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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Adeno-associated viral vector containing the human *ARSB* gene for the treatment of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome)

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Rev.1: sponsor's change of address	26 November 2013
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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 13 May 2011, orphan designation (EU/3/11/864) was granted by the European Commission to Fondazione Telethon, Italy, for adeno-associated viral vector containing the human *ARSB* gene for the treatment of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome).

What is mucopolysaccharidosis VI (Maroteaux-Lamy syndrome)?

Mucopolysaccharidosis VI (also known as Maroteaux-Lamy syndrome) is an inherited disease that is caused by the lack of an enzyme called arylsulfatase B (ARSB). This enzyme is needed to break down substances in the body called glycosaminoglycans (GAGs). If the enzyme is not present, GAGs cannot be broken down and they build up in the cells and damage them. This causes a wide range of symptoms, the most noticeable being a short body, a large head, difficulty moving about, clouding of the eyes and hearing loss. The disease is usually diagnosed in children between one and five years of age.

Mucopolysaccharidosis VI is a debilitating disease that is long lasting and life threatening because of the damage to various parts of the body, particularly the spine, heart and lungs.

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

At the time of designation, mucopolysaccharidosis VI affected less than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 51,000* 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, galsulfase was authorised in the EU for the treatment of mucopolysaccharidosis VI. This is an enzyme replacement therapy which works by providing patients with the enzyme they are lacking. Some patients underwent transplantation to receive haematopoietic (blood) stem cells that are able to produce the missing enzyme.

The sponsor has provided sufficient information to show that this medicine might be of significant benefit for patients with mucopolysaccharidosis VI because early studies in experimental models show that it might improve the treatment of patients with this condition. This assumption will need to be confirmed at the time of marketing authorisation, in order to maintain the orphan status.

How is this medicine expected to work?

The medicine is made of a virus that contains normal copies of the *ARSB* gene, which is responsible for the production of the ARSB enzyme. When injected into the patient, it is expected that the virus carries the *ARSB* gene into the liver cells, enabling them to produce the missing enzyme. The enzyme is then expected to be transferred to cells throughout the body, where it will break down the accumulated GAGs, 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?

The effects of for adeno-associated viral vector containing the human *ARSB* gene 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 VI had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for mucopolysaccharidosis VI 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 9 February 2011 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 27), Norway, Iceland and Liechtenstein.
At the time of designation, this represented a population of 507,700,000 (Eurostat 2011).

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 *ARSB* gene	Treatment of mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)
Bulgarian	Адено-свързан вирусен вектор съдържащ човешки *ARSB* ген	Лечение на Мукополизахаридоза тип VI (Maroteaux-Lamy syndrome)
Czech	Adeno asociovaný virový vektor, obsahující lidský gen *ARSB*	Léčba mukopolysacharidosy typu VI (Maroteaux-Lamy syndrom)
Danish	Adenoassocieret viral vektor indeholdende det humane gen *ARSB*	Behandling af mukopolysakkaridose type VI (Maroteaux-Lamy syndrom)
Dutch	Adenogeassociëerde virale vector, welke het humane gen *ARSB* bevat	Behandeling van mucopolysaccharidosis, type VI (Maroteaux-Lamy syndroom)
Estonian	Adenoviirusega assotsieeruv viirusvektor, mis sisaldab inimese *ARSB* geeni	Mukopolüsahharidoos tüüp VI (Maroteaux-Lamy sündroom) ravi
Finnish	Adenoassosioitu virusvektori, joka sisältää ihmisen *ARSB* geenin	Mukopolysakkaridoosi VI:n (Maroteaux-Lamy-syndrooma) hoito
French	Vecteur viral adéno-associé contenant le gène humain *ARSB*	Traitement de mucopolysaccharidose, type VI (syndrome de Maroteaux et Lamy)
German	Adeno-assoziierter viraler Vektor mit humanem *ARSB* Gen	Behandlung der Mukopolysaccharidose Typ VI (Maroteaux-Lamy-Syndrom)
Greek	Ιϊκός φορέας σχετιζόμενος με αδενοϊό που περιέχει το ανθρώπινο γονίδιο *ARSB*	Θεραπεία της βλεννοπολυσακχαρίδωσης τυπου VI (σύνδρομο Maroteaux-Lamy)
Hungarian	Humán *ARSB* gént tartalmazó adeno-társított vírus vektor	VI-típusú mucopolysaccharidosis (Maroteaux-Lamy szindróma) kezelèse
Italian	Vettore virale adeno-associato contenente il gene umano *ARSB*	Trattamento della mucopolisaccaridosi tipo VI (sindrome di Maroteaux-Lamy)
Latvian	Cilvēka *ARSB* gēnu saturošs adenosaistīts vīrusa vektors ,	VI tipa mukopolisaharidozes (Marota-Lamī (*Marateux-Lamy*) sindroma) ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektorius, pernešantis žmogaus *ARSB* geną	VI tipo mukopolisacharidozės (*Maroteaux-Lamy* sindromo) gydymas
Maltese	Vettur imnissel mill-adenovirus li fih il-ġene *ARSB* uman	Kura tal-mukopolisakkaridoži tat-tip VI (sindrome ta' Maroteaux-Lamy)
Polish	Wektor sprzężony z adenowiresum zawierający ludzki gen *ARSB*	Leczenie mukopolisacharydozy typu VI (zespół Maroteaux-Lamy)
Portuguese	Vector viral adeno-associado contendo o gene humano *ARSB*	Tratamento da Mucopolissacaridose tipo VI (síndrome de Maroteaux-Lamy)
Romanian	Vector viral adeno-asociat conţinând gena umană *ARSB*	Tratamentul mucopolizaharidozei tip VI (sindromul Maroteaux-Lamy)
Slovak	Adeno-asociovaný vírusový vektor obsahujúci l'udský gén *ARSB*	Liečba mukopolysacharidózy typu VI (Maroteauxov-Lamyho syndróm)
Slovenian	Adenovirusom pridruženi virusni vektor, ki vsebuje človeški gen *ARSB*	Zdravljenje mukoplisaharidoze tipa VI (Maroteaux-Lamyov sindrom)

[1] At the time of designation

Language	Active ingredient	Indication
Spanish	Vector viral adenoasociado que conteniene el gen humano *ARSB*	Tratamiento de la Mucopolisacaridosis tipo VI (síndrome de Maroteaux-Lamy)
Swedish	Adenoassocierad virusvektor, som innehåller den mänskliga *ARSB* genen	Behandling av mukopolysackaridos typ VI (Maroteaux-Lamy syndrom)
Norwegian	Adenoassosiert virusvektor som inneholder det humane genet *ARSB*	Behandling av mukopolysakkaridose type VI (Maroteaux-Lamy syndrom)
Icelandic	Adenótengd veiruferja sem inniheldur manna *ARSB* gen	Meðferð á múkópólýsakkaríðósis tegund VI (Maroteaux-Lamy heilkenni)