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Public summary of opinion on orphan designation

Adeno-associated virus serotype 2/8 vector containing the human *PDE6A* gene for the treatment of retinitis pigmentosa

On 21 August 2020, orphan designation EU/3/20/2310 was granted by the European Commission to Institute for Ophthalmic Research, Germany, for adeno-associated virus serotype 2/8 vector containing the human *PDE6A* gene 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 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, retinitis pigmentosa affected approximately 2.2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 114,000 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, no satisfactory methods were authorised in the EU for the treatment of retinitis pigmentosa caused by mutations in the *PDE6A* gene. The medicine Luxturna was authorised in the EU for the treatment of retinitis pigmentosa caused by mutations in a different gene. Patients with the condition were given sunglasses to slow down the damage to the retina, genetic counselling (discussion of the risks of passing the condition on to children) and general support.

*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?

One of the causes of retinitis pigmentosa is a mutation (change) in the *PDE6A* gene, which is responsible for the production of a component of an enzyme (phosphodiesterase 6) needed for the normal functioning of retinal cells. In patients with this form of the disease, the enzyme cannot be properly formed and, as a result, does not function properly.

The medicine consists of a virus that contains a normal copy of the *PDE6A* gene. When injected into the patient's eye, under the retina, it is expected that the virus will carry the *PDE6A* gene into the retinal cells, enabling them to produce the missing component of the phosphodiesterase 6 enzyme. This is then expected to help the cells in the retina to function better, reducing progression of the disease.

The 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 adeno-associated virus serotype 2/8 vector containing the human PDE6A gene 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 retinitis pigmentosa had 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 16 July 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.

Translations of the active ingredient and indication in all official EU languages[1], Norwegian and Icelandic

Language	Active ingredient	Indication
English	Adeno-associated virus serotype 2/8 vector containing the human *PDE6A* gene	Treatment of retinitis pigmentosa
Bulgarian	Адено-асоцииран вирусен вектор серотип 2/8, съдържащ човешкия *PDE6A* ген	Лечение на пигментен ретинит
Croatian	Adeno-pridruženi virus serotip 2/8 vektora koji sadrži humani PDE6A gen	Liječenje retinitisa pigmentoze
Czech	Adeno-asociovaný virový vektor sérotypu 2/8 obsahující lidský *PDE6A* gen	Léčba pigmentosní retinitidy
Danish	Adeno-associeret virusserotype 2/8 vektor, der indeholdender det humane *PDE6A*-gen	Behandling af retinitis pigmentosa
Dutch	Adeno-geassocieerde virus serotype 2/8 vector die het humane *PDE6A*-gen bevat	Behandeling van retinitis pigmentosa
Estonian	Inimese *PDE6A* geeni sisaldav adeno-assotsieerunud viirusvektori serotüüp 2/8	Pigmentoosse võrkkestapõletiku ravi
Finnish	Adeno-assosioituneen viruksen serotyypin 2/8 vektori, joka sisältää ihmisen *PDE6A*-geenin	Retinitis pigmentosan hoito
French	Vecteur de virus adéno-associé de sérotype 2/8 contenant le gène humain *PDE6A*	Traitement de la rétinite pigmentaire
German	Adeno-assoziierter viraler Vektor Serotyp 2/8 der das humane *PDE6A*-Gen enthält.	Behandlung der Retinopathia Pigmentosa
Greek	Αδενο-σχετιζόμενος ιικός φορέας ορότυπου 2/8 που περιέχει το ανθρώπινο γονίδιο *PDE6A*	Θεραπεία της μελαγχρωστικής αμφιβληστροειδοπάθειας
Hungarian	Adeno-asszociált vírus 2/8 szerotípusú vektor, amely a humán *PDE6A* gént tartalmazza	Retinitis pigmentosa kezelése
Italian	Vettore virale virus adeno-associato di sierotipo 2/8 contenente il gene umano *PDE6A*	Trattamento della retinite pigmentosa

[1] At the time of designation

Language	Active ingredient	Indication
Latvian	Adeno asociētais vīrusa 2/8 serotipa vektors, kas satur cilvēka *PDE6A* gēnu	Retinitis pigmentosa ārstēšana
Lithuanian	Adeno-asocijuoto viruso serotipas 2/8, pernešantis žmogaus *PDE6A* geną	Pigmentinio retinito gydymas
Maltese	Vettur ta' serotip 2/8 ta' virus assoċjat ma' adeno li fih il-ġene umana *PDE6A*	Kura tar-retinite pigmentuża
Polish	Wektor wirusowy związany z adenowirusem serotypu 2/8 zawierający ludzki gen *PDE6A*	Leczenie retinopatii barwnikowej
Portuguese	Vetor viral adeno-associado de serotipo 2/8 que contém o gene *PDE6A* humano	Tratamento da retinite pigmentosa
Romanian	Vectoru adeno-asociat cu virusul de serotip 2/8 care conține gena *PDE6A* umană	Tratamentul retinitei pigmentare
Slovak	Adeno-asociovaný vírusový vektor sérotypu 2/8 obsahujúci ľudský gén *PDE6A*	Liečba retinitis pigmentosa
Slovenian	Adeno pridružen virus serotipa 2/8, ki vsebuje človeški gen *PDE6A*	Zdravljenje pigmentozne retinopatije
Spanish	Vector de serotipo 2/8 de virus adenoasociado que contiene el gen *PDE6A* humano	Tratamiento de retinosis pigmentaria
Swedish	Adeno-associerad virusserotyp 2/8 vektor innehållande den humana *PDE6A*-genen	Behandling av retinitis pigmentosa
Norwegian	Adeno-assosiert virusserotype 2/8 vektor som inneholder det humane *PDE6A*-genet	Behandling av retinitis pigmentosa
Icelandic	Adenótengd veiruferja, sermisgerð 2/8, sem inniheldur *PDE6A* genið úr mönnum	Meðferð á sjónufreknum