disease.

This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

How is this medicine expected to work?

This medicine is made of a blood protein called transferrin which transports iron around the body. The transferrin in this medicine is extracted from healthy human plasma (the liquid component of the blood) and has had the iron attached to it removed. The medicine is expected to attach to any free iron in the blood and deliver it to the bone marrow, where red blood cells are produced. This is expected to increase the production of red blood cells and also to reduce the deposition of free iron in organs and tissues which can damage them.

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 the medicine in patients with beta-thalassaemia intermedia and major were being planned.

At the time of submission, human apotransferrin was not authorised anywhere in the EU for beta-thalassaemia intermedia and major 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 18 October 2018 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:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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 beta-thalassaemia intermedia and major
Bulgarian	Човешки апотрансферин	Лечение на бета таласемия интермедия и майор
Croatian	Ljudski apotransferin	Liječenje beta-talasemije intermedije i major
Czech	Lidský apotransferin	Léčení beta thalasémie intermedia a major
Danish	Humant apotransferrin	Behandling af beta-thalassæmia intermedia og major
Dutch	Humane apotransferrine	Behandeling van bètathalassemie intermedia en major
Estonian	Inimese apotransferriin	Keskmise ja raske beetatalasseemia ravi
Finnish	Ihmisen apotransferriini	Beetatalassemia intermedia-ja major-tyypin hoito
French	Apotransferrine humaine	Traitement de la bêta-thalassémie intermédiaire et majeure
German	Humanes Apotransferrin	Behandlung der Beta-Thalassämie (Intermediäre und Major-Form)
Greek	Ανθρώπινη Αποτρανσφερίνη	Θεραπεία της β-μεσογειακής αναιμίας, ενδιάμεσης και μείζονος
Hungarian	Humán apotranszferrin	Béta-talasszémia intermedia és major kezelése
Italian	Apotransferrina umana	Trattamento della beta-talassemia intermedia e major
Latvian	Cilvēka apotransferīns	Vidēji izteiktas un izteiktas bēta talasēmijas ārstēšana
Lithuanian	Žmogaus apotransferinas	Vidutinio sunkumo ir sunkios β-talasemijos gydymas
Maltese	Apotransferrin uman	Kura tal-beta talassemija intermedja u maġġuri
Polish	Apotransferyna ludzka	Leczenie talasemii beta-intermedia i major
Portuguese	Apotransferrina humana	Tratamento da beta talassémia intermédia e major
Romanian	Apotransferină umană	Tratamentul beta talasemiei intermediare şi majore
Slovak	Ľudský apotransferín	Liečba stredne závažnej a závažnej beta talasémie
Slovenian	Humani apotransferin	Zdravljenje srednje in velike talasemije beta
Spanish	Apotransferrina humana	Tratamiento de la beta talasemia intermedia y mayor
Swedish	Humant apotransferrin	Behandling av beta-thalassaemia intermedia och major
Norwegian	Humant apotransferrin	Behandling av beta-thalassemia intermedia og beta-thalassemia major
Icelandic	Apótransferrín úr mönnum	Meðferð á meðalsvæsnu og svæsnu dvergkornablóðleysi (beta-thalassemia)

[1] At the time of designation