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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 *HGSNAT* gene for the treatment of mucopolysaccharidosis IIIC (Sanfilippo C syndrome)

On 21 May 2015, orphan designation (EU/3/15/1491) was granted by the European Commission to Cochamo Systems Ltd, United Kingdom, for adeno-associated viral vector serotype 9 containing the human *HGSNAT* gene for the treatment of mucopolysaccharidosis IIIC (Sanfilippo C syndrome).

What is mucopolysaccharidosis IIIC?

Mucopolysaccharidosis type IIIC (also known as Sanfilippo C syndrome) is an inherited disease that is caused by the lack of a substance called heparin-alpha-glucosaminide N-acetyltransferase (HGSNAT). This enzyme is needed to break down a substance in the body called heparan sulphate.

Because patients with mucopolysaccharidosis type IIIC cannot break this substance down, it gradually builds up in cells in the body, particularly in the brain, and damages them. The disease is usually diagnosed in children between two and seven years of age.

Mucopolysaccharidosis type IIIC is a seriously debilitating and life-threatening disease because it leads to a decline in mental abilities, behavioural problems, hyperactivity, seizures (fits), hearing loss, difficulty swallowing and frequent infections of the airways. The disease usually leads to death in the patient's thirties or in their fifties and sixties for those with milder forms of the condition.

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

At the time of designation, mucopolysaccharidosis type IIIC affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 26,000 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 512,900,000 (Eurostat 2015).

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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 IIIC. Patients received supportive treatment to temporarily relieve the symptoms of the disease, such as physiotherapy and antiepileptic medicines to treat seizures.

How is this medicine expected to work?

This medicine is made of a virus containing the gene for the HGSNAT enzyme, which is lacking in patients with mucopolysaccharidosis type IIIC. When injected into the brain of the patient, the virus is expected to carry the gene into the nerve cells, enabling the cells to start producing HGSNAT. 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 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 IIIC had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for mucopolysaccharidosis type IIIC 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 16 April 2015 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:

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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 *HGSNAT* gene	treatment of mucopolysaccharidosis IIIC (Sanfilippo C syndrome)
Bulgarian	Аденосвързан вирусен вектор серотип 9, съдържащ човешки *HGSNAT* ген	Лечение на мукополизахаридоза тип IIIC (синдром на Санфилипо С)
Croatian	Adeno-povezani virusni vektor serotipa 9 koji sadrži ljudski gen *HGSNAT*	Liječenje mukopolisaharidoze tipa IIIC (Sanfilippov C sindrom)
Czech	Sérotyp 9 adeno asociovaný virový vektor, obsahující lidský gen *HGSNAT*	Léčba mukopolysacharidozy typu IIIC (syndrom Sanfilippo C)
Danish	Adenoassocieret viral vektor serotype 9 indeholdende det humane *HGSNAT* gen	Behandling af mucopolysaccharidose type IIIC (Sanfilippo C syndrom)
Dutch	Adenogeassocieerde virale vector, serotype 9, welke het humane gen *HGSNAT* bevat	Behandeling van mucopolysacharidose type IIIC (Sanfilippo-C-syndroom)
Estonian	Adenoga assotsieeruv viirusvektori serotüüp 9, mis sisaldab inimese *HGSNAT* geeni	IIIC-tüüpi mukopolüsahharidoosi (A-tüüpi Sanfilippo sündroomi) ravi
Finnish	Adenoassosioitu virusvektori, serotyyppi 9, joka sisältää ihmisen *HGSNAT* geenin	Tyypin IIIC (Sanfilippo C) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé de sérotype 9 contenant le gène humain *HGSNAT*	Traitement de la mucopolysaccharidose de type IIIC (maladie de Sanfilippo C)
German	Das humane *HGSNAT* Gen beinhaltender, adeno-assoziierter viraler Vektor Serotyp 9	Behandlung der Mukopolysaccharidose Typ IIIC (Sanfilippo-Syndrom Typ C)
Greek	Αδενο-σχετιζόμενος ιικός φορέας ορότυπου 9 που περιέχει το ανθρώπινο γονίδιο *HGSNAT*	Θεραπεία βλεννοπολυσακχαρίδωσης τύπου IIIC (σύνδρομο Sanfilippo C)
Hungarian	Humán *HGSNAT* gént tartalmazó 9-as szerotípusú adeno-asszociált vírus vector	IIIC típusú mukopoliszaharidózis (Sanfilippo C szindróma) kezelése
Italian	Vettore virale adeno-associato del serotipo 9 contenente il gene umano *HGSNAT*	Trattamento della mucopolisaccaridosi di tipo IIIC (sindrome di Sanfilippo C)
Latvian	Adeno-saistītā vīrālā vektora 9. serotips, kas satur cilvēka *HGSNAT* gēnu	IIIC tipa mukopolisaharidozes (Sanfilipo C sindroma) ārstēšana

[1] At the time of designation

Language	Active ingredient	Indication
Lithuanian	Adeno-asocijuotas viruso vektoriaus 9 serotipas, pernešantis žmogaus *HGSNAT* geną	Mukopolisacharidozės, IIIC tipo gydymas (Sanfilippo C sindromas)
Maltese	Vettur imnissel mill-adenovirus tas-serotip 9 li fih il-ġene uman *HGSNAT*	Kura tal-mukopolisakkaridoži tat-tip IIIĊ (sindrome ta' Sanfilippo tat-tip Ċ)
Polish	Wektor adenowirusowy serotypu 9 zawierający ludzki gen *HGSNAT*	Leczenie mukopolisacharydozy, typ IIIC (zespół Sanfilippo C)
Portuguese	Vector viral adeno-associado de serotipo 9 contendo o gene humano *HGSNAT*	Tratamento da mucopolissacaridose, tipo IIIC (síndrome de Sanfilippo de tipo C)
Romanian	Vector viral adeno-asociat de serotip 9 conţinând gena umană *HGSNAT*	Tratamentul mucopolizaharidozei de tip IIIC (sindromul Sanfilippo tip C)
Slovak	Adeno-asociovaný vírusový vektor sérotypu 9 obsahujúci l'udský gén *HGSNAT*	Liečba mukopolysacharidózy typu IIIC (Sanfilippov syndróm C)
Slovenian	Adeno-pridruženi virusni vektor serotipa 9, ki vsebuje človeški gen *HGSNAT*	Zdravljenje mukopolisaharidoze vrste IIIC (sindroma Sanfilippo C)
Spanish	Vector viral adenoasociado del serotipo 9 que conteniene el gene humano *HGSNAT*	Tratamiento de la mucopolisacaridosis tipo IIIC (síndrome de Sanfilippo C)
Swedish	Adenoassocierad virusvektor serotyp 9, innehållande den mänskliga genen *HGSNAT*	Behandling av mukopolysackaridos typ IIIC (Sanfilippos syndrom typ C)
Norwegian	Adenoassosiert virusvektor serotype 9 som inneholder det humane genet *HGSNAT*	Behandling av mukopolysakkaridose, type IIIC (Sanfilippos syndrom type C)
Icelandic	Adenótengd veiruferja af sermisgerð 9 sem inniheldur manna *HGSNAT* gen	Meðferð við slímsykrukvilla gerð IIIC (Sanfilippo C heilkenni)