6 May 2020
EMADOC-628903358-1826

Public summary of opinion on orphan designation

Adeno-associated virus serotype 2/6 encoding human alpha-galactosidase A cDNA for the treatment of Fabry disease

On 9 January 2020, orphan designation EU/3/19/2241 was granted by the European Commission to ERA Consulting GmbH, Germany, for adeno-associated virus serotype 2/6 encoding human alpha-galactosidase A cDNA (also known as ST-920) for the treatment of Fabry disease.

What is Fabry disease?

Fabry disease is an inherited disease that is caused by the lack of an enzyme called alpha galactosidase A, which breaks down and removes Gb3, a molecule made up of sugars and fats.

In patients with this disease, large amounts of Gb3 build up in tissues of vital organs, such as the kidneys and heart, leading to kidney failure and heart problems. Gb3 also builds up in the tissues of the skin, eye and nervous system leading to skin damage, clouding of the front part of the eye, pain in the hands and feet and complications affecting the brain.

Fabry disease is a long-term debilitating disease due to recurrent episodes of severe pain that cannot be relieved by painkillers. It is also life-threatening due to kidney, heart and brain complications.

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

At the time of designation, Fabry disease affected less than 2.6 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 135,000 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, Fabrazyme (agalsidase beta), Galafold (migalastat) and Replagal (agalsidase alfa) were authorised in the EU to treat Fabry disease.

*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 518,400,000 (Eurostat 2019).

Official address Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
Address for visits and deliveries Refer to www.ema.europa.eu/how-to-find-us
Send us a question Go to www.ema.europa.eu/contact **Telephone** +31 (0)88 781 6000 An agency of the European Union

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with Fabry disease. Laboratory studies indicate that a single dose of the medicine could restore the activity of alpha-galactosidase A for a long time and thereby reduce the need for regular treatment. 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 virus that contains the gene for alpha galactosidase A, the enzyme that patients lack. When given by injection, the virus is expected to carry the gene into the patient's liver cells, which would then start making the missing enzyme, thereby improving the symptoms of the disease.

The type of virus used in this medicine (adeno-associated virus) does not cause viral disease in humans.

What is the stage of development of this medicine?

The effects of the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with the medicine in patients with Fabry disease had started.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of Fabry disease or designated as an orphan medicinal product elsewhere for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 5 December 2019, recommending the granting of this designation.

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.

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	Adeno-associated virus serotype 2/6 encoding human alpha-galactosidase A cDNA	Treatment of Fabry disease
Bulgarian	Адено-асоцииран вирус серотип 2/6 кодиращ комплементарна ДНК за човешка алфа-галактозидаза А	Лечение на болест на Fabry
Croatian	Adeno-povezani virus serotipa 2/6 koji kodira cDNA humane alfa-galaktolakidaze A	Liječenje Fabryjeve bolesti
Czech	Adeno-asociovaný virus sérotypu 2/6 kódující cDNA lidské alfa-galaktosidázy A	Léčba Fabryho choroby
Danish	Adeno-associeret virus serotype 2/6 som koder for human alpha-galactosidase A cDNA	Behandling af Fabrys sygdom
Dutch	Adeno-geassociëerd virus serotype 2/6 welke codeert voor humaan alpha-galactosidase A cDNA	Behandeling van de ziekte van Fabry
Estonian	Inimese alfa-galaktosidaas A cDNA-d kodeeriv adeno-assotsieerunud viiruse serotüüp 2/6	Fabry tõve ravi
Finnish	Adenoassosioitu serotyypin 2/6 virus, joka koodaa ihmisen alfa-galaktosidaasi A:n cDNA:ta	Fabryn taudin hoito
French	Virus adeno-associé de serotype 2/6 codant l'alpha-galactosidase A cDNA humain	Traitement de la maladie de Fabry
German	Adeno-assoziierter Virus vom Serotyp 2/6 der für humane Alpha-Galactosidase A cDNA kodiert	Behandlung des Fabry-Syndroms
Greek	Αδενο-σχετιζόμενος ιός οροτύπου 2/6 που κωδικοποιεί το cDNA της α-γαλακτοσιδάσης galactosidase A	Θεραπεία της νόσου του Fabry
Hungarian	Humán alfa-galaktozidáz A cDNS-t kódoló 2/6 szerotípusú adeno-asszociált vírus	Fabry betegség kezelése
Italian	Virus adeno-associato di serotipo 2/6 che codifica per il cDNA dell'alfa-galattosidasi A umana	Trattamento della malattia di Fabry
Latvian	Adeno-saistītā vīrusa serotips 2/6, kas kodē cilvēka alfa-galktozidāzes A cDNS	Fabrī slimības ārstēšana
Lithuanian	Adeno-asocijuoto viruso serotipas 2/6, koduojantis žmogaus alfa-galaktozidazės A cDNR	Fabry ligos gydymas
Maltese	Virus adeno-assoċjat tas-serotip 2/6 li jikkodifika l-alfa galaktosidaži tal-bniedem A cDNA	Kura tal-marda ta' Fabry

[1] At the time of designation

Language	Active ingredient	Indication
Polish	Wektor wirusowy związany z adenowirusami serotypu 2/6 kodujacy cDNA ludzkiej alfa-galaktozydazy A	Leczenie choroby Fabry'ego
Portuguese	Vetor viral adeno-associado de serotipo 2/6 que codifica o ADN complementar (cDNA) da alfa-galactosidase A humana	Tratamento da doença de Fabry
Romanian	Vector viral adeno-asociat de serotip 2/6 ce codifică alfa-galactozidaza ADNc umană	Tratamentul bolii Fabry
Slovak	Adeno-asociovaný vírus sérotypu 2/6 kódujúci ľudskú cDNA alfa-galaktozidázy A	Liečba Fabryho choroby
Slovenian	Adeno-pridružen virus serotipa 2/6 kodiran za humano alfa-galaktozidazo A cDNA	Zdravljenje Fabryjeve bolezni
Spanish	Vector viral adeno asociado de serotipo 2/6 aue codifica el ADNc para el alfagactosidasa A humano.	Tratamiento de la enfermedad de Fabry
Swedish	Adenoassocierat virus serotype 2/6 kodande för humant alfagalaktosidas A cDNA	Behandling av Fabrys sjukdom
Norwegian	Adenoassosiert virus serotype 2/6 som koder for humant alfa-galaktosidase A cDNA	Behandling av Fabrys sykdom
Icelandic	Adenótengdar veirur af sermisgerð 2/6 sem umtákna (encode) manna alfa-galaktósídasa A cDNA	Meðferð Fabry-sjúkdóms