10 March 2015
EMA/COMP/317797/2013 Rev.1
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

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

First publication	9 July 2013
Rev.1: sponsor's change of address	10 March 2015

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 June 2013, orphan designation (EU/3/13/1144) was granted by the European Commission to Synageva BioPharma Ltd., United Kingdom, for recombinant human alpha-N-acetylglucosaminidase 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 and sleep disturbances.

Symptoms of mucopolysaccharidosis type IIIB typically start in children between three and six years of age. It is a seriously debilitating and life-threatening disease that progress to serious mental disability. 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 0.02 in 10,000 people in the European Union (EU). This was equivalent to a total of around 1,000 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, no satisfactory methods were authorised in the EU for treating mucopolysaccharidosis type IIIB.

How is this medicine expected to work?

The medicine consists of a copy of the enzyme, alpha-N-acetylglucosaminidase, which is missing in patients with mucopolysaccharidosis type IIIB. When it is given to the patient (either into a vein or into the space that surrounds the spinal cord), it is expected to replace the missing enzyme thereby slowing down the build-up of heparan sulphate. This is expected to reduce the symptoms of the disease.

The medicine is produced by a method known as 'recombinant DNA technology': it is purified from the egg white of hens into which a gene (DNA) has been introduced, which makes them able to produce the enzyme.

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 recombinant human alpha-N-acetylglucosaminidase in experimental models was ongoing.

At the time of submission of the application for orphan designation, no clinical trials with recombinant human alpha-N-acetylglucosaminidase in patients with mucopolysaccharidosis type IIIB had been started.

At the time of submission, recombinant human alpha-N-acetylglucosaminidase 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 15 May 2013 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.
At the time of designation, this represented a population of 512,200,000 (Eurostat 2013).

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	Recombinant human alpha-N-acetylglucosaminidase	Treatment of mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)
Bulgarian	Рекомбинантна човешка Алфа-N-ацетилглюкозаминидаза	Лечение на мукополизахаридоза тип IIIB (синдром на Санфилипо B)
Czech	Rekombinantní lidská alfa-N-acetylglukosaminidáza	Léčba mukopolysacharidozy typu IIIB (syndrom Sanfilippo B)
Danish	Rekombinant human-alfa-N-acetylglucosaminidase	Behandling af mucopolysaccharidose type IIIB (Sanfilippo B syndrom)
Dutch	Recombinant humane alfa-N-acetylglucosaminidase	Behandeling van mucopolysacharidose type IIIB (Sanfilippo-B-syndroom)
Estonian	Rekombinantne inimese alfa-N-atsetüülglükoosaminidaas	IIIB-tüüpi mukopolüsahharidoosi (B-tüüpi Sanfilippo sündroomi) ravi
Finnish	Ihmisen rekombinantti alfa-N-asetyyliglukosaminidaasi	Tyypin IIIB (Sanfilippo B) mukopolysakkaridoosin hoito
French	Alpha-N-acetylglucosaminidase recombinante humaine	Traitement de la mucopolysaccharidose de type IIIB (maladie de Sanfilippo B)
German	Rekombinante humane N-alpha-Acetylglucosaminidase	Behandlung der Mukopolysaccharidose Typ IIIB (Sanfilippo-Syndrom Typ B)
Greek	Ανασυνδυασμένη ανθρώπινη α-N-ακετυλογλυκοζαμινιδάση	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIB (σύνδρομο Sanfilippo B)
Hungarian	Rekombináns humán alfa-N-acetilglükózamil	IIIB típusú mucopolisacharidosis (Sanfilippo B szindróma) kezelése
Italian	Alfa-N-acetilglucosaminidasi umana ricombinante	Trattamento della mucopolisaccaridosi di tipo IIIB (sindrome di Sanfilippo B)
Latvian	Rekombinantā cilvēka alfa-N-acetil glikozaminidāze	IIIB tipa mukopolisaharidozes (Sanfilipo B sindroms) ārstēšana
Lithuanian	Rekombinantinė žmogaus alfa-N-acetilgliukozaminidazė	Mukopolisacharidozės, IIIB tipo gydymas (Sanfilippo B sindromas)
Maltese	Alpha-N-acetylglucosaminidase uman rikombinanti	Kura tal-mukopolisakkaridoži tat-tip IIIB (sindrome ta' Sanfilippo tat-tip B)
Polish	Rekombinowana ludzka N-alfa-acetylglukozaminidaza	Leczenie mukopolisacharydozy, typ III B (zespół Sanfilippo B)
Portuguese	Alfa-N-acetilglucosamina humana recombinante	Tratamento da mucopolissacaridose, tipo IIIB (síndrome de Sanfilippo de tipo B)
Romanian	Alfa-N-acetilglucozaminidază umană recombinantă	Tratamentul mucopolizaharidozei de tip IIIB (sindromul Sanfilippo tip B)
Slovak	Rekombinantná ľudská alfa-N-acetylglukózaminidáza	Liečba mukopolysacharidózy typu IIIB (Sanfilippov syndróm B)
Slovenian	Rekombinantna človeška alfa-N-acetil glukozaminidaza	Zdravljenje mukopolisaharidoze vrste IIIB (sindroma Sanfilippo B)

[1] At the time of designation

Language	Active ingredient	Indication
Spanish	Alfa-N-acetilglucosaminidasa humana recombinante	Tratamiento de la mucopolisacaridosis tipo IIIB (síndrome de Sanfilippo B)
Swedish	Rekombinant humant alfa-N-acetylglukosaminidas	Behandling av mukopolysackaridos typ IIIB (Sanfilippos syndrom typ B)
Norwegian	Rekombinant human alfa-N-acetylglukoseaminidase	Behandling av mukopolysakkaridose, type IIIB (Sanfilippos syndrom type B)
Icelandic	Raðbrigða manna alfa-N-asetýlglúkósamínídasi	Meðferð við slímsykrukvilla gerð IIIB (Sanfilippo B heilkenni)