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Public summary of opinion on orphan designation

Human laminin-111, recombinant for the treatment of congenital muscular dystrophy

On 6 January 2021, orphan designation EU/3/20/2388 was granted by the European Commission to Maxia Strategies-Europe Limited, Ireland, for human laminin-111, recombinant (also known as rhLAM-111) for the treatment of congenital muscular dystrophy.

What is congenital muscular dystrophy?

Congenital muscular dystrophy (CMD) represents a group of hereditary disorders, usually presenting at birth or within the first six months of life. There are many different forms of CMD and each form is caused by a specific defect in a gene. All forms of CMD share some symptoms and signs, such as weakness and degeneration of muscles, contractures and joint deformities. Usually CMD leads to difficulty in movement, skeletal deformation (scoliosis) and respiratory failure. Mental retardation is sometimes present.

The most common form of CMD is caused by a deficiency (lack) of a protein called laminin alpha 2 (also called merosin). Laminins are found in tissues where they provide support to the cells and they also have other functions such as protecting the cells from dying. Laminin alpha 2 supports muscle cells and a deficiency of laminin alpha 2 in muscle tissue leads to increased muscle cell death and progressive muscle weakness.

Congenital muscular dystrophy with merosin (laminin alpha 2) deficiency is a chronically debilitating and life-threatening disease.

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

At the time of designation, congenital muscular dystrophy affected approximately 0.2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 10,400 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).

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What treatments are available?

No satisfactory methods were authorised at the time of designation for the treatment of CMD. Patients received supportive care to manage the symptoms of the condition.

How is this medicine expected to work?

The medicine contains a synthetic version of a natural protein called laminin-111 that is normally produced during development in the womb. Laminin-111 works in the same way as the missing laminin alpha 2 in patients with this form of CMD. By supplying laminin-111, the medicine is expected to replace the missing protein and allow muscle cells to survive and function more normally, controlling symptoms of the condition.

What is the stage of development of this medicine?

The effects of human laminin-111, recombinant, 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 congenital muscular dystrophy had been started.

At the time of submission, human laminin-111, recombinant, was not authorised anywhere in the EU for the treatment of congenital muscular dystrophy. Orphan designation of the medicine had been granted in the United States for treatment of merosin (laminin-alpha2) deficient congenital muscular dystrophy type 1A.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 3 December 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.