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Public summary of opinion on orphan designation

Adeno-associated virus serotype 9 containing the human ASPA gene for the treatment of Canavan disease

On 4 June 2020, orphan designation EU/3/20/2287 was granted by the European Commission to Raremoon Consulting Limited, United Kingdom, for adeno-associated virus serotype 9 containing the human ASPA gene (also known as BBP-812) for the treatment of Canavan disease.

What is Canavan disease?

Canavan disease is an inherited disease that affects the normal functioning of the nerves, the brain and spinal cord.

The disease is caused by a mutation (change) in the gene responsible for producing an enzyme called ASPA, which is involved in maintaining myelin, a protective sheath around nerve cells that also speeds up the transmission of impulses (messages) along nerves. Because of these mutations, ASPA does not work properly leading to problems with the development of the brain and spinal cord and messaging between nerves throughout the body.

The disease usually starts at birth and affected infants do not develop normally and have an unusually large head size. As the disease progresses, patients develop loss of muscle strength, damage to the optic nerve and stiffening of muscles. Patients often have swallowing difficulties, sleep disturbances, inability to move voluntarily and experience seizures (fits).

Canavan disease is long-term debilitating and life threatening with a life expectancy of less than 10 years for the severe form of the disease.

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

At the time of designation, Canavan disease affected approximately 0.04 in 10,000 people in the European Union (EU). This was equivalent to a total of around 2,000 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).

*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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What treatments are available?

At the time of designation, no satisfactory methods were authorised in the EU for the treatment of Canavan disease. Patients received supportive treatment to protect airways and provide nutrition and hydration. Patients also received treatment to temporarily relieve the symptoms of the disease, such as medicines to treat epilepsy and spasticity.

How is this medicine expected to work?

This medicine is made of a virus that contains a normal copy of the gene for ASPA. When given to the patient, the virus is expected to carry the gene into the nerve cells in the brain, enabling them to produce a working ASPA, thus reducing the damage to myelin and maintaining their normal functioning.

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 Canavan disease had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for the treatment of Canavan disease 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 23 April 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 9 containing the human *ASPA* gene	Treatment of Canavan disease
Bulgarian	Адено-асоцииран вирус, серотип 9, съдържащ човешкия ген *ASPA*	Лечение на болестта на Канаван
Croatian	Adeno-pridruženi virus, serotipa 9 koji sadrži humani gen *ASPA*	Liječenje Kanavanove bolesti
Czech	Adeno-asociovaný virus sérotypu 9 obsahující lidský *ASPA* gen	Léčba nemoci Canavanové
Danish	Adenoassocieret virus serotype 9, indeholdende det humane *ASPA*-gen	Behandling af Canavans sygdom
Dutch	Adeno-geassocieerd virus, serotype 9 dat het menselijke *ASPA*-gen bevat	Behandeling van de ziekte van Canavan
Estonian	Inimese *ASPA* geeni sisaldav adeno-assotsieerunud viiruse serotüüp 9	Canavani haiguse ravi
Finnish	Adeno-assosioituneen viruksen serotyyppi 9, joka sisältää ihmisen *ASPA*-geenin	Canavanin taudin hoito
French	Virus adéno-associé de sérotype 9 contenant le gène *ASPA* humain	Traitement de la maladie de Canavan
German	Adeno-assoziiertes Virus, Serotyp 9, welches das menschliche *ASPA*-Gen enthält	Behandlung der Canavan-Krankheit
Greek	Αδενο-σχετιζόμενος ιός ορότυπου 9 που περιέχει το ανθρώπινο γονίδιο *ASPA*	Θεραπεία της νόσου Canavan
Hungarian	Adeno-asszociált vírus 9. szerotípus, amely humán *ASPA* gént tartalmaz	Canavan-betegség kezelése
Italian	Virus adeno-associato del sierotipo 9 recante il gene umano *ASPA*	Trattamento della sindrome di Canavan
Latvian	Adenosaistītā vīrusa 9 serotips, kas satur cilvēka *ASPA* gēnu	Kanavana slimības ārstēšana
Lithuanian	Adeno asocijuoto viruso 9 serotipas, turintis žmogaus *ASPA* geną	Canavan ligos gydymas
Maltese	Serotip 9 tal-virus adenoassoċjat li fih il-ġene *ASPA* uman	Trattament tal-marda ta' Canavan
Polish	Wektor wirusowy związany z adenowirusami serotypu 9 zawierający ludzki gen *ASPA*	Leczenie choroby Canavana
Portuguese	Vírus adeno-associado de serotipo 9 contendo o gene humano *ASPA*	Tratamento da doença de Canavan
Romanian	Virus adeno-asociat de serotip 9 care conține gena umană *ASPA*	Tratamentul bolii Canavan
Slovak	Adeno-asociovaný vírus, sérotypu 9 obsahujúci ľudský gén *ASPA*	Liečba Canavanovej choroby

[1] At the time of designation

Language	Active ingredient	Indication
Slovenian	Adeno-povezan virusni serotip 9, ki vsebuje človeški gen *ASPA*	Zdravljenje Canavanove bolezni
Spanish	Virus adenoasociado de serotipo 9 que contiene el gen humano de *ASPA*	Tratamiento de la enfermedad de Canavan
Swedish	Adenoassosierat virus serotyp 9 som innehåller den mänskliga *ASPA*-genen	Behandling av Canavans sjukdom
Norwegian	Adenoassosiert virus serotype 9 som inneholder det humane *ASPA*-genet	Behandling av Canavans sykdom
Icelandic	Adenótengd veiruferja af sermisgerð 9 sem inniheldur *ASPA*-gen manna.	Meðferð við Canavan-sjúkdómi