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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Lentiviral vector carrying the Fanconi anaemia-A (*FANCA*) gene for the treatment of Fanconi anaemia type A

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Disclaimer

Please note that revisions to the Public Summary of Opinion are purely administrative updates. Therefore, the scientific content of the document reflects the outcome of the Committee for Orphan Medicinal Products (COMP) at the time of designation and is not updated after first publication.

On 17 December 2010, orphan designation (EU/3/10/822) was granted by the European Commission to the Centre for Biomedical Network Research on Rare Diseases (CIBERER), Spain, for lentiviral vector carrying the Fanconi anaemia-A (*FANCA*) gene for the treatment of Fanconi anaemia type A.

In July 2014, Centre for Biomedical Network Research on Rare Diseases (CIBERER) changed name to Centro de Investigación Biomédica en Red (CIBER).

What is Fanconi anaemia type A?

Fanconi anaemia is an inherited genetic disorder. 'Type A' is the most common form of Fanconi anaemia. Patients with Fanconi anaemia type A have a defect in the Fanconi-A (*FANCA*) gene normally involved in the recognition and repair of damaged cell DNA. The defect prevents cells from replicating normally. Patients with Fanconi anaemia are generally born with physical malformations, and over the years, they develop a wide range of complications such as bone marrow failure (the body's inability to produce new blood cells) and cancers such as acute myeloid leukaemia (a cancer of the white blood cells). Bone marrow failure normally occurs during childhood.

Fanconi anaemia is a severely debilitating and life-threatening condition because of the bone marrow failure which leads to shortened life expectancy.

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

At the time of designation, Fanconi anaemia type A affected between 0.01 and 0.05 in 10,000 people in the European Union (EU). This was equivalent to a total of between 500 and 2,500 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, while different medicines were authorised to treat specific complications resulting from the disease, there were no satisfactory treatments authorised in the EU for Fanconi anaemia. The only treatment available was allogeneic haematopoietic (blood) stem cell transplantation (a complex procedure where the patient receives stem cells from a matched donor to help restore the bone marrow), but the availability of suitable donors was limited.

How is this medicine expected to work?

Lentiviral vector carrying the *FANCA* gene is a 'gene therapy product'. This is a medicine that works by delivering genes into the body. The medicine is made up of a virus that contains normal copies of the *FANCA* gene. Haematopoietic stem cells (cells that can develop into different types of blood cells) are first taken from the patient and then 'infected' with the virus so that the healthy *FANCA* gene is carried into the cells. These modified cells are transplanted back into the patient where they are expected to start producing healthy blood cells thus altering or halting the disease.

The type of virus used in this medicine (lentivirus) is modified so that it does not cause disease in humans.

What is the stage of development of this medicine?

The effects of lentiviral vector carrying the *FANCA* gene have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with the medicine had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for Fanconi anaemia type A 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 7 October 2010 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.

[*]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.
At the time of designation, this represented a population of 506,300,000 (Eurostat 2010).

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:

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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	Lentiviral vector carrying the Fanconi anaemia-A *(FANCA)* gene	Treatment of Fanconi anaemia type A
Bulgarian	Лентивирусен вектор, носещ ген на анемията на Фанкони А *(FANCA)*	Лечение на пациенти с подтип А на анемията на Фанкони
Czech	Lentivirový nosič přenášející Fanconiho anémii A *(FANCA)* gen	Léčba Fanconiho anémie typu A
Danish	Lentivirus-vektor bærende Fanconis anæmi A *(FANCA)* genet	Behandling af Fanconis anæmi type A
Dutch	Lentivirale vector dewelke het Fanconi-anemie A *(FANCA)* gen draagt	Behandeling van Fanconi-anemie type A
Estonian	Lentiviraalne vektor, mis sisaldab Fanconi aneemia A *(FANCA)* geeni	Fanconi aneemia tüübi A ravi
Finnish	Fanconin anemian A *(FANCA)* -geenin sisältävä lentivirusvektori	Fanconin anemian tyypin A hoito
French	Vector lentiviral portant le gène de l'anémie de Fanconi A *(FANCA)*	Traitement de l´anémie de Fanconi type A
German	Lentiviraler Vektor der das Fanconi-Anämie A *(FANCA)* Gen beinhaltet	Behandlung der Fanconi-Anämie Gruppe A
Greek	Λεντι-ιικοί φορείς που περιέχουν το γονίδιο της αναιμίας Fanconi A *(FANCA)*	Θεραπεία ασθενών με αναιμία Fanconi τύπου Α
Hungarian	Lentivirális vektor mely a Fanconi-anémia A *(FANCA)* gént tartalmazza	A Fanconi-anémia A altípus kezelése
Italian	Vettore lentivirale contenente il gene dell'anemia di Fanconi A *(FANCA)*	Trattamento dell'anemia di Fanconi tipo A
Latvian	Lentivīrusa vektors ar Fanconi anēmijas A *(FANCA)* gēnu	Fankoni A tipa anēmijas ārstēšana
Lithuanian	Lentivirusinis vektorius, turintis Fankonio *(Fanconi)* anemijos A *(FANCA)* geną	Fanconi anemijos A tipo gydymas
Maltese	Vettur lentivirali li fih il-gene tal-anemija ta' Fanconi A *(FANCA)*	Kura tal-anemija ta' Fanconi tat-tip A
Polish	Wektor lentiwirusowy zawierający gen niedokrwistości A Fanconiego *(FANCA)*	Leczenie niedokrwistości Fanconiego typu A
Portuguese	Vector lentiviral contendo o gene da anemia de Fanconi A *(FANCA)*	Tratamento da anemia de Fanconi tipo A
Romanian	Vector lentiviral ce conţine gena anemiei Fanconi A *(FANCA)*	Tratamentul anemiei Fanconi tip A
Slovak	Lentivírusový vektor obsahujúci gén Fanconiho anémie A *(FANCA)*	Liečba Fanconiho anémie typu A
Slovenian	Lentivirusni vektor, ki vsebuje gen Fanconijeve anemije A *(FANCA)*	Zdravljenje Fanconijeve anemije tipa A
Spanish	Vector lentiviral conteniendo el gen de la anemia de Fanconi A *(FANCA)*	Tratamiento de la anemia de Fanconi variante A

[1] At the time of designation

Swedish	Lentiviral vektor innehållande Fanconi anemi A *(FANCA)* -genen	Behandling av Fanconis anemi typ A
Norwegian	Lentiviral vektor som inneholder Fanconi-anemi A *(FANCA)*-genet	Behandling av Fanconis anemi type A
Icelandic	Lentiveiru ferja sem inniheldur Fanconi blóðleysi A *(FANCA)* erfðavísi	Meðferð á Fanconi blóðleysi, gerð A