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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human N acetylgalactosamine 6 sulfatase for the treatment of mucopolysaccharidosis, type IVA (Morquio A syndrome)

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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 24 July 2009, orphan designation (EU/3/09/657) was granted by the European Commission to BioMarin Europe Limited, United Kingdom, for recombinant human N-acetylgalactosamine-6-sulfatase for the treatment of mucopolysaccharidosis, type IVA (Morquio A syndrome).

What is mucopolysaccharidosis type IVA (Morquio A syndrome)?

Mucopolysaccharidosis type IVA (also known as Morquio A syndrome) is an inherited disease that is caused by the lack of an enzyme called N-acetylgalactosamine-6-sulfatase. This enzyme is needed to break down substances in the body called glycosaminoglycans (GAGs). Because patients with mucopolysaccharidosis type IVA cannot break these substances down, the GAGs gradually build up in most of the bones and organs in the body and damage them. This causes a wide range of symptoms, including dwarfism, deformities in the spine, shortened bones, a bell-shaped chest, a short neck, difficulty moving, difficulty breathing, clouding of the eyes and hearing loss. The disease differs from other types of mucopolysaccharidosis in that it does not affect the patient's intelligence. It is usually diagnosed in infants between two and three years of age.

Mucopolysaccharidosis type IVA is a debilitating disease that is long lasting and may be life threatening because of the damage to the spine and the heart, and problems with breathing.

7 Westferry Circus ● Canary Wharf ● London E14 4HB ● United Kingdom
Telephone +44 (0)20 7418 8400 **Facsimile** +44 (0)20 7418 8416
E-mail info@ema.europa.eu **Website** www.ema.europa.eu
An agency of the European Union

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

At the time of designation, mucopolysaccharidosis type IVA affected less than 1.5 in 10,000 people in the European Union (EU). This was equivalent to a total of 76,000 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, no satisfactory methods were authorised in the EU for the treatment of mucopolysaccharidosis type IVA. Treatments were aimed at relieving the symptoms of the disease, and included surgery, medicines to fight infection and reduce inflammation and pain, and oxygen for patients with breathing problems.

How is this medicine expected to work?

Recombinant human N-acetylgalactosamine-6-sulfatase is expected to act in the same way as the human enzyme N-acetylgalactosamine-6-sulfatase, which is missing in patients with mucopolysaccharidosis type IVA. The replacement enzyme is expected to help to break down GAGs and stop them accumulating in the body, relieving the symptoms of the disease.

What is the stage of development of this medicine?

The effects of recombinant human N-acetylgalactosamine-6-sulfatase have been evaluated in experimental models

At the time of submission of the application for orphan designation, no clinical trials in patients with mucopolysaccharidosis type IVA had been started.

At the time of submission, recombinant human N-acetylgalactosamine-6-sulfatase was not authorised anywhere in the EU for mucopolysaccharidosis type IVA or designated as 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 4 June 2009 recommending the granting of this designation.

Update: recombinant human N-acetylgalactosamine-6-sulfatase (Vimizim) has been authorised in the EU since 28 April 2014 for the treatment of mucopolysaccharidosis, type IVA (Morquio A Syndrome, MPS IVA) in patients of all ages.

More information on Vimizim can be found in the European public assessment report (EPAR) on the Agency's website: ema.europa.eu/Find medicine/Human medicines/European Public Assessment Reports

*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 504,800,000 (Eurostat 2009).

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:

BioMarin Europe Limited
164 Shaftesbury Avenue
London WC12 8HL
United Kingdom
Tel. +44 207 420 0800
Fax +44 207 420 0829
E-mail: biomarin-europe@bmrn.com

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	Recombinant human N-acetylgalactosamine-6-sulfatase	Treatment of mucopolysaccharidosis, type IVA (Morquio A syndrome)
Bulgarian	Рекомбинатна човешка N-ацителгалактозамин-6-сулфатаза	Лечение на мукополизахаридоза тип IVA (Синдром на Morquio A)
Czech	Rekombinantní humánní N-acetylgalaktosamin-6-sulfatáza	Léčba mukopolysacharidózy typu IVA(Morquiho A syndrome)
Danish	Rekombinant human N-acetylgalaktosamin-6-sulfatase	Behandling af mucopolysaccharidose type IV (Morquio syndrom)
Dutch	Recombinant N-acetylgalactosamine-6-sulfatase	Behandeling van mucopolysaccharidose , type IVA (Morquio A syndroom)
Estonian	Rekombinantne inimese N-atsetüülgalaktoosamiin-6-sulfataas	IVA tüüpi mukopolüsahharidoosi (Morquio A sündroomi) ravi
Finnish	Rekombinantti ihmisen N-asetyyligalaktosamiini-6-sulfataasi	Tyypin IVA mukopolysakkaridoosin hoito (Morquio A:n oireyhtymä)
French	N-acétylgalactosamine-6-sulfatase recombinante humaine	Traitement de la mucopolysaccharidose de type IVA (syndrome de Morquio de type A)
German	Rekombinante humane N-Acetylgalaktosamin-6-Sulfatase	Behandlung von Mukopolysaccharidose Typ IVA (Morquio A Syndrom)
Greek	Ανασυνδιασμένη ανθρώπινη N-ακετυλογαλακτοζάμινο-6-σουλφατάση	Θεραπεία βλεννοπολυσακχαρίδωσης, τύπου IVA (σύνδρομο Morquio A)
Hungarian	Rekombináns humán N-acetilgalaktózamin-6-szulfatáz	IVA-típusú mucopolysaccharidosis (Morquio A szindróma) kezelése
Italian	N-Acetilgalattosamina-6-solfatasi umana ricombinante	Trattamento della mucopolisaccaridosi IVa (Sindrome di Morquio tipo A)
Latvian	Rekombinanta cilvēka N-acetilgalaktozamīna-6-sulfatāze	IVA tipa mukopolisaharidozes (Morkijo sindroma) ārstēšanai
Lithuanian	Rekombinantinė žmogaus N-acetilgalaktozamino-6-sulfatazė	Mukopolisacharidozės, IVA tipo (*Morquio* A sindromo) gydymas
Maltese	N-acetylgalactosamine-6-sulfatase uman rikombinanti	Kura tal-mukopolisakkaridożi tat-tip IVA (Sindrome ta' Morquio tat-tip A)
Polish	Rekombinowana ludzka 6-sulfataza N-acetylogalaktozaminy	Leczenie mukopolisacharydozy IVA (Zespół Morquio)
Portuguese	N-acetilgalactosamina-6-sulfatase humana recombinante	Tratamento da mucopolissacaridose tipo IVA (Sindrome de Morquio A)
Romanian	N-acetilgalactozamino-6-sulfatază umană recombinantă	Tratamentul mucopolizaharidozei tip IVA (Sindrom Morquio A)
Slovak	Rekombinantná humánna N-acetylgalaktozamín-6-sulfatáza	Liečba mukopolysacharidózy typu IVA (syndróm Morquio A)

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Rekombinantna človeška N-acetilgalaktozamin-6-sulfataza	Zdravljenje mukopolisaharidoze tipa IV A (sindrom Morquio A)
Spanish	N-acetilgalactosamina 6-sulfatasa recombinante humano	Tratamiento de la mucopolisacaridosis tipo IV A (síndrome de Morquio)
Swedish	Rekombinant human N-acetylgalaktosamin-6-sulfatas	Behandling av mukopolysackaridos typ IVA (Morquio A Syndrom)
Norwegian	Rekombinant human N-acetylgalaktosamin-6-sulfatase	Behandling av mukopolysakkaridose type IVA (Morquio A syndrom)
Icelandic	Raðbrigða manna N-asetýlgalaktósamín-6-súlfatasi	Meðferð við slímfjölsykrukvilla gerð IVA (Morquio A heilkenni)