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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Sacrosidase for the treatment of congenital sucrase-isomaltase deficiency

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Disclaimer

Please note that revisions to the Public Summary of Opinion are purely administrative updates. Therefore, the scientific content of the document reflects the outcome of the Committee for Orphan Medicinal Products (COMP) at the time of designation and is not updated after first publication.

On 5 August 2013, orphan designation (EU/3/13/1183) was granted by the European Commission to QOL Therapeutics EU Ltd, United Kingdom, for sacrosidase for the treatment of congenital sucrase-isomaltase deficiency.

In March 2014, QOL Therapeutics EU Ltd changed name to QOL Therapeutics UK Ltd.

What is congenital sucrase-isomaltase deficiency?

Congenital sucrase-isomaltase deficiency is a disease caused by defects in the gene for sucrase-isomaltase, an enzyme involved in breaking down sucrose (sugar) and starch in the intestines. Patients with this disease do not have enough of the correctly functioning enzyme and therefore cannot break down the sucrose and starch in their diet. This leads to a build-up of sucrose and starch in the gut, which can cause severe gastrointestinal symptoms. It also stops the patient from absorbing these nutrients, leading to long-term malnutrition and failure to thrive and develop.

Congenital sucrase-isomaltase deficiency is a long-term debilitating disease as patients fail to thrive and develop normally. It can also cause kidney stones and high calcium levels in the kidneys and in the blood, and increased acidity of the blood due to kidney problems.

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

At the time of designation, congenital sucrase-isomaltase deficiency affected approximately 2 in 10,000 people in the European Union (EU). This was equivalent to a total of around 102,000 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 treatments for congenital sucrase-isomaltase deficiency were authorised in the EU. The disease was usually managed through lifelong dietary measures to eliminate sucrose from the diet.

How is this medicine expected to work?

Sacrosidase is an enzyme found in yeast which is able to break down sucrose. When given orally to the patient, sacrosidase is expected to break down sucrose in the patient's body, thereby substituting an important aspect of the action of the missing enzyme. Since sacrosidase does not break down starch, it cannot fully substitute the function of the missing enzyme, and it is therefore expected to be used together with a low starch diet.

What is the stage of development of this medicine?

The effects of sacrosidase have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with sacrosidase in patients with congenital sucrase-isomaltase deficiency had finished.

At the time of submission, sacrosidase was authorised in the United States for congenital sucrase-isomaltase deficiency.

At the time of submission, sacrosidase was not authorised anywhere in the EU for congenital sucrase-isomaltase deficiency but was available for use on a named-patient basis.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 11 July 2013 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.

[*]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 27), Norway, Iceland and Liechtenstein.
At the time of designation, this represented a population of 512,200,000 (Eurostat 2013).

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	Sacrosidase	Treatment of congenital sucrase-isomaltase deficiency
Bulgarian	Сакроидаза	Лечение на вродена недостатъчност на сукраза-изомалтаза
Croatian	Sakrozidaza	Liječenje prirođenog manjka sukraze-izomaltaze
Czech	Sacroidasa	Léčba vrozeného deficitu sucras- isomaltázy
Danish	Sacrosidase	Behandling af congenital sucras-isomaltas brist
Dutch	Sacrosidase	Behandeling van congenitale sucrase-isomaltase deficiëntie
Estonian	Sakrosidaas	Kaasasündinud sukraas-isomaltaasi puudulikkuse ravi
Finnish	Sakrosidaasi	Synnynnäisen sakkaroosi-isomaltaasin puutoksen hoito
French	Sacrosidase	Déficit congénital en sucrase-isomaltase (DCSI)
German	Sacrosidase	Behandlunng angeborener Sucrase-Isomaltase Defizienz
Greek	Σακροσιδάση	Θεραπεία της συγγενούς ανεπάρκειας σουκράσης-ισομαλτάσης.
Hungarian	Szakrozidáz	Kongenitális szukráz-izomaltáz elégtelenség kezelése
Italian	Sacrosidasi	Trattamento del deficit congenito di sucrasi-isomaltasi
Latvian	Sakrozidāze	Iedzimta sukrāzes-izomaltāzes deficīta ārstēšana
Lithuanian	Sakrozidazė	Įgimto sacharozės – izomaltazės stokos gydymas
Maltese	Sacrosidase	Kura tan-nuqqas konġenitu ta' sucrase-isomaltase
Polish	Sakrozydaza	Leczenie wrodzonego niedoboru sukrazo-izomaltazy
Portuguese	Sacrosidase	Tratamento da deficiência congénita de sucrase-isimaltase
Romanian	Sacrozidază	Tratamentul deficientei congenitale de sucrază-izomaltază
Slovak	Sakrozidáza	Liečba vrodeného deficitu sukrázy - izomaltázy
Slovenian	Sukrozidaza	Zdravljenje vrojenega pomanjkanja sukraza – izomaltaze
Spanish	Sacrosidase	Tratamiento de la deficiencia congenital de sucrose-isomaltase
Swedish	Sakrosidas	Behandling av congenital sucras-isomaltas brist
Norwegian	Sakrosidase	Behandling av medfødt sukrase-isomaltase mangel
Icelandic	Sakrósídasi	Meðferð á meðfæddum súkrasa-ísómaltasa skorti

[1] At the time of designation