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Public summary of opinion on orphan designation

Adeno-associated viral vector serotype 9 encoding a codon-optimised human aspartylglucosaminidase transgene for the treatment of aspartylglucosaminuria

On 19 October 2020, orphan designation EU/3/20/2334 was granted by the European Commission to Real Regulatory Limited, Ireland, for adeno-associated viral vector serotype 9 encoding a codon-optimised human aspartylglucosaminidase transgene (also known as NGN-AGA) for the treatment of aspartylglucosaminuria.

What is aspartylglucosaminuria?

Aspartylglucosaminuria is an inherited disease belonging to the family of metabolic disorders called 'lysosomal storage diseases'. Patients with this condition lack the enzyme aspartylglucosaminidase, which is involved in the breakdown of molecules called glycoproteins within cells. As a result, glycoproteins accumulate in tissues, causing cell damage, particularly in the nerves and bones.

Patients with the disease have developmental delays (such as in speech and movement) in the early years of life, followed by a gradual decline in mental abilities in later years. Patients may also have problems with their bones, such as abnormalities in the curvature of their spine and in their facial features.

Aspartylglucosaminuria is a debilitating and life-threatening disease because of the decline in mental functions and other problems such as epilepsy and infections.

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

At the time of designation, aspartylglucosaminuria 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).

*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 for treating aspartylglucosaminuria were authorised in the EU at the time of designation.

How is this medicine expected to work?

The medicine contains the genetic material for producing aspartylglucosaminidase. When given to the patient, it is expected to enter the patient's cells and make them produce the enzyme, which can break down the glycoproteins that have been building up and thereby relieve symptoms of the disease.

The genetic material in this medicine is contained within a virus. The virus does not cause disease in humans.

What is the stage of development of this medicine?

The effects of this medicine have been evaluated in experimental models.

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

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of aspartylglucosaminuria 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 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.