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Public summary of opinion on orphan designation

Adeno-associated virus serotype 8 containing the human RdCVF sequence and the human RdCVFL sequence for the treatment of inherited retinal dystrophies

On 28 February 2020, orphan designation EU/3/20/2249 was granted by the European Commission to SparingVision, France, for adeno-associated virus serotype 8 containing the human RdCVF sequence and the human RdCVFL sequence (also known as SPVN06) for the treatment of inherited retinal dystrophies.

What are inherited retinal dystrophies?

Inherited retinal dystrophies are a group of hereditary diseases of the eye that lead to progressive loss of sight. In patients with inherited retinal dystrophies, cells in the retina (the light-sensitive surface at the back of the eye) become damaged and eventually die.

Inherited retinal dystrophies are debilitating in the long term because they cause 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, IRD affected approximately 3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 156,000 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 medicines Raxone (idebenone) and Luxturna (voretigene neparvovec) were authorised in the EU for the treatment of certain subtypes of inherited retinal dystrophies.

The sponsor has provided sufficient information to show that the medicine might improve vision in patients with certain types of inherited retinal dystrophies, for whom no authorised products are

*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 519,200,000 (Eurostat 2020).

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available. This assumption is based on data from studies in the laboratory and 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?

Inherited retinal dystrophies are caused by mutations (changes) in the genes responsible to produce the proteins necessary for the normal functioning of retinal cells. In patients with these mutations the proteins are lacking.

The medicine consists of a virus that contains two genes, RdCVF and RdCVFL, involved in normal functioning of retinal cells and preventing them from damage. When injected into the patient's eye, under the retina, it is expected that the virus will carry the genes into the retinal cells, thereby helping the cells to function better and reducing progression of the condition, regardless of the genetic mutation causing 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 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 inherited retinal dystrophies had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of IRD 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 22 January 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

Sponsor’s contact details:

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 8 containing the human RdCVF sequence and the human RdCVFL sequence	Treatment of inherited retinal dystrophies
Bulgarian	Адено-асоцииран вирус, серотип 8, съдържащ човешката RdCVF последователност и човешката RdCVFL последователност	Лечение на наследствени ретинални дистрофии
Croatian	Adeno-povezani virus serotipa 8 koji sadrži ljudsku sekvencu RdCVF i ljudsku sekvencu RdCVFL	Liječenje nasljednih mrežničnih distrofija
Czech	Adeno-asociovaný virus sérotypu 8 obsahující humánní RdCVF sekvenci a humánní RdCVFL sekvenci	Léčba dědičných retinálních dystrofií
Danish	Adenoassocieret virus serotype 8 indeholdende den humane RdCVF-sekvens og den humane RdCVFL-sekvens	Behandling af arvelige nethinde dystrofier
Dutch	Adeno-geassocieerd virus serotype 8 met de humane RdCVF-sequentie en de humane RdCVFL-sequentie	Behandeling van erfelijke retinale dystrofieën
Estonian	Inimese RdCVF järjestust ja inimese RdCVFL järjestust sisaldav adeno-assotsieerunud viiruse serotüüp 8.	Pärilike reetina düstroofiate ravi
Finnish	AA-viruksen serotyyppi 8, joka sisältää ihmisperäisen RdCVF-sekvenssin ja ihmisperäisen RdCVFL-sekvenssin	Perinnöllisten verkkokalvorappeumien hoito
French	Virus adéno-associé de sérotype 8 contenant la séquence humaine RdCVF et la séquence humaine RdCVFL	Traitement des dystrophies rétiniennes héréditaires
German	Adeno-assoziiertes Virus Serotyp 8 mit der humanen RdCVF-Sequenz und der humanen RdCVFL-Sequenz	Behandlung von hereditären Netzhautdystrophien
Greek	Αδενο-σχετιζόμενος ιός οροτύπου 8 που περιέχει την ανθρώπινη αλληλουχία RdCVF και την ανθρώπινη αλληλουχία RdCVFL	Θεραπεία των κληρονομικών δυστροφιών του αμφιβληστροειδούς
Hungarian	Humán RdCVF és RdCVFL szekvenciákat tartalmazó, 8-as szerotípusú adeno-asszociált vírusvektor	Örökletes retina dystrophiák kezelése
Italian	Virus adeno-associato di sierotipo 8 contenente le sequenze umane RdCVF e RdCVFL	Trattamento della distrofia retinica ereditaria

[1] At the time of designation

Language	Active ingredient	Indication
Latvian	Cilvēka RdCVF sekvenci un cilvēka RdCVFL sekvenci saturošs adenoasociētais vīrusa 8. serotips	Iedzimtu tīklenes distrofiju ārstēšana
Lithuanian	Su adeno virusu susijęs 8 serotipas, turintis žmogaus RdCVF seką ir žmogaus RdCVFL seką	Paveldimų tinklainės distrofijų gydymas
Maltese	Virus serotip 8 adeno-assoċjat li fih is-sekwenza RdCVF tal-bniedem u s-sekwenza RdCVFL tal-bniedem	Kura ta' distrofiji tar-retina li jintirtu
Polish	Wirus związany z adenowirusami serotypu 8 zawierający ludzką sekwencję RdCVF i ludzką sekwencję RdCVFL	Leczenie dziedzicznych dystrofii siatkówki
Portuguese	Vírus adeno-associado de serotipo 8 contendo a sequência RdCVF humana e a sequência RdCVFL humana	Tratamento de distrofias retinianas congénitas
Romanian	Virus adeno-asociat de serotip 8 conținând secvența RdCVF umană și secvența RdCVFL umană	Tratamentul distrofiilor retiniene ereditare
Slovak	Adeno-asociovaný vírus sérotypu 8 vybavený sekvenciou ľudského RdCVF a sekvenciou ľudského RdCVFL	Liečba vrodených retinálnych dystrofií
Slovenian	Adenoasociacijski virusni serotip 8, ki vsebuje človeško zaporedje RdCVF in človeško zaporedje RdCVFL	Zdravljenje dedne distrofije mrežnice
Spanish	Virus adenoasociado de serotipo 8 que contiene la secuencia humana RdCVF y la secuencia humana RdCVFL	Tratamiento de distrofias retinianas hereditarias
Swedish	Adenoassocierad virus serotyp 8 innehållande den humana RdCVF-sekvensen och den humana RdCVFL-sekvensen	Behandling av ärftliga retinala dystrofier
Norwegian	Adenoassosiert virus, serotype 8, som inneholder den humane RdCVF-sekvensen og den humane RdCVFL-sekvensen	Behandling av arvelige netthinnedystrofier
Icelandic	Adenótengd veira af sermisgerð 8 sem inniheldur manna RdCVF-röðina og manna RdCVFL-röðina	Meðferð við arfgengum sjónukyrking