



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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Public summary of opinion on orphan designation

2'-O-(2-methoxyethyl) phosphorothioate antisense oligonucleotide targeting CD49d RNA for the treatment of Duchenne muscular dystrophy

On 9 December 2020, orphan designation EU/3/20/2375 was granted by the European Commission to Pharma Gateway AB, Sweden, for 2'-O-(2-methoxyethyl) phosphorothioate antisense oligonucleotide targeting CD49d RNA (also known as ATL1102) for the treatment of Duchenne muscular dystrophy.

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy is a genetic disease that causes increasing weakness and atrophy (wasting) of muscles. It mainly affects boys, and usually starts before the age of 6 years. The muscle weakness usually starts in the hips and legs, before affecting the arms, chest and the heart. Patients with DMD lack normal dystrophin, a protein found in muscles. Because this protein helps to protect muscles from injury as muscles contract and relax, the muscles become progressively weaker and eventually stop working.

Duchenne muscular dystrophy causes long-term disability and is life threatening because of its effects on the heart and the respiratory muscles (muscles that are used to breathe). The disease usually leads to death in early adulthood.

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

At the time of designation, Duchenne muscular dystrophy affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 26,000 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, the medicine Translarna (ataluren) was authorised in the EU for the treatment of a group of patients with Duchenne muscular dystrophy caused by a particular type of

*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).



mutation (change), called a nonsense mutation, in the dystrophin gene. Patients also received supportive treatment such as physiotherapy.

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with Duchenne muscular dystrophy because early studies suggest it might reduce loss of muscle function in a wider group of patients compared to the authorised treatment.

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?

This medicine is an 'antisense oligonucleotide', a short strand of synthetic genetic material that blocks production of a protein called CD49d. Increased levels of CD49d occur in patients with Duchenne muscular dystrophy and high levels are associated with more severe disease. By blocking production of CD49d, the medicine is expected to reduce inflammation, thereby improving the symptoms and slowing progression of the disease.

What is the stage of the 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 the medicine in patients with Duchenne muscular dystrophy were ongoing.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of Duchenne muscular dystrophy or designated as an orphan medicinal product elsewhere for this condition.

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, a 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.