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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant adeno-associated viral vector containing the human *RPGR* gene for the treatment of retinitis pigmentosa caused by mutations in the *RPGR* gene

On 30 May 2016, orphan designation (EU/3/16/1665) was granted by the European Commission to TMC Pharma Services Ltd, United Kingdom, for recombinant adeno-associated viral vector containing the human *RPGR* gene for the treatment of retinitis pigmentosa caused by mutations in the *RPGR* gene.

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.

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 this form of the disease the RPGR protein is lacking.

Retinitis pigmentosa caused by mutations in the *RPGR* gene 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 caused by mutations in the *RPGR* gene was estimated to affect approximately 0.7 in 10,000 people in the European Union (EU). This was equivalent to a total of around 36,000 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).

*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 513,700,000 (Eurostat 2016).

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 the damage to the retina, genetic counselling (discussion of the risks of passing the condition on to children) and general support.

How is this medicine expected to work?

The medicine consists of a virus that contains a normal 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 function 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 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 retinitis pigmentosa caused by mutations in the *RPGR* gene had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for retinitis pigmentosa caused by mutations in the *RPGR* gene 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 21 April 2016 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

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 languages¹, Norwegian and Icelandic

Language	Active ingredient	Indication
English	Recombinant adeno-associated viral vector containing the human <i>RPGR</i> gene	Treatment of retinitis pigmentosa caused by mutations in the <i>RPGR</i> gene
Bulgarian	Рекомбинантен адено-асоцииран вирусен вектор, съдържащ човешкия ген <i>RPGR</i>	Лечение на пигментен ретинит, предизвикан от мутации в <i>RPGR</i> гена
Croatian	Rekombinantni adeno-povezani virusni vektor koji sadrži ljudski <i>RPGR</i> gen	Liječenje retinitis pigmentoze uzrokovane mutacijama gena <i>RPGR</i>
Czech	Rekombinantní adeno-asociovaný virový vektor obsahující lidský gen <i>RPGR</i>	Léčba pigmentosní retinitidy způsobené mutacemi v genu <i>RPGR</i>
Danish	Rekombinant adeno-associeret viral vektor som indeholder det humane <i>RPGR</i> -gen	Behandling af retinitis pigmentosa, forårsaget af mutationer i <i>RPGR</i> -genet
Dutch	Recombinant adeno-geassocieerde virale vector welke het humaan <i>RPGR</i> gen	Behandeling van door mutaties in het <i>RPGR</i> -gen veroorzaakte retinitis pigmentosa
Estonian	Inimese <i>RPGR</i> geeni sisaldav rekombinantne adenoviirusega seotud viirusvektor	<i>RPGR</i> geeni mutatsioonidest tingitud pigmentoosse võrkkestapõletiku ravi
Finnish	Rekombinantti adeno-pohjainen virusvektori, joka sisältää ihmisen <i>RPGR</i> -geenin	<i>RPGR</i> -geenin mutaation aiheuttaman verkkokalvorappeuman hoito
French	Vecteur viral recombinant adéno-associé contenant le gène humain <i>RPGR</i>	Traitement de la rétinite pigmentaire causée par des mutations du gène <i>RPGR</i>
German	Rekombinanter Adeno-assoziiierter viraler Vektor, der das menschliche Gen <i>RPGR</i> enthält	Behandlung der Retinopathia Pigmentosa infolge von Mutationen des <i>RPGR</i> -Gens
Greek	Ανασυνδυασμένος ιϊκός φορέας σχετιζόμενος με αδενοϊό που περιέχει το ανθρώπινο γονίδιο <i>RPGR</i>	Θεραπεία της μελαγχρωστικής αμφιβληστροειδοπάθειας που προκαλείται από μεταλλάξεις στο γονίδιο <i>RPGR</i>
Hungarian	Humán <i>RPGR</i> gén tartalmazó rekombináns adeno asszociált virális vektor	<i>RPGR</i> gén mutációk által okozott Retinitis pigmentosa kezelése
Italian	Vettore virale ricombinante adeno-associato contenente il gene della <i>RPGR</i> umana	Trattamento della retinite pigmentosa causata da mutazioni del gene <i>RPGR</i>
Latvian	Rekombinants adenoasociētā vīrusa vektors, kas satur cilvēka <i>RPGR</i> gēnu	<i>RPGR</i> gēna mutāciju izraisīta retinitis pigmentosa ārstēšana

¹ At the time of designation

Language	Active ingredient	Indication
Lithuanian	Rekombinantinis adeno-asocijuoto viruso vektorius, pernešantis žmogaus <i>RPGR</i> geną	<i>RPGR</i> geno mutacijų sąlygoto pigmentinio retinito gydymas
Maltese	Vettur virali rikombinanti adeno-assocjat li fih il-ġene <i>RPGR</i> uman	Kura tar-retinite pigmentuża kkawżata minn mutazzjonijiet fil-ġene <i>RPGR</i>
Polish	Rekombinowany wektor adenowirusowy zawierający ludzki gen <i>RPGR</i>	Leczenie retinopatii barwnikowej wywołanej mutacjami genu <i>RPGR</i>
Portuguese	Vetor viral recombinante adeno-associado, contendo o gene <i>RPGR</i> humano	Tratamento da retinite pigmentosa causada pelas mutações no gene <i>RPGR</i>
Romanian	Vector viral adeno-asociat recombinant ce conține gena <i>RPGR</i> umană	Tratamentul retinitei pigmentare produse de mutații ale genei <i>RPGR</i>
Slovak	Rekombinantný adeno-asociovaný vírusový vektor obsahujúci ľudský gén <i>RPGR</i>	Liečba retinitis pigmentosa spôsobenej mutáciami v géne <i>RPGR</i>
Slovenian	Rekombinantni adenovirusni vector, ki vsebuje humani <i>RPGR</i> gen	Zdravljenje pigmentozne retinopatije, ki jo povzročijo mutacije gena <i>RPGR</i>
Spanish	Vector viral adenoasociado recombinante que contiene el gen humano de la <i>RPGR</i>	Tratamiento de la retinosis pigmentaria causada por mutaciones en <i>RPGR</i>
Swedish	Rekombinant adeno-associerad virusvektor innehållande den humana <i>RPGR</i> -genen	Behandling av retinitis pigmentosa orsakad av mutationer i <i>RPGR</i> -genen
Norwegian	Rekombinant adenoassosiert viral vektor som inneholder det humane <i>RPGR</i> -genet	Behandling av retinitis pigmentosa forårsaket av mutasjoner i <i>RPGR</i> -genet
Icelandic	Raðbrigða adenótengd veiru ferja sem inniheldur manna <i>RPGR</i> gen	Meðferð á retinitis pigmentosa af völdum stökkbreytinga í <i>RPGR</i> geninu