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Public summary of opinion on orphan designation

Ribitol for the treatment of limb-girdle muscular dystrophy

On 19 October 2020, orphan designation EU/3/20/2337 was granted by the European Commission to Premier Research Group S.L., Spain, for ribitol (also known as BBP-418) for the treatment of limb-girdle muscular dystrophy.

What is limb-girdle muscular dystrophy?

Limb-girdle muscular dystrophy describes a group of genetic diseases that cause progressive weakness and wasting of muscles mainly of the hip and shoulders. The diseases are caused by mutations (changes) in the genes for certain proteins that are important for maintaining muscle fibres and are needed for muscles to work properly. Signs and symptoms vary in severity depending on the type of limb-girdle muscular dystrophy. In some cases, the disease may affect the heart and breathing muscles.

Limb-girdle muscular dystrophy is a long-term debilitating and life-threatening condition because it causes progressive muscle wasting and may lead to heart and breathing problems.

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

At the time of designation, limb-girdle muscular dystrophy affected approximately 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 10,000 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 to treat limb-girdle muscular dystrophy. Treatment of patients was mainly supportive and included physiotherapy and medicines to manage muscle pain and inflammation.

*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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How is this medicine expected to work?

In some patients with limb-girdle muscular dystrophy, an enzyme called FKRP, which helps maintain the muscle cell walls, does not work well enough.

Ribitol is a type of sugar that is used by FKRP. By providing more ribitol for FKRP to use, the medicine is expected to improve the maintenance of the cell walls, preventing deterioration of the muscle cells. This is expected to reduce muscle weakness and improve muscle function in patients with this form of the disease.

What is the stage of development of this medicine?

The effects of ribitol have been evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials with ribitol in patients with limb-girdle muscular dystrophy had been started.

At the time of submission, ribitol was not authorised anywhere in the EU for the treatment of limb-girdle muscular dystrophy. Orphan designation of ribitol had been granted in the United States for the condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 10 September 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.