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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Self-complementary adeno-associated viral vector serotype 9 containing the *SGSH* gene for the treatment of mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)

On 14 October 2016, orphan designation (EU/3/16/1761) was granted by the European Commission to Ser-mes Planificación SL, Spain, for self-complementary adeno-associated viral vector serotype 9 containing the *SGSH* gene for the treatment of mucopolysaccharidosis type IIIA (Sanfilippo A syndrome).

What is mucopolysaccharidosis type IIIA?

Mucopolysaccharidosis type IIIA (also known as Sanfilippo A syndrome) is an inherited disease that is caused by the lack of an enzyme called sulfamidase. This enzyme is needed to break down a substance in the body called heparan sulphate. Because patients with mucopolysaccharidosis type IIIA 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 is usually diagnosed in children between two and six years of age.

Mucopolysaccharidosis type IIIA 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 IIIA 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).

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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 IIIA. Patients received supportive treatment to temporarily relieve the symptoms of the disease, such as physiotherapy, speech therapy and behavioural therapy.

How is this medicine expected to work?

This medicine is made of a virus containing the gene for the enzyme that is lacking in patients with mucopolysaccharidosis type IIIA. 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, clinical trials with the medicine in patients with mucopolysaccharidosis type IIIA were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for mucopolysaccharidosis type IIIA 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 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	Self-complementary adeno-associated viral vector serotype 9 containing the *SGSH* gene	Treatment of mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)
Bulgarian	Самодопълващ се адено-свързан вирусен вектор, серотип 9, съдържащ човешкия ген *SGSH*	Лечение на мукополизахаридоза тип IIIA (синдром на Санфилипо А)
Croatian	Samostalno-komplementarni adeno-povezani virusni vektor serotipa 9 koji sadrži ljudski gen *SGSH*	Liječenje mukopolisaharidoze tipa IIIA (Sanfilippov A sindrom)
Czech	Self-komplementární adeno-asociovaný virový vektor serotyp 9 obsahujícím lidský gen *SGSH*	Léčba mukopolysacharidozy typu IIIA (syndrom Sanfilippo A)
Danish	Selv-komplementære adenoassocieret virusvektor serotype 9, der indeholder det humane *SGSH* genet	Behandling af mucopolysaccharidose type III (Sanfilippo A syndrom)
Dutch	Zelf complementaire adeno-geassociëerde virale vector serotype 9 welke die het humane gen *SGSH* bevat	Behandeling van mucopolysacharidose type IIIA (Sanfilippo-A-syndroom)
Estonian	*SGSH* geeni sisaldav isetäiendav adenoviirusega assotseerunud viirusvektor serotüüp 9	IIIA-tüüpi mukopolüsahharidoosi (A-tüüpi Sanfilippo sündroomi) ravi
Finnish	Itseään kohti takaisin taittuva, serotyyppiä 9 oleva adenoassosioitu virusvektori, joka sisältää ihmisen *SGSH*-geenin	Tyypin IIIA (Sanfilippo A) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé auto-complémentaire de sérotype 9 contenant le gène humain *SGSH*	Traitement de la mucopolysaccharidose de type IIIA (maladie de Sanfilippo A)
German	Selbstkomplementäre Adeno-assoziierter Virusvektor des Serotyps 9, der das menschliche Gen *SGSH* enthält	Behandlung der Mukopolysaccharidose Typ IIIA (Sanfilippo-Syndrom Typ A)
Greek	Αυτο-συμπληρωματικός αδενο-σχετιζόμενος ϊικός φορέας οροτύπου 9, ο οποίος περιέχει το γονίδιο *SGSH*	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIA (σύνδρομο Sanfilippo A)
Hungarian	Humán *SGSH* gént tartalmazó önkiegészítő 9-es szerotípusú adeno-asszociált vírusvektor	IIIA típusú mucopolisaccharidosis (Sanfilippo A szindróma) kezelése
Italian	Vettore virale auto-complementare adeno-associato di sierotipo 9 contenente il gene umano *SGSH*	Trattamento della mucopolisaccaridosi di tipo IIIA (sindrome di Sanfilippo A)
Latvian	Pašpapildinošs adenosaistītā vīrusa vektora 9. serotips, kas satur *SGSH* gēnu	IIIA tipa mukopolisaharidozes (Sanfilipo A sindroms) ārstēšana
Lithuanian	Sau komplementarus adeno-asocijuoto viruso 9 serotipo vektorius, pernešantis žmogaus *SGSH* geną	Mukopolisacharidozės, IIIA tipo gydymas (Sanfilippo A sindromas)

[1] At the time of designation

Language	Active ingredient	Indication
Maltese	Vettur awto komplementari mnissel mill-adenovirus tas-serotip 9 li fih il-ġene *SGSH* uman	Kura tal-mukopolisakkaridożi tat-tip IIIA (sindrome ta' Sanfilippo tat-tip A)
Polish	Komplementarny do siebie, wirusowy wektor związany z adenowirusami serotypu 9 zawierającym ludzki gen *SGSH*	Leczenie mukopolisacharydozy, typ III A (zespół Sanfilippo A)
Portuguese	Vector viral adeno-associado do serotipo 9 autocomplementar contendo o gene humano *SGSH*	Tratamento da mucopolissacaridose, tipo IIIA (síndrome de Sanfilippo de tipo A)
Romanian	Vector viral adeno-asociat de serotip 9 auto-complementar ce conţine gena *SGSH*	Tratamentul mucopolizaharidozei de tip IIIA (sindromul Sanfilippo tip A)
Slovak	Samokomplementárny adeno-asociovaný vírusový vektor sérotypu 9 obsahujúcim ľudský gén *SGSH*	Liečba mukopolysacharidózy typu III.A (Sanfilippov syndróm A)
Slovenian	Samokomplementarni adenovirusom pridruženi virusni vektor serotipa 9 ki vsebuje gen humanega *SGSH*	Zdravljenje mukopolisaharidoze vrste IIIA (sindroma Sanfilippo A)
Spanish	Vector viral adenoasociado del serotipo 9 auto complementario que contiene el gen humano *SGSH*	Tratamiento de la mucopolisacaridosis tipo IIIA (síndrome de Sanfilippo A)
Swedish	Självkomplementär adenoassocierad virusvektor, serotyp 9 innehållande den mänskliga *SGSH* genen	Behandling av mukopolysackaridos typ IIIA (Sanfilippos syndrom typ A)
Norwegian	Selvkomplementær adenoassosiert virusvektor, serotype 9 inneholdende humant *SGSH* gen	Behandling av mukopolysakkaridose, type IIIA (Sanfilippos syndrom type A)
Icelandic	Sjálfkomplimentary adenó-tengd veiruferja af sermisgerð 9 sem inniheldur SGSH genið	Meðferð við slímsykrukvilla gerð IIIA (Sanfilippo A heilkenni)