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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 sulfamidase gene for the treatment of mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)

On 21 June 2011, orphan designation (EU/3/11/877) was granted by the European Commission to Laboratorios del Dr. Esteve, S.A., Spain, for adeno-associated viral vector serotype 9 containing the human sulfamidase 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 (a specialised type of protein) called N-sulfoglucosamine sulfohydrolase. 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 approximately 0.05 in 10,000 people in the European Union (EU)*. This is equivalent to a total of around 3,000 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 27), Norway, Iceland and Liechtenstein. This represents a population of 506,300,000 (Eurostat 2011).

7 Westferry Circus ● Canary Wharf ● London E14 4HB ● United Kingdom
Telephone +44 (0)20 7418 8400 **Facsimile** +44 (0)20 7523 7040
E-mail info@ema.europa.eu **Website** www.ema.europa.eu

What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for treating mucopolysaccharidosis type IIIA. Bone marrow transplantation had been used to try to slow down the progression of the disease.

How is this medicine expected to work?

Adeno-associated viral vector serotype 9 containing the human sulfamidase 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, N-sulfoglucosamine sulfohydrolase. 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 this 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 IIIA had been started.

At the time of submission, this 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 7 April 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:

Laboratorios del Dr. Esteve, S.A.
Av. Mare de Déu de Montserrat, 221
08041 Barcelona
Spain
Telephone: +34 93 446 60 00
Telefax: +34 93 450 48 99
E-mail: evalenti@esteve.es

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 sulfamidase gene	Treatment of mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)
Bulgarian	Адено-свързан вирусен вектор, серологичен тип 9, съдържащ ген за човешка сулфамидаза	Лечение на мукополизахаридоза тип IIIA (синдром на Санфилипо А)
Czech	Adeno-asociovaný virový vektor (serotyp 9) obsahující gen humánní sulfaminidasy	Léčba mukopolysacharidozy typu IIIA (syndrom Sanfilippo A)
Danish	Adenoassocieret virusvektor serotype 9der indeholder det humane sulfamidase-gen	Behandling af mucopolysaccharidose type III (Sanfilippo syndrom)
Dutch	Adeno-geassociëerde virale vector serotype 9 welke het humaan sulfamidase gen bevat	Behandeling van mucopolysacharidose type IIIA (Sanfilippo-A-syndroom)
Estonian	Inimese sulfamidaasi geeni sisaldav adenoviirusega assotseerunud viirusvektor serotüüp 9.	IIIA-tüüpi mukopolüsahharidoosi (A-tüüpi Sanfilippo sündroomi) ravi
Finnish	Serotyyppiä 9 oleva adenoassosioitu virusvektori, joka sisältää ihmisen sulfamidaasi-geenin	Tyypin IIIA (Sanfilippo A) mukopolysakkaridoosin hoito
French	Vecteur viral adéno-associé de sérotype 9 qui contient le gène de la sulfamidase humaine	Traitement de la mucopolysaccharidose de type IIIA (maladie de Sanfilippo A)
German	Adeno-assoziierter Virusvektor des Serotyps 9, der das humane Sulfamidase-Gen enthält	Behandlung der Mukopolysaccharidose Typ IIIA (Sanfilippo-Syndrom Typ A)
Greek	Ιϊκός φορέας σχετιζόμενος με αδενοϊό οροτύπου 9, ο οποίος περιέχει το γονίδιο για την ανθρώπινη υαλουρονιδάση	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IIIA (σύνδρομο Sanfilippo A)
Hungarian	Humán szulfamidázt expresszáló, 9-es szerotípusú adeno-asszociált vírusvektor	IIIA típusú mucopolisacharidosis (Sanfilippo A szindróma) kezelése
Italian	Vettore virale adeno-associato di sierotipo 9 che contiene il gene della sulfamidasi umana	Trattamento della mucopolisaccaridosi di tipo IIIA (sindrome di Sanfilippo A)
Latvian	Cilvēka sulfamidāzes gēnu saturošs adenosaistīts vīrusa vektora 9. serotips	IIIA tipa mukopolisaharidozes (Sanfilipo A sindroms) ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektoriaus 9 serotipas, pernešantis žmogaus sulfamidazės geną	Mukopolisacharidozės, IIIA tipo gydymas (Sanfilippo A sindromas)
Maltese	Vettur mnissel mill-adenovirus tas-serotip 9 li fih il-ġene sulfamidase uman	Kura tal-mukopolisakkaridożi tat-tip IIIA (sindrome ta' Sanfilippo tat-tip A)
Polish	Wektor wirusowy zawierający gen ludzkiej sulfamidazy, związany z adenowirusem serotypu 9	Leczenie mukopolisacharydozy, typ III A (zespół Sanfilippo A)
Portuguese	Vector viral adeno-associado do serotipo 9 expressando o gene da sulfamidase humana	Tratamento da mucopolissacaridose, tipo IIIA (síndrome de Sanfilippo de tipo A)

[1] At the time of designation

Language	Active ingredient	Indication
Romanian	Vector viral adeno-asociat de serotip 9 ce conţine gena sulfamidazei umane	Tratamentul mucopolizaharidozei de tip IIIA (sindromul Sanfilippo tip A)
Slovak	Adeno-asociovaný vírusový vektor sérotypu 9 obsahujúci gén pre ľudskú sulfamidázu	Liečba mukopolysacharidózy typu III.A (Sanfilippov syndróm A)
Slovenian	Adenovirusni pridruženi virusni vektor serotipa 9, kivsebuje gen za humano sulfamidazo	Zdravljenje mukopolisaharidoze vrste IIIA (sindroma Sanfilippo A)
Spanish	Vector viral adenoasociado del serotipo 9 que contiene el gen de la sulfamidasa humana	Tratamiento de la mucopolisacaridosis tipo IIIA (síndrome de Sanfilippo A)
Swedish	Adenoassocierad virusvektor, serotyp 9, innehållande den humana sulfamidasgenen	Behandling av mukopolysackaridos typ IIIA (Sanfilippos syndrom typ A)
Norwegian	Adenoassosiert virusvektor serotype 9 som inneholder humant sulfamidase gen	Behandling av mukopolysakkaridose, type IIIA (Sanfilippos syndrom type A)
Icelandic	Adeno-tengd veiruferja af sermisgerð 9 sem tjáir manna súlfamíðasa	Meðferð við slímsykrukvilla gerð IIIA (Sanfilippo A heilkenni)