EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Cysteamine for the treatment of cystic fibrosis

On 19 February 2014, orphan designation (EU/3/14/1240) was granted by the European Commission to 'Istituto Europeo per la Ricerca sulla Fibrosi Cistica' – ONLUS, Italy, for cysteamine for the treatment of cystic fibrosis.

What is cystic fibrosis?

Cystic fibrosis is a hereditary disease that affects the cells in the lungs, and the glands in the gut and pancreas, that secrete fluids such as mucus and digestive juices. In cystic fibrosis, these fluids become thick and viscous, blocking the airways and the flow of digestive juices. This leads to long-term infection and inflammation of the lungs because of excess mucus not being cleared away, and to problems with the digestion and absorption of food, resulting in poor growth.

Cystic fibrosis is caused by abnormalities in a gene that makes a protein called 'cystic-fibrosis transmembrane conductance regulator' (CFTR), which is involved in regulating the production of mucus and digestive juices.

Cystic fibrosis is a long-term debilitating and life-threatening disease because it severely damages the lung tissue, leading to problems with breathing and to recurrent chest infections.

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

At the time of designation, cystic fibrosis affected 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).

What treatments are available?

At the time of designation, lung infection in cystic fibrosis was mainly treated with antibiotics. Kalydeco (ivacaftor) was authorised to correct the defect of the CFTR protein in a subgroup of patients with

*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 511,100,000 (Eurostat 2014).

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cystic fibrosis with the G551D mutation. Other medicines used to treat the lung disease included anti-inflammatory agents, bronchodilators (medicines that help to open up the airways in the lungs) and mucolytics (medicines that help dissolve the mucus in the lungs). In addition, patients with cystic fibrosis were often given other types of medicines such as pancreatic enzymes (substances that help to digest and absorb food) and food supplements. They were also advised to exercise and to undergo physiotherapy.

The sponsor has provided sufficient information to show that cysteamine might be of significant benefit for patients with cystic fibrosis because early studies in experimental models show that it works in a different way to existing treatments and that it might be used to treat cystic fibrosis patients with the most common genetic abnormality (F508D). 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?

Cysteamine is expected to work in patients with cystic fibrosis by improving the ability of the defective CFTR protein to function in patients with the most common genetic abnormality (F508D). Cysteamine may also reduce inflammation in the airways of cystic fibrosis patients, thereby improving the symptoms of the disease.

This medicine is expected to be taken by mouth.

What is the stage of development of this medicine?

The effects of cysteamine have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with this medicine in patients with cystic fibrosis were planned.

At the time of submission, cysteamine was authorised as Cystagon and Procysbi in the EU for the treatment of nephropathic (kidney) cystinosis.

At the time of submission, this medicine was not authorised anywhere in the EU for cystic fibrosis. Cysteamine had already been granted orphan designation in the EU for cystic fibrosis.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 9 January 2014 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:

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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	Cysteamine	Treatment of cystic fibrosis
Bulgarian	Цистеамин	Лечение на кистозна фиброза
Czech	Cysteamin	Léčba cystické fibrózy
Croatian	Cisteamin	Liječenje cistične fibroze
Danish	Cysteamin	Behandling af cystisk fibrose
Dutch	Cysteamine	Behandeling van cystische fibrose
Estonian	Tsüsteamiin	Tsüstilise fibroosi ravi
Finnish	Kysteamiini	Kystisen fibroosin hoito
French	Cystéamine	Traitement de la mucoviscidose
German	Cysteamin	Behandlung zystischer Fibrose
Greek	Κυστεαμίνη	Θεραπεία της κυστικής ίνωσης
Hungarian	Ciszteamin	Cisztikus fibrózis kezelése
Italian	Cisteamina	Trattamento della fibrosi cistica
Latvian	Cisteamīns	Cistiskās fibrozes ārstēšana
Lithuanian	Cisteaminas	Cistinės fibrozės gydymas
Maltese	Cysteamine	Kura tal-fibrożi ċistiku
Polish	Cysteamina	Leczenie zwłóknienia torbielowatego
Portuguese	Cisteamina	Tratamento da fibrose quística
Romanian	Cisteamină	Tratamentul fibrozei chistice
Slovak	Cysteamín	Terapia cystickej fibrózy
Slovenian	Cisteamin	Zdravljenje cistične fibroze
Spanish	Cisteamina	Tratamiento de la fibrosis quística
Swedish	Cysteamin	Behandling av cystisk fibros
Norwegian	Cysteamin	Behandling av cystisk fibrose
Icelandic	Sýsteamín	Meðferð við slímseigjusjúkdómi

[1] At the time of designation