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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 N-acetylglucosaminidase alpha gene for the treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)

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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 24 January 2013, orphan designation (EU/3/12/1095) was granted by the European Commission to Laboratorios del Dr. Esteve S.A., Spain, for adeno-associated viral vector serotype 9 containing the human N-acetylglucosaminidase alpha 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 an inherited disease that is caused by the lack of an enzyme called alpha-N-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, difficulty moving and sleep disturbances.

Mucopolysaccharidosis type IIIB typically starts in children between three and six years of age. It is a seriously debilitating and life-threatening disease because it leads to poor development of language skills and movement, hyperactivity and slow development. The disease usually leads to death during youth.

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

At the time of designation, mucopolysaccharidosis type IIIB affected approximately 0.009 in 10,000 people in the European Union (EU). This was equivalent to a total of around 450 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, no satisfactory methods were authorised in the EU for treating mucopolysaccharidosis type IIIB.

How is this medicine expected to work?

Adeno-associated virus serotype 9 vector containing the human alpha-N-acetylglucosaminidase gene is a medicine that works by delivering genes into the body. It is made up of a virus that contains the gene for producing alpha-N-acetylglucosaminidase, the enzyme missing in patients with mucopolysaccharidosis type IIIB. The virus is expected to carry the gene into cells, enabling the cells to produce the missing enzyme for breaking down the accumulated heparan sulphate. This is expected to help relieve the symptoms of the disease.

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?

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

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 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 6 December 2012 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. This represents a population of 509,000,000 (Eurostat 2013).

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 *N-acetylglucosaminidase alpha* gene	Treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)
Bulgarian	Адено-асоцииран вирусен вектор серотип 9, носещ човешки *N-ацетилглюкозаминидаза Алфа* ген	Лечение на мукополизахаридоза тип IIIB (синдром на Санфилипо B)
Czech	Adeno-asociovaný virový vektor sérotypu 9 přenášející humánní DNA komplementární k *N-acetylglukosaminidáze alfa*	Léčba mukopolysacharidozy typu IIIB (syndrom Sanfilippo B)
Danish	Adeno-associeret viralvektor, serotype 9, indeholdende det humane *N-acetylglukosaminidase alfa* gen	Behandling af mucopolysaccharidose type IIIB (Sanfilippo B syndrom)
Dutch	Adeno-geassocieerde virale vector serotype 9 welke het humaan *N-acetylglucosaminidase alfa* gen bevat	Behandeling van mucopolysacharidose type IIIB (Sanfilippo-B-syndroom)
Estonian	Inimese *N-atsetüülglükoosaminidaasi alfa* geeni sisaldav adenoviirusega assotsieerunud vektor serotüüp 9	IIIB-tüüpi mukopolüsahharidoosi (B-tüüpi Sanfilippo sündroomi) ravi
Finnish	Adenoassosioitu virusvektori, serotyyppi 9, joka sisältää ihmisen *alfa-N-asetyyliglukoosiaminidaasigeenin*	Tyypin IIIB (Sanfilippo B) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé de sérotype 9 portant le gène de *l'N-acetylglucosaminidase alpha* humaine	Traitement de la mucopolysaccharidose de type IIIB (maladie de Sanfilippo B)
German	Adeno-assoziierter Virusvektor Serotyp 9, der das humane *N-Acetylgalactosaminidase alpha* Gen enthält	Behandlung der Mukopolysaccharidose Typ IIIB (Sanfilippo-Syndrom Typ B)
Greek	Αδενο-σχετιζόμενος ιικός φορέας οροτύπου 9 που περιέχει το ανθρώπινο γονίδιο της *α-N-ακετυλογλυκοζαμινιδάσης άλφα*	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIB (σύνδρομο Sanfilippo B)
Hungarian	Humán *N-acetil-glükózaminidáz alfa* gént hordozó, 9-es szerotípusú adeno-asszociált vírusvektor	IIIB típusú mucopolisacharidosis (Sanfilippo B szindróma) kezelése
Italian	Vettore virale adenoassociato di sierotipo 9 recante il gene dell *N-acetilglucosaminidasi alpha* umana	Trattamento della mucopolisaccaridosi di tipo IIIB (sindrome di Sanfilippo B)
Latvian	9 serotipa ar adenovīrusu saistīta vīrusa vectors, kas satur *cilvēka N-acetilglikozaminidāzes alpha* gēnu	IIIB tipa mukopolisaharidozes (Sanfilipo B sindroms) ārstēšana
Lithuanian	Adeno asocijuoto viruso vektoriaus 9 serotipas, pernešantis *žmogaus N-acetilgliukozaminidazės alfa* geną	Mukopolisacharidozės, IIIB tipo gydymas (Sanfilippo B sindromas)

[1] At the time of designation

Language	Active ingredient	Indication
Maltese	Vettur imnissel mill-adenovirus tas-serotip 9 li fih il-ġene *N-acetylglucosaminidase alfa* uman	Kura tal-mukopolisakkaridoži tat-tip IIIB (sindrome ta' Sanfilippo tat-tip B)
Polish	Wektor adenowirusowy serotypu 9 zawierający gen ludzkiej *N-acetyloglukozaminidazy alfa*	Leczenie mukopolisacharydozy, typ III B (zespół Sanfilippo B)
Portuguese	Vector viral adeno-associado do serotipo 9 contendo ADN complementar da *N-acetilglucosaminidase alfa* humana	Tratamento da mucopolissacaridose, tipo IIIB (síndrome de Sanfilippo de tipo B)
Romanian	Vector viral adeno-asociat de serotip 9 care conţine gena umană *N-acetilglucozaminidaza alfa*	Tratamentul mucopolizaharidozei de tip IIIB (sindromul Sanfilippo tip B)
Slovak	Adeno-asociovaný vírusový vektor sérotypu 9 obsahujúci ľudský gén pre *N-acetylglukozaminidázu alfa*	Liečba mukopolysacharidózy typu III.B (Sanfilippov syndróm B)
Slovenian	Vektor adenoasociacijskega virusa serotipa 9, ki prenaša komplementarno DNA humane *N-acetilglukozaminidaze alfa*	Zdravljenje mukopolisaharidoze vrste IIIB (sindroma Sanfilippo B)
Spanish	Vector viral adeno-asociado del serotipo 9 que contiene el gen de la *N-acetilglucosaminidasa alfa* humana	Tratamiento de la mucopolisacaridosis tipo IIIB (síndrome de Sanfilippo B)
Swedish	Adenoassocierad virusvektor serotype 9 innehållande den mänskliga N-acetylglukosaminidas alfa genen	Behandling av mukopolysackaridos typ IIIB (Sanfilippos syndrom typ B)
Norwegian	Adenoassosiert virusvektor serotype 9 som inneholder genet for human *N-acetylglukosaminidase alfa*	Behandling av mukopolysakkaridose, type IIIB (Sanfilippos syndrom type B)
Icelandic	Adeno-tengd veiruferja af sermisgerð 9 sem inniheldur manna *N-asetýlglúkósamínidasa alfa* gen	Meðferð við slímsykrukvilla gerð IIIB (Sanfilippo B heilkenni)