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Public summary of opinion on orphan designation

Venglustat for treatment of autosomal dominant polycystic kidney disease

On 14 December 2018, orphan designation (EU/3/18/2122) was granted by the European Commission to Genzyme Europe BV, the Netherlands, for venglustat for treatment of autosomal dominant polycystic kidney disease.

What is autosomal dominant polycystic kidney disease?

Polycystic kidney disease is an inherited condition marked by the growth of numerous fluid-filled cysts mainly in the kidneys. The growth of cysts eventually affects kidney function and can cause the kidneys to fail. Symptoms include abdominal (belly) pain, problems with urinating, high blood pressure and infection.

In most cases, polycystic kidney disease is 'autosomal dominant', which means that it is caused by gene mutations (changes) that are 'dominant' because a person can have the disease even if only one parent has passed on the mutated gene. Autosomal dominant polycystic kidney disease is caused by a mutation of either of two genes, *PKD1* and *PKD2*.

Autosomal dominant polycystic kidney disease is debilitating in the long term and life threatening because patients can develop kidney failure and also problems in other organs such as the heart and the gut.

What is the estimated number of patients affected by autosomal dominant polycystic kidney disease?

At the time of designation, orphan autosomal dominant polycystic kidney disease affected approximately 4.7 in 10,000 people in the European Union (EU). This was equivalent to a total of around 243,000 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 autosomal dominant polycystic kidney 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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What treatments are available?

At the time of designation, Jinarc (tolvaptan) was authorised in the EU for the treatment of autosomal dominant polycystic kidney disease.

The sponsor has provided sufficient information to show that venglustat might be of significant benefit for patients with autosomal dominant polycystic kidney disease. Early data indicate that the medicine may have beneficial effects in patients who cannot take tolvaptan. This assumption 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?

Venglustat is expected to work in autosomal dominant polycystic kidney disease by blocking the production of certain fatty subtances called glycosphingolipid. An excess of these fatty subtances in cells is thought to contribute to the formation of cysts. By blocking the production of these substances, venglustat is expected to reduce the formation of cysts in the kidney and also in the liver. This is expected to improve the symptoms of the disease.

What is the stage of development of this medicine?

The effects of venglustat have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with venglustat in patients with autosomal dominant polycystic kidney disease were ongoing.

At the time of submission, venglustat was not authorised anywhere in the EU for autosomal dominant polycystic kidney disease 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 November 2018 recommending the granting of this designation.

Opinions on orphan medicinal product designations are based on the following three criteria:

- the seriousness of the autosomal dominant polycystic kidney disease;
- the existence of alternative methods of diagnosis, prevention or treatment;
- either the rarity of the autosomal dominant polycystic kidney disease (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 the 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	Venglustat	Treatment of autosomal dominant polycystic kidney disease
Bulgarian	Венглустат	Лечение на автозомно-доминантна поликостозна бъбречна болест
Croatian	Venglustat	Liječenje autosomno dominantne policistične bolesti bubrega
Czech	Venglustat	Léčba autozomálně dominantní polycystózy ledvin
Danish	Venglustat	Behandling af autosomal dominant polycystisk nyresygdom
Dutch	Venglustat	Behandeling van autosomale dominante polycystische nierziekte
Estonian	Venglustaat	Autosoom-dominantse polütsüstilise neeruhaiguse ravi
Finnish	Venglustaatti	Autosomaalisen dominantin polykystisen munuaistaudin hoito
French	Venglustat	Traitement de la polykystose rénale autosomique dominante
German	Venglustat	Behandlung der autosomal-dominanten polyzystischen Nierenerkrankung
Greek	Βενγλουστάτη	Θεραπεία της αυτοσωματικής κυρίαρχης πολυκυστικής νόσου των νεφρών
Hungarian	Venglusztát	Autoszomális domináns policisztás vesebetegség kezelése
Italian	Venglustat	Trattamento della malattia renale policistica autosomica dominante
Latvian	Venglustats	Autosomāli dominantas nieru policistozes ārstēšana
Lithuanian	Venglustatas	Autosominės dominantinės policistinės inkstų ligos gydymas
Maltese	Venglustat	Kura tal-marda poliċistika tal-kliewi awtosomali dominanti
Polish	Wenglustat	Leczenie autosomalnie dominującej wielotorbielowatości nerek
Portuguese	Venglustate	Tratamento da doença renal poliquística autossómica dominante
Romanian	Venglustat	Tratamentul bolii polichistice renale cu transmitere autozomal dominantă
Slovak	Venglustat	Liečba autozomálneho dominantného polycystického ochorenia obličiek
Slovenian	Venglustat	Zdravljenje avtosomne dominantne policistične bolezni ledvic
Spanish	Venglustat	Tratamiento de la poliquistosis renal autosómica dominante
Swedish	Venglustat	Behandling av autosomalt dominant polycystisk njursjukdom
Norwegian	Venglustat	Behandling av autosomal dominant polycystisk nyresykdom
Icelandic	Venglustat	Meðferð við nýrnafjölblöðrusjúkdómi með ríkjandi erfðamáta

[1] At the time of designation