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Public summary of opinion on orphan designation

Adeno-associated virus serotype 5 containing the human RDH12 gene for the treatment of RDH12 mutation associated retinal dystrophy

On 19 October 2020, orphan designation EU/3/20/2351 was granted by the European Commission to MeiraGTx B.V., Netherlands, for adeno-associated virus serotype 5 containing the human RDH12 gene (also known as AAV-RDH12) for the treatment of RDH12 mutation associated retinal dystrophy.

What is RDH12 mutation associated retinal dystrophy?

RDH12 mutation associated retinal dystrophy is a hereditary disease of the eye that leads to progressive loss of sight. Patients with this condition lack an enzyme called retinol dehydrogenase 12 (RDH12) which has a key role in the correct functioning of the light-sensitive cells of the eye. Without this enzyme, the light-sensitive cells in the eye get damaged and die.

RDH12 mutation associated retinal dystrophy is a long-term debilitating disease because it causes the patient's sight to get worse, eventually leading to blindness.

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

At the time of designation, RDH12 mutation associated retinal dystrophy affected approximately 0.4 in 10,000 people in the European Union (EU). This was equivalent to a total of around 20,000 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 application for orphan designation, there were no satisfactory treatments authorised in the EU for treating RDH12 mutation associated retinal dystrophy. Patients with the condition usually received supportive treatment such as vision aids.

*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?

This medicine is made of a virus that contains a working copy of the RDH12 gene. When injected into the patient's eye, it is expected that the virus will carry the RDH12 gene into the light-sensitive cells of the eye, enabling the cells to develop normally and thereby helping to improve the patient's sight.

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, no clinical trials with the medicine in patients with RDH12 mutation associated retinal dystrophy had started.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of RDH12 mutation associated retinal 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 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.