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Public summary of opinion on orphan designation

Copper histidinate for the treatment of Menkes disease

On 21 August 2020, orphan designation EU/3/20/2320 was granted by the European Commission to CambPharma Solutions (CY) Limited, Cyprus, for copper histidinate (also known as CUTX-101) for the treatment of Menkes disease.

What is Menkes disease?

Menkes disease is a genetic disorder that affects how copper from food, which is essential for the good functioning of many organs, is distributed in the body. In people with Menkes disease, because of a genetic mutation (change), copper is not efficiently transported from the gut into the bloodstream and distributed throughout the body. As a result, copper is not able to adequately reach some organs such as the brain and liver, while it builds up in other organs such as the gut and the kidneys, damaging them. Symptoms usually appear within the first few months of life and may include sparse, kinky and brittle hair, slow growth, seizures (fits), low muscle tone, sagging facial features, and developmental and intellectual disability.

Menkes disease is a debilitating disease that can be life-threatening due to progressive damage to the brain and problems affecting blood vessels, the bones and several organs.

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

At the time of designation, Menkes Disease affected less than 1 in 10,000 people in the European Union (EU). This was equivalent to a total of fewer than 52,000 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 submission of the application for orphan designation, no treatments were authorised in the EU for the treatment of Menkes disease. Patients were sometimes given injections of copper supplements and their symptoms treated with painkillers, medicines for seizures and physical therapy.

*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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How is this medicine expected to work?

This medicine is made of copper histidinate, a form of copper that can be given by injection under the skin. Once given to the patient, the medicine can replenish copper histidinate normally found in the blood, restore the balance of copper in the body and thereby improve the symptoms of the disease.

What is the stage of development of this medicine?

The effects of copper histidinate have been evaluated in experimental models.

At the time of submission of the application for orphan designation, clinical trials with copper histidinate in patients with Menkes disease were ongoing.

At the time of submission, copper histidinate was not authorised anywhere in the EU for the treatment of Menkes disease. Orphan designation of copper histidinate had been granted in the United States for the condition.

In accordance with Regulation (EC) No 141/2000, the COMP adopted a positive opinion on 16 July 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

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	Copper histidinate	Treatment of Menkes disease
Bulgarian	Меден хистидинат	Лечение на болестта на Менкес
Croatian	Bakreni histidinat	Liječenje Menkesove bolesti
Czech	Histidinát mědi	Léčba Menkesovy choroby
Danish	Kobberhistidinat	Behandling af Menkes sygdom
Dutch	Koperhistidinaat	Behandeling van de ziekte van Menkes
Estonian	Vaskhistidinaat	Menkese haiguse ravi
Finnish	Kuparihistidinaatti	Menken taudin hoito
French	Histidinate de cuivre	Traitement de la maladie de Menkes
German	Kupferhistidinat	Behandlung der Menkes Krankheit
Greek	Ιστιδινικός χαλκός	Θεραπεία της νόσου Menkes
Hungarian	Réz hisztidinát	Menkes-betegség kezelése
Italian	Istidinato di rame	Trattamento della malattia di Menkes
Latvian	Vara histidināts	Menkes slimības ārstēšana
Lithuanian	Vario histidinatas	*Menkes* ligos gydymas
Maltese	Istidina tar-ram	Kura tal-marda Menkes
Polish	Histydynian miedzi	Leczenie choroby Menkesa
Portuguese	Histidinato de cobre	Tratamento da doença de Menkes.
Romanian	Histidinat de cupru	Tratamentul bolii Menkes
Slovak	Copper histidinate	Liečba Menkesovej choroby
Slovenian	Bakrov histidinat	zdravljenje Menkesove bolezni
Spanish	Histidinato de cobre	Tratamiento de la enfermedad de Menkes
Swedish	kopparhistidinat	Behandling av Menkes sjukdom
Norwegian	Kobberhistidinat	Behandling av menkes sykdom
Icelandic	Koparhistidínat	Meðferð á Menkes-sjúkdómi

[1] At the time of designation