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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene for the treatment of Gaucher disease

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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 19 March 2015, orphan designation (EU/3/15/1468) was granted by the European Commission to Gauchers Association, United Kingdom, for adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene for the treatment of Gaucher disease.

What is Gaucher disease?

Gaucher disease is an inherited disorder that is caused by the lack of an enzyme called glucocerebrosidase. This enzyme normally breaks down a fatty waste product called glucocerebroside. Without the enzyme, glucocerebroside builds up in the body, typically in the liver, spleen and bone marrow. This causes a wide range of symptoms, including anaemia (low red blood cell counts), tiredness, easy bruising and a tendency to bleed, an enlarged spleen and liver, and bone pain and fractures. Some severe forms of the disease affecting children also involve the brain, which may cause problems such as abnormal eye and head movements and seizures (fits).

Gaucher disease is a long-term debilitating and life-threatening disease that is associated with a reduced life expectancy if left untreated.

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

At the time of designation, Gaucher disease affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 26,000 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, four medicines (eliglustat, imiglucerase, miglustat and velaglucerase alfa) were authorised for the treatment of Gaucher disease in the EU. Imiglucerase and velaglucerase alfa are 'enzyme replacement therapies' that work by replacing the missing enzyme. Eliglustat and miglustat block the production of glucocerebroside.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with Gaucher disease because it works in different way to existing treatments and early studies in experimental models have shown that it has the potential to cure 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?

The medicine is made of a virus that contains the gene for glucocerebrosidase, the enzyme that is missing in patients with Gaucher disease. After being injected into the patient, the virus is expected to carry the gene into the cells, enabling them to produce the missing enzyme, thus restoring the body's ability to break down glucocerebroside.

The type of virus used in this medicine ('adeno-associated virus') does not cause 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 Gaucher disease had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for Gaucher disease 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 12 February 2015 recommending the granting of this designation.

*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 512,900,000 (Eurostat 2015).

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:

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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	Adeno-associated viral vector serotype 9 containing the human glucocerebrosidase gene	Treatment of Gaucher disease
Bulgarian	Адено-асоцииран вирусен вектор от серотип 9, съдържащ човешкия ген за глюкоцереброзидаза	Лечение на болест на Гоше
Croatian	Adeno-povezani virusni vektor serotipa 9 koji sadrži ljudski gen za glukocerebrozidazu	Liječenje Gaucherove bolesti
Czech	Adeno-asociovaný virový vektor sérotypu 9, který obsahuje lidský glukocerebrosidáza gen	Léčba Gaucherovy choroby
Danish	Adeno-associeret viral vektor serotype 9 indeholdende det humane glukocerebrosidase gen	Behandling af Gauchers sygdom
Dutch	Adeno-geassocieerde virale vector van serotype 9 die het humane cardiale gen calsequestrin bevat	Behandeling van de ziekte van Gaucher
Estonian	Adeno-assotsieerunud viirusvektor serotüüp 9, mis sisaldabd inimese glükotserebrosidaasi geeni	Gaucher' tõve ravi
Finnish	Adeno-associated virus-vektori, serotyyppi 9, joka sisältää ihmisen glukoserebrosidaasigeenin	Gaucherin taudin hoito
French	Vecteur viral adéno-associé de sérotype 9 contenant le gène humain glucocérébrosidase	Traitement de la maladie de Gaucher
German	Adeno-assoziierter viraler Vektor Serotyp 9, der das Gen für das humane Glucocerebrosidase-Gen enthält	Behandlung der Gaucher-Krankheit
Greek	Αδενο-σχετιζόμενος ιικός φορέας ορότυπου 9 που περιέχει το ανθρώπινο γονίδιο της ανθρώπινης γλυκοκερεβροσιδάσης	Θεραπεία της νόσου του Gaucher
Hungarian	Humán glükocerebrozidáz gént tartalmazó 9-es szerotípusú adeno-asszociált vírusvektor	Gaucher-kór kezelése
Italian	Vettore virale adeno-associato del sierotipo 9, contenente il gene della glucocerebrosidasi umana	Trattamento della malattia di Gaucher
Latvian	Ar adenovīrusu-saistīts 9. serotipa vīrusa vektors, kas satur cilvēka glikocerebrosidāzes gēnu	Gošē slimības ārstēšana
Lithuanian	Adeno asocijuotas viruso vektoriaus 9 serotipas, pernešantis žmogaus gliukocerebrozidazės geną	Gošė ligos gydymas
Maltese	Vettur imnissel mill-adenovirus tas-serotip 9 li fih il-ġene glucocerebrosidase uman	Kura tal-marda ta' Gaucher
Polish	Wektor adenowirusowy serotypu 9 zawierający gen ludzkiej glukocerebrozydazy	Leczenie choroby Gaucher'a
Portuguese	Vector viral adeno-associado de serotipo 9, contendo o gene humano da glucocerebrosidase	Tratamento da doença de Gaucher
Romanian	Vector viral adeno-asociat de serotip 9 ce conține gena glucocerebrozidazei umane	Tratamentul bolii Gaucher

[1] At the time of designation

Language	Active ingredient	Indication
Slovak	Adeno-asociovaný vírusový vektor sérotypu 9, ktorý obsahuje ľudský gén glukocerebrozidázy	Liečba Gaucherovej choroby
Slovenian	Adenoasociiran virusni vektor serotipa 9, ki vsebuje gen humane glukocerebrozidaze	Zdravljenje Gaucherove bolezni
Spanish	Vector viral adeno-asociado de serotipo 9 que contiene el gen glucocerebrosidasa humana	Tratamiento de la enfermedad de Gaucher
Swedish	Adenoassocierad virusvektor av serotyp 9 innehållande den humana glukocerebrosidas–genen	Behandling av Gauchers sjukdom
Norwegian	Adenoassosiert virusvektor serotype 9 som inneholder det humane glukocerebrosidase genet	Behandling av Gauchers sykdom
Icelandic	Adenótengd veiruferja sermisgerð 9 sem inniheldur manna glúkóseribrósidasa gen	Meðferð á Gauchersveiki