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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

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

On 27 October 2011, orphan designation (EU/3/11/917) was granted by the European Commission to Institut Pasteur, France, for adeno-associated viral vector containing the human alpha-N-acetylglucosaminidase gene for the treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome).

What is mucopolysaccharidosis type IIIB?

Mucopolysaccharidosis type IIIB (also known as Sanfilippo B syndrome) is an inherited disease that is caused by the lack of an enzyme (a specialised type of protein) 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. The disease typically starts in children between three and six years of age.

Mucopolysaccharidosis type IIIB 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 adolescence.

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

At the time of designation, mucopolysaccharidosis type IIIB affected approximately 0.005 in 10,000 people in the European Union (EU)*. This is equivalent to a total of around 250 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 27), Norway, Iceland and Liechtenstein. This represents a population of 506,300,000 (Eurostat 2011).

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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 viral 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 the missing enzyme, alpha-N-acetylglucosaminidase. The type of virus used in this medicine (adeno-associated virus) does not cause disease in humans.

When injected into the brain, the virus is expected to carry the gene mainly into the brain cells. These cells are then expected to produce the missing enzyme so that it can break down the accumulated heparan sulphate and help to relieve the symptoms of the disease.

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 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 8 September 2011 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:

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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 containing the human alpha-N-acetylglucosaminidase gene	Treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)
Bulgarian	Адено-асоцииран вирусен вектор, носещ човешки Алфа-N-ацетилглюкозаминидаза ген.	Лечение на мукополизахаридоза тип IIIB (синдром на Санфилипо B)
Czech	Adeno-asociovaný virový vektor přenášející humánní DNA komplementární k alfa-N-acetylglukosaminidáze	Léčba mukopolysacharidozy typu IIIB (syndrom Sanfilippo B)
Danish	Adeno-associeret viral vektor, indeholdende det humane alfa-N-acetylglukosaminidase gen	Behandling af mucopolysaccharidose type IIIB (Sanfilippo B syndrom)
Dutch	Adeno-geassocieerde virale vector welke het humaan alfa-N-acetylglucosaminidase gen bevat	Behandeling van mucopolysacharidose type IIIB (Sanfilippo-B-syndroom)
Estonian	Inimese alfa-N-atsetüülglükoosaminidaasi geeni sisaldav adenoviirusega assotsieerunud vektor	IIIB-tüüpi mukopolüsahharidoosi (B-tüüpi Sanfilippo sündroomi) ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää ihmisen alfa-N-asetyyliglukoosiaminidaasi geenin	Tyypin IIIB (Sanfilippo B) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé portant le gène de l'alpha-N-acetylglucosaminidase humaine	Traitement de la mucopolysaccharidose de type IIIB (maladie de Sanfilippo B)
German	Adeno-assoziierter Virusvektor, der das humane α-N-Acetylgalactosaminidase Gen trägt	Behandlung der Mukopolysaccharidose Typ IIIB (Sanfilippo-Syndrom Typ B)
Greek	Αδενο-σχετιζόμενος ιικός φορέας που μεταφέρει συμπληρωματικό DNA ανθρώπινης α-N-ακετυλογλυκοζαμινιδάσης	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIB (σύνδρομο Sanfilippo B)
Hungarian	Humán alfa-N-acetil-glükózaminidáz gént hordozó, adeno-társított vírus vector	IIIB típusú mucopolisacharidosis (Sanfilippo B szindróma) kezelése
Italian	Vettore virale adenoassociato recante il gene dell'alfa-N-acetilglucosaminidasi umana	Trattamento della mucopolisaccaridosi di tipo IIIB (sindrome di Sanfilippo B)
Latvian	Ar adenovīrusu saistīta vīrusa vectors, kas satur cilvēka Alpha-N-acetilglikozaminidāzes gēnu	IIIB tipa mukopolisaharidozes (Sanfilipo B sindroms) ārstēšana
Lithuanian	Adeno asocijuotas virusinis vektorius, pernešantis žmogaus alfa-N-acetilgliukozaminidazės geną	Mukopolisacharidozės, IIIB tipo gydymas (Sanfilippo B sindromas)

[1] At the time of designation

Language	Active ingredient	Indication
Maltese	Vettur imnissel mill-adenovirus li fih il-ġene alfa-N-acetylglucosaminidase uman	Kura tal-mukopolisakkaridożi tat-tip IIIB (sindrome ta' Sanfilippo tat-tip B)
Polish	Wektor wirusowy, związany z adenowirusem, zawierający gen ludzkiej alfa-N-acetyloglukozaminidazy	Leczenie mukopolisacharydozy, typ III B (zespół Sanfilippo B)
Portuguese	Vector viral adeno-associado contendo ADN complementar da alfa-N-acetilglucosaminidase humana	Tratamento da mucopolissacaridose, tipo IIIB (síndrome de Sanfilippo de tipo B)
Romanian	Vector viral adeno-asociat care conţine gena ce codifică alfa-N-acetilglucozaminidaza umană	Tratamentul mucopolizaharidozei de tip IIIB (sindromul Sanfilippo tip B)
Slovak	Adeno-asociovaný vírusový vektor obsahujúci gén pre alfa-N-acetylglukozaminidázu	Liečba mukopolysacharidózy typu III.B (Sanfilippov syndróm B)
Slovenian	Vektor adenoasociacijskega virusa, ki prenaša komplementarno DNA humane alfa-N-acetilglukozaminidaze	Zdravljenje mukopolisaharidoze vrste IIIB (sindroma Sanfilippo B)
Spanish	Vector viral adeno-asociado conteniendo el gen de la alfa-N-acetilglucosaminidasa humana	Tratamiento de la mucopolisacaridosis tipo IIIB (síndrome de Sanfilippo B)
Swedish	Adenoassocierad virusvektor, som innehåller den mänskliga alfa-N-acetylglukosaminidasgenen	Behandling av mukopolysackaridos typ IIIB (Sanfilippos syndrom typ B)
Norwegian	Adenoassosiert virusvektor som inneholder genet for human alfa-N-acetylglukosaminidase	Behandling av mukopolysakkaridose, type IIIB (Sanfilippos syndrom type B)
Icelandic	Kirtil-tengdur veiruferja sem inniheldur manna alfa-N-asetýlglúkósaamíníðasa gen	Meðferð við slímsykrukvilla gerð IIIB (Sanfilippo B heilkenni)