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Public summary of opinion on orphan designation

Pentosan polysulfate sodium for the treatment of mucopolysaccharidosis type VI

On 21 August 2020, orphan designation EU/3/20/2315 was granted by the European Commission to Paradigm Biopharmaceuticals (Ireland) Limited, Ireland, for pentosan polysulfate sodium for the treatment of mucopolysaccharidosis type VI.

What is mucopolysaccharidosis type VI?

Mucopolysaccharidosis type VI (also known as Maroteaux-Lamy syndrome) is an inherited disease 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). Because the enzyme is not present, GAGs 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 long-lasting and life-threatening disease because of the damage to various parts of the body, particularly the spine, heart and lungs

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

At the time of designation, mucopolysaccharidosis type VI affected approximately 0.01 in 10,000 people in the European Union (EU). This was equivalent to a total of around 500 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 (ERT) which provides patients with the enzyme they are lacking. Some patients may undergo transplantation to receive haematopoietic (blood) stem cells that are able to produce the missing enzyme.

*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, Iceland, Liechtenstein, Norway and the United Kingdom. This represents a population of 519,200,000 (Eurostat 2020).

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The sponsor has provided sufficient information to show that pentosan polysulfate sodium might be of significant benefit for patients with mucopolysaccharidosis VI when combined with ERT. Laboratory studies indicate that pentosan polysulfate sodium may improve symptoms not managed by the currently authorised medicine, such as mobility, and preliminary clinical data showed lower levels of GAGs in the urine. 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?

Pentosan polysulfate sodium is thought to block the effects of inflammatory proteins produced as a result of a build-up of GAGs within the structures of the joint, especially in cartilage. The medicine may also reduce the amount of GAGs in some tissues but it is not understood if this results from reduced production of GAGs or their increased break-down. When used in combination with enzyme replacement therapy, the medicine is expected to reduce inflammation and pain in patients with mucopolysaccharidosis VI.

What is the stage of development of this medicine?

The effects of pentosan polysulfate sodium have been evaluated in experimental models

At the time of submission of the application for orphan designation, no clinical trials with pentosan polysulfate sodium in patients with mucopolysaccharidosis VI had been started.

Pentosan polysulfate sodium is a well-established substance authorised in the EU for several conditions, including the relief of bladder pain and certain vascular disorders.

At the time of submission, pentosan polysulfate sodium was not authorised anywhere in the EU for mucopolysaccharidosis VI. Orphan designation of the medicine had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 16 July 2020, 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

Contact details of the current sponsor for this orphan designation can be found on EMA website.

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	Pentosan polysulfate sodium	Treatment of mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)
Bulgarian	Пентозан полисулфат натрий	Лечение на Мукополизахаридоза тип VI (Maroteaux-Lamy syndrome)
Croatian	Natrijev pentozanpolisulfat	Liječenje mukopolisaharidoze tipa VI (Maroteaux-Lamy)
Czech	Pentosan polysulfát sodný	Léčba mukopolysacharidosy typu VI (Maroteaux-Lamy syndrom)
Danish	Pentosan polysulfat natrium	Behandling af mukopolysakkaridose type VI (Maroteaux-Lamy syndrom)
Dutch	Pentosanpolysulfaatnatrium	Behandeling van mucopolysaccharidosis, type VI (Maroteaux-Lamy syndroom)
Estonian	Pentosaanpolüsulfaatnaatrium	VI tüüpi mukopolüsahharidoosi (Maroteaux-Lamy sündroom) ravi
Finnish	Pentosaani polysulfaattinatrium	Mukopolysakkaridoosi VI:n (Maroteaux-Lamy-syndrooma) hoito
French	Polysulfate de pentosan sodique	Traitement de mucopolysaccharidose, type VI (syndrome de Maroteaux et Lamy)
German	Pentosanpolysulfat-Natrium	Behandlung der Mukopolysaccharidose Typ VI (Maroteaux-Lamy-Syndrom)
Greek	Νατριούχος πολυθειική πεντοσάνη	Θεραπεία της βλεννοπολυσακχαρίδωσης τυπου VI (σύνδρομο Maroteaux-Lamy)
Hungarian	Pentozan poliszulfát nátrium	VI-típusú mucopolysaccharidosis (Maroteaux-Lamy szindróma) kezelèse
Italian	Polisolfato di pentosano sodico	Trattamento della mucopolisaccaridosi tipo VI (sindrome di Maroteaux-Lamy)
Latvian	Pentozāna polisulfāta nātrija sāls	VI tipa mukopolisaharidozes (Marota-Lamī (Marateux-Lamy) sindroma) ārstēšana
Lithuanian	Natrio pentozano polisulfatas	VI tipo mukopolisacharidozės (Maroteaux-Lamy sindromo) gydymas
Maltese	Pentosan polysulfate sodium	Kura tal-mukopolisakkaridożi tat-tip VI (sindrome ta’ Maroteaux-Lamy)

[1] At the time of designation

Language	Active ingredient	Indication
Polish	Polisiarczan pentozanu sodu	Leczenie mukopolisacharydozy typu VI (zespół Maroteaux-Lamy)
Portuguese	Pentosano polissulfato de sódio	Tratamento da Mucopolissacaridose tipo VI (síndrome de Maroteaux-Lamy)
Romanian	Polisulfat de pentosan sodic	Tratamentul mucopolizaharidozei tip VI (sindromul Maroteaux-Lamy)
Slovak	Pentózan polysulfát sodný	Liečba mukopolysacharidózy typu VI (Maroteauxov-Lamyho syndróm)
Slovenian	Natrijev pentosan polisulfat	Zdravljenje mukoplisaharidoze tipa VI (Maroteaux-Lamyov sindrom)
Spanish	pentosano polisulfato sódico	Tratamiento de la Mucopolisacaridosis tipo VI (síndrome de Maroteaux-Lamy)
Swedish	Pentosanpolysulfat natrium	Behandling av mukopolysackaridos typ VI (Maroteaux-Lamy syndrom)
Norwegian	Pentosanpolysulfat natrium	Behandling av mukopolysakkaridose type VI (Maroteaux-Lamy syndrom)
Icelandic	Pentósan pólýsúlfat natríum	Meðferð á slímsykrukvilla, gerð VI