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Public summary of opinion on orphan designation

Adenovirus-associated viral vector serotype 8 containing the human *RPGR* gene for the treatment of retinitis pigmentosa

On 22 February 2018, orphan designation (EU/3/18/1975) was granted by the European Commission to Nightstar Therapeutics plc, the United Kingdom, for adenovirus-associated viral vector serotype 8 containing the human *RPGR* 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 3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 155,000 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 treating retinitis pigmentosa. Patients with the condition were given sunglasses to slow down damage to the retina, genetic counselling (discussion of the risks of passing the condition on to children) and general support.

[*]Disclaimer: 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 (EU 28), Norway, Iceland and Liechtenstein. This represents a population of 517,400,000 (Eurostat 2018).

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How is this medicine expected to work?

One of the causes of retinitis pigmentosa is mutations (changes) in the *RPGR* gene responsible for the production of the RPGR protein, which is necessary for the normal functioning of retinal cells. In patients with these mutations the RPGR protein is lacking.

The medicine consists of a virus that contains a working copy of the *RPGR* gene. When injected into the patient's eye, under the retina, it is expected that the virus will carry the *RPGR* gene into the retinal cells, enabling them to produce the missing RPGR protein. This is then expected to help the cells in the retina to work better, reducing progression of the condition.

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 this medicine have been evaluated in experimental models.

At the time of submission, clinical trials with the medicine in patients with retinitis pigmentosa were ongoing.

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

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 8 January 2018 recommending the granting of this designation.

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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

Sponsor's contact details:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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	Adenovirus-associated viral vector serotype 8 containing the human *RPGR* gene	Treatment of retinitis pigmentosa
Bulgarian	Аденовирусно-асоцииран вирусен вектор серотип 8, съдържащ човешкия ген *RPGR*	Лечение на пигментен ретинит
Croatian	Adeno-povezani virusni vektor serotipa 8 koji sadrži ljudski gen *RPGR*	Liječenje retinitisa pigmentoze
Czech	Adeno-asociovaný virus sérotypu 8 obsahující lidský gen *RPGR*	Léčba pigmentosní retinitidy
Danish	Adenovirus associeret viral vektor serotype 8 indeholdende det humane gen *RPGR*	Behandling af retinitis pigmentosa
Dutch	Adenovirus geassocieerde virale vector, serotype 8, welke het humane gen *RPGR* bevat	Behandeling van retinitis pigmentosa
Estonian	Adenoviirusega seotud viirusvektor serotüüp 8, mis sisaldab inimese *RPGR* geeni	Pigmentoosse võrkkestapõletiku ravi
Finnish	Serotyypin 8 adenovirusvektori, joka sisältää ihmisen *RPGR*-geenin	Verkkokalvorappeuman hoito
French	Vecteur viral adéno-associé de type 8 contenant le gène humain *RPGR*	Traitement de la rétinite pigmentaire
German	Adeno-assoziierter viraler Vektor Serotyp 8, der das humane *RPGR* Gen enthält	Behandlung der Retinopathia Pigmentosa
Greek	Αδενοσχετιζόμενος ιικός φορέας ορότυπου 8 που περιέχει το ανθρώπινο γονίδιο *RPGR*	Θεραπεία της μελαγχρωστικής αμφιβληστροειδοπάθειας
Hungarian	Humán *RPGR* gént tartalmazó 8-as szerotípusú adenovírus-asszociált virális vektor	Retinitis pigmentosa kezelése
Italian	Vettore virale adenovirus-associato del serotipo 8 contenente il gene umano *RPGR*	Trattamento della retinite pigmentosa
Latvian	Adenovīrusa asociētais 8. seroptipa vīrusa vektors, kas satur cilvēka *RPGR* gēnu	*Retinitis pigmentosa* ārstēšana
Lithuanian	Adeno-asocijuoto viruso vektoriaus 8 serotipas, turintis žmogaus *RPGR* geną	Pigmentinio retinito gydymas
Maltese	Vektor virali assoċjat mal-adenovirus tas-serotip 8 li għandu l-ġene uman *RPGR*	Kura tar-retinite pigmentuża
Polish	Wektor adenowirusowy serotypu 8 zawierający ludzki gen *RPGR*	Leczenie retinopatii barwnikowej
Portuguese	Vetor viral adeno-associado de serotipo 8 contendo o gene humano *RPGR*	Tratamento da retinite pigmentosa
Romanian	Vector viral adeno-asociat de serotip 8 ce conţine gena umană *RPGR*	Tratamentul retinitei pigmentare
Slovak	Adeno-asociovaný vírusový vektor sérotypu 8 obsahujúci ľudský gén *RPGR*	Liečba retinitis pigmentosa
Slovenian	Adenovirusom pridružen virusni vektor serotipa 8,	Zdravljenje pigmentozne

[1] At the time of designation

Language	Active ingredient	Indication
	ki vsebuje človeški gen *RPGR*	retinopatije
Spanish	Vector vírico adenoasociado del serotipo 8 que conteniene el gen humano *RPGR*	Tratamiento de la retinosis pigmentaria
Swedish	Adenoassocierad virusvektor av serotyp 8, innehållande den humana *RPGR* genen	Behandling av retinitis pigmentosa
Norwegian	Adenoassosiert virusvektor serotype 8 som inneholder det humane genet *RPGR*	Behandling av retinitis pigmentosa
Icelandic	Adenó tengd veiruferja af sermisgerð 8 sem inniheldur manna *RPGR* gen	Meðferð á retinitis pigmentosa