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Public summary of opinion on orphan designation

DNA plasmid encoding human transferrin gene for the treatment of retinitis pigmentosa

On 13 November 2020, orphan designation EU/3/20/2364 was granted by the European Commission to Eyevensys S.A.S, France, for DNA plasmid encoding human transferrin gene (also known as pEYS611) for the treatment of retinitis pigmentosa.

What is retinitis pigmentosa?

Retinitis pigmentosa is a group of hereditary diseases of the eye that lead to progressive loss of sight. In patients with retinitis pigmentosa, cells in the retina (the light-sensitive surface at the back of the eye) become damaged and eventually die.

Retinitis pigmentosa is a long-term debilitating disease because it gradually reduces the patient's sight, eventually leading to blindness.

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

At the time of designation, retinitis pigmentosa affected approximately 2.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 130,000 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 gene therapy medicine voretigene neparvovec was authorised for treating some patients with retinitis pigmentosa who had mutations (changes) in a specific gene. Patients with retinitis pigmentosa were given sunglasses to slow down damage to the retina and genetic counselling (discussion of the risks of passing the condition on to children).

The sponsor has provided sufficient information to show that the medicine might be of significant benefit for patients with retinitis pigmentosa because experimental models of the disease indicate that

*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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the medicine can be used for a broader group of patients than can be treated with voretigene neparvovec. 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?

It is thought that patients with retinitis pigmentosa have an excessive amount of iron in the eye, which damages the retina. The medicine is made up of a small fragment of DNA (genetic material) that can make a protein called transferrin, which attaches to iron. By injecting the medicine into muscles in the eye, the cells are expected to start producing transferrin, so reducing the damaging effect of iron and slowing down damage to the retina.

Technology using tiny electrical pulses transfers DNA into the cells when the medicine is injected into the eye.

What is the stage of development of this medicine?

At the time of submission of the application for orphan designation, the evaluation of the effects of the medicine in experimental models was ongoing.

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

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of retinitis pigmentosa 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 8 October 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.