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Public summary of opinion on orphan designation

Adeno-associated virus serotype rh74 containing the human sarcoglycan beta gene for the treatment of limb-girdle muscular dystrophy

On 9 December 2020, orphan designation EU/3/20/2365 was granted by the European Commission to Sarepta Therapeutics Ireland Limited, Ireland, for adeno-associated virus serotype rh74 containing the human sarcoglycan beta gene (also known as SRP-9003) 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).

*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).

Official address Domenico Scarlattilaan 6 ● 1083 HS Amsterdam ● The Netherlands
Address for visits and deliveries Refer to www.ema.europa.eu/how-to-find-us
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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.

How is this medicine expected to work?

Patients with type 2E limb-girdle muscular dystrophy have a mutation (change) in the gene for a protein called beta-sarcoglycan. This results in lack of the protein, which is needed for muscles to function normally. The medicine is made of a virus that has been modified to contain normal copies of the gene for producing the beta-sarcoglycan protein. After injection, the virus is expected to deliver the gene into the muscle cells, allowing them to produce the protein. This is expected to help relieve 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 the medicine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with this medicine in patients with limb-girdle muscular dystrophy type 2E were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of limb-girdle muscular dystrophy. Orphan designation had been granted in the United States for treatment of limb girdle muscular dystrophy type 2E.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 5 November 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.