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Public summary of opinion on orphan designation

Recombinant adeno-associated viral vector serotype 5 carrying the gene for the human frataxin protein for the treatment of Friedreich's ataxia

On 23 August 2017, orphan designation (EU/3/17/1906) was granted by the European Commission to Voisin Consulting S.A.R.L., France, for recombinant adeno-associated viral vector serotype 5 carrying the gene for the human frataxin protein (also known as AGIL-FA) for the treatment of Friedreich's ataxia.

What is Friedreich's ataxia?

Friedreich's ataxia is an inherited disease that causes a range of symptoms that worsen over time, including difficulty walking, inability to co-ordinate movements, muscle weakness, speech problems, damage to the heart muscle, and diabetes.

Patients with Friedreich's ataxia do not have enough frataxin, a protein that regulates iron in mitochondria (energy-producing components of cells). As a result iron builds up within the cells, which in turns results in the production of toxic forms of oxygen that damage cells in the brain, the spinal cord and nerves, as well as in the heart and pancreas.

Friedreich's ataxia is a debilitating and life-threatening disease because of the worsening of symptoms over time. The disease is usually fatal in early adulthood.

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

At the time of designation, Friedreich's ataxia affected approximately 0.5 in 10,000 people in the European Union (EU). This was equivalent to a total of around 26,000 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 Friedreich's ataxia. Different treatments were used to relieve the symptoms of the disease, such as

*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 515,700,000 (Eurostat 2017).

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medicines for diabetes and heart problems. Patients were also offered walking aids to allow them to remain as independent as possible, and other devices to assist them with everyday tasks such as eating and taking care of themselves. Speech therapy and physiotherapy were also used.

How is this medicine expected to work?

This medicine consists of a virus that contains a gene that produces frataxin, the protein lacking in patients with Friedreich's ataxia. When given directly into the brain of patients, the virus is expected to carry this gene into the brain cells. This would enable these cells to produce frataxin, thereby helping the cells to regulate iron normally, and is expected to improve the symptoms 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 this medicine in patients with Friedreich's ataxia had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for Friedreich's ataxia. Orphan designation of the medicine had been granted in the United States for this condition.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 13 July 2017 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	Recombinant adeno-associated viral vector serotype 5 carrying the gene for the human frataxin protein	Treatment of Friedreich's ataxia
Bulgarian	Рекомбинантен адено-свързан вирусен вектор, серологичен тип 5, носител на гена за човешкия протеин фратаксин	Лечение на атаксия на Фридрайх
Croatian	Rekombinantni adeno-povezani virusni vektor serotipa 5 koji nosi gen za ljudski protein fraktiksin	Liječenje Friedreichove ataksije
Czech	Rekombinantní adeno-asociovaný vector serotypu 5 nesoucí gen lidského frataxin proteinu	Léčba Friedrichovy ataxie
Danish	Rekombinant adeno-associeret viral vektor serotype 5, der bærer genet for humant frataxin proteinet	Behandling af Friedreichs ataksi
Dutch	Recombinant adeno-associëerde virale vector serotype 5 welke het gen voor humaan frataxin proteïne bevat	Behandeling van de ataxie van Friedreich
Estonian	Rekombinantne adenoviirusega assotseerunud viirusvektori serotüüp 5, mis kannab inimese frataksiini proteiini geeni	Friedreichi ataksia ravi
Finnish	Rekombinantti adenoassosioitu virusvektori, serotyyppi 5, joka sisältää ihmisen frataksiini proteiinin	Friedreichin ataksian hoito
French	Vecteur viral adéno-associé de sérotype 5 portant le gene humain de la protein frataxine	Traitement de l'ataxie de Friedreich
German	Rekombinanter adeno-assoziierter viraler Vektor vom Serotyp 5, der das Gen für das humane Frataxin Protein enthält	Therapie der Friedreichschen Ataxie
Greek	Ανασυνδυασμένος αδενο-σχετιζόμενος ιικός φορέας οροτύπου 5 φέρων το γονίδιο της ανθρώπινης πρωτεΐνης φραταξίνης	Θεραπεία της αταξίας Friedreich
Hungarian	Humán frataxin protein génjét hordozó 5-ös szerotípusú rekombináns adeno-asszociált vírus vektor	Friedreich ataxia kezelése
Italian	Vettore virale adeno-associato di sierotipo 5 contenente il gene umano della proteina frataxina	Trattamento dell'atassia di Friedreich
Latvian	Rekombinants adeno-asociētā vīrusa 5. serotipa vektors, kas pārnes cilvēka frataksīna proteīna gēnu	Frīdreiha ataksijas ārstēšana
Lithuanian	Rekombinantinis adenoasocijuoto viruso vektoriaus serotipas 5, nešantis žmogaus frataksino baltymo geną	Fridreicho ataksijos gydymas
Maltese	Vettur virali serotip 5 rikombinanti assoċjat ma□ adeno li jġorr il-ġene għall-proteina umana fratassin	Kura tal-atassja ta' Friedreich

[1] At the time of designation

Language	Active ingredient	Indication
Polish	Rekombinowany wektory wirusowy związany z adenowirusami serotypu 5 niosący gen ludzkiego białka frataksyny	Leczenie ataksji Friedreicha
Portuguese	Vetor viral recombinante adeno-associado de serotipo 5 contendo o gene da proteína de frataxina humana	Tratamento da ataxia de Friedreich
Romanian	Vector viral adeno-asociat de serotip 5 ce conţine gena umană a proteinei frataxină	Tratamentul ataxiei Friedreich
Slovak	Rekombinantný adeno-asociovaný vírusový vektor sérotypu 5 nesúci gén pre ľudský frataxín proteín	Liečba Friedreichovej ataxie
Slovenian	Rekombninantni adenovirusom pridruženi virusni vektor serotipa 5, ki nosi gen za humani frataksin protein	Zdravljenje Friedreichove atakcije
Spanish	Vector viral adenoasociado recombinante de serotipo 5 que contiene el gen para la proteina fractaxina	Tratamiento de la ataxia de Friedreich
Swedish	Rekombinant adeno-associerad viral vektor av serotype 5 innehållande genen för humanproteinet frataxin	Behandling av Friedreichs ataxi
Norwegian	Rekombinant adeno-assosiert viral vektor serotype 5 som bærer genet for humant frataxin protein	Behandling av Friedreichs ataksi
Icelandic	Raðbrigða adenó-tengdur veiru vector sermisgerð 5 sem ber gen fyrir manna frataxin próteini	Meðferð arfgengs mænuslingurs