8 March 2017
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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant adeno-associated viral vector serotype 9 containing the human N-alpha-acetylglucosaminidase gene for the treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)

On 12 January 2017, orphan designation (EU/3/16/1825) was granted by the European Commission to Ser-mes Planificación SL, Spain, for recombinant adeno-associated viral vector serotype 9 containing the human N-alpha-acetylglucosaminidase gene for the treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome).

What is mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)?

Mucopolysaccharidosis type IIIB (also known as Sanfilippo B syndrome) is a genetic disease that is caused by the lack of an enzyme called N-alpha-acetylglucosaminidase. This enzyme is needed to break down a substance in the body called heparan sulphate. Because patients with mucopolysaccharidosis type IIIB cannot break this substance down, it gradually builds up in cells in the body, particularly in the brain, and damages them. This causes a wide range of symptoms, including behavioural problems, learning disabilities and sleep disturbances. The disease is usually diagnosed in children between two and six years of age.

Mucopolysaccharidosis type IIIB is a seriously debilitating and life-threatening disease that progress to serious mental disability. The disease usually leads to death during adolescence or early adulthood.

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

At the time of designation, mucopolysaccharidosis type IIIB affected less than 0.1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 5,000 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).

[*]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 513,700,000 (Eurostat 2016).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for treating mucopolysaccharidosis type IIIB. Patients received supportive treatment to temporarily relieve the symptoms of the disease.

How is this medicine expected to work?

This medicine is made of a virus that has been modified to contain a copy of the gene for N-alpha-acetylglucosaminidase, the enzyme that is lacking in patients with mucopolysaccharidosis type IIIB. When injected into the patient, the virus is expected to carry the gene into the cells, including the nerve cells of the brain, enabling them to start producing the enzyme. As a result the cells will be able to break down the accumulated heparan sulphate, thereby helping to relieve the symptoms of the disease.

The 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 mucopolysaccharidosis type IIIB had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for mucopolysaccharidosis type IIIB. Orphan designation of the medicine had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 8 December 2016 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	Recombinant adeno-associated viral vector serotype 9 containing the human N-alpha-acetylglucosaminidase gene	Treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)
Bulgarian	Рекомбинантен адено-асоцииран вирусен вектор серотип 9, съдържащ човешки N-алфа--ацетилглюкозаминидаза ген	Лечение на мукополизахаридоза тип IIIB (синдром на Санфилипо B)
Croatian	Rekombinantni adeno-vezani virusni vektor serotipa 9 koji sadržava čovječji gen za N-alfa-acetilglukozaminidazu	Liječenje mukopolisaharidoze tipa IIIB (Sanfilippov B sindrom)
Czech	Rekombinantní adeno-asociovaný virový vektor serotypu 9 nesoucí gen lidské N-alfa-acetylglucosaminidázy	Léčba mukopolysacharidozy typu IIIB (syndrom Sanfilippo B)
Danish	Rekombinant adenoassocieret virusvektor serotype 9 indeholdende det menneskelige N-alfa-acetylglucosaminidase-gen	Behandling af mucopolysaccharidose type IIIB (Sanfilippo B syndrom)
Dutch	Recombinant adeno-geassociëerde virale vector serotype 9 dat het menselijke N-alpha-acetylglucosaminidasegen bevat	Behandeling van mucopolysacharidose type IIIB (Sanfilippo-B-syndroom)
Estonian	Inimese N-alfa-atsetüülglükoosaminidaasi geeni sisaldav rekombinantne adenoviirusega assotsieerunud viirusvektori serotüüp 9	IIIB-tüüpi mukopolüsahharidoosi (B-tüüpi Sanfilippo sündroomi) ravi
Finnish	Rekombinantti adenoassosioitu virusvektori, serotyyppiä 9, joka sisältää ihmisen N-asetyyli-alfa-glukosaminidaasigeenin	Tyypin IIIB (Sanfilippo B) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé recombinant de sérotype 9 contenant le gène humain N-alpha-acétylglucosaminidase	Traitement de la mucopolysaccharidose de type IIIB (maladie de Sanfilippo B)
German	Rekombinanter Adeno-assoziierter viraler Vektor des Serotyps 9, der das menschliche N-alpha-Acetylglucosaminidase Gen enthält	Behandlung der Mukopolysaccharidose Typ IIIB (Sanfilippo-Syndrom Typ B)
Greek	Ανασυνδυασμένος αδενο-σχετιζόμενος ιικός φορέας ορότυπου 9 που περιέχει το ανθρώπινο γονίδιο N-α-ακετυλογλυκοζαμινιδάσης	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIB (σύνδρομο Sanfilippo B)
Hungarian	9-es szerotípusú rekombináns adeno-asszociált vírus vektor, amely tartalmazza a humán N-alfa-acetil-glükózaminidáz gént	IIIB típusú mucopolisaccharidosis (Sanfilippo B szindróma) kezelése
Italian	Vettore virale adeno-associato ricombinante del sierotipo 9 recante il gene dell'alfa-N-acetilglucosaminidasi umano	Trattamento della mucopolisaccaridosi di tipo IIIB (sindrome di Sanfilippo B)
Latvian	Rekombinants adeno-asociēts 9. serotipa vīrusa vektors, kas satur cilvēka N-alfa-acetilglikozaminidāzes gēnu	IIIB tipa mukopolisaharidozes (Sanfilipo B sindroms) ārstēšana

[1] At the time of designation

Language	Active ingredient	Indication
Lithuanian	Rekombinantinis adeno-asocijuotas 9 serotipo virusinis vektorius, turintis žmogaus N-alfa-acetilglukozaminidazės geną	Mukopolisacharidozės, IIIB tipo gydymas (Sanfilippo B sindromas)
Maltese	Vettur virali rikombinanti assoċjat ma' adeno serotip 9 li fih il-ġene ta' N-alfa-acetylglucosaminidase	Kura tal-mukopolisakkaridożi tat-tip IIIB (sindrome ta' Sanfilippo tat-tip B)
Polish	Rekombinowany wektor wirusowy związany z adenowirusami serotypu 9 zawierający ludzki gen N-alfa-acetyloglukozaminidazy	Leczenie mukopolisacharydozy, typ III B (zespół Sanfilippo B)
Portuguese	Vetor viral adeno-associado de serotipo 9 recombinante que contém o gene da N-alfa-acetilglucosaminidase humana	Tratamento da mucopolissacaridose, tipo IIIB (síndrome de Sanfilippo de tipo B)
Romanian	Vector viral adeno-asociat recombinant de serotip 9 ce conține gena umană a N-alfa-acetilglucozaminidazei	Tratamentul mucopolizaharidozei de tip IIIB (sindromul Sanfilippo tip B)
Slovak	Rekombinantný adeno-asociovaný vírusový vektor sérotypu 9, obsahujúci ľudský gén N-alfa-acetylglukozaminidázu	Liečba mukopolysacharidózy typu III.B (Sanfilippov syndróm B)
Slovenian	Rekombinantni adeno-pridruženi virusni vector serotip 9, ki vsebuje človeški gen N-alfa-acetilglukozaminidaza	Zdravljenje mukopolisaharidoze vrste IIIB (sindroma Sanfilippo B)
Spanish	Vector viral adenoasociado recombinante del serotipo 9 conteniendo el gen de la alfa-N-acetilglucosaminidasa humana	Tratamiento de la mucopolisacaridosis tipo IIIB (síndrome de Sanfilippo B)
Swedish	Rekombinant adenoassocierad virusvektor av serotyp 9 innehållande human N-alfa-acetylglukosaminidasgen	Behandling av mukopolysackaridos typ IIIB (Sanfilippos syndrom typ B)
Norwegian	Rekombinant adenoassosiert virusvektor serotype 9 som inneholder genet for humant N-alfa-acetylglukosaminidase	Behandling av mukopolysakkaridose, type IIIB (Sanfilippos syndrom type B)
Icelandic	Raðbrigða adenó-tengd a veiruferja af sermisgerð 9 sem inniheldur manna N-alfa-asetýlglúkósamínídasa genið	Meðferð við slímsykrukvilla gerð IIIB (Sanfilippo B heilkenni)