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Public summary of opinion on orphan designation

Hydroxychloroquine sulphate for the treatment of LIPIN1 disease

On 17 January 2018, orphan designation (EU/3/17/1963) was granted by the European Commission to Professor Pascale De Lonlay, France, for hydroxychloroquine sulphate for the treatment of LIPIN1 disease.

What is LIPIN1 disease?

LIPIN1 disease is a condition that causes regular bouts of rhabdomyolysis (where muscles tissue is broken down), resulting in muscle pain and weakness. Patients may also have kidney and heart problems. The condition usually starts in childhood and is caused by a mutation (change) in a gene responsible for making a protein known as lipin-1, which is needed for muscles to function normally. Patients with the mutation cannot produce the protein properly.

LIPIN1 disease is debilitating and life threatening because of complications of rhabdomyolysis such as cardiac arrest (when the heart suddenly stops beating) or kidney failure.

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

At the time of designation, LIPIN1 disease affected approximately 0.0005 in 10,000 people in the European Union (EU). This was equivalent to a total of around 20 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?

No satisfactory methods were authorised in the EU for treatment of LIPIN1 disease at the time of application. Patients with rhabdomyolysis were given with fluids and had their heart and blood levels of minerals monitored regularly.

*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 515,700,000 (Eurostat 2017).

30 Churchill Place ● Canary Wharf ● London E14 5EU ● United Kingdom
Telephone +44 (0)20 3660 6000 **Facsimile** +44 (0)20 3660 5555
Send a question via our website www.ema.europa.eu/contact

How is this medicine expected to work?

This medicine targets a receptor known as Toll-like receptor 9 (TLR9), which is involved in stimulating the inflammatory reactions associated with rhabdomyolysis. By attaching to this receptor, the medicine is expected to reduce TLR9 action and so help relieve symptoms of rhabdomyolysis in patients with LIPIN1 disease.

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 LIPIN1 disease had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for LIPIN1 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 7 December 2017 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:

Contact details of the current sponsor for this orphan designation can be found on EMA website, on the medicine's rare disease designations page.

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	Hydroxychloroquine sulphate	Treatment of LIPIN1 disease
Bulgarian	Хидроксихлорохин сулфат	Лечение на LIPIN1 дефицит
Croatian	Hidroksiklorokin sulfat	Liječenje deficita u LIPIN1
Czech	Hydroxychlorochin sulfát	Léčba onemocnění LIPIN1
Danish	Hydroxychloroquinsulfat	Behandling af underskuddet i LIPIN1
Dutch	Hydroxychloroquinesulfaat	Behandeling van het tekort in LIPIN1
Estonian	Hüdroksüklorokviinsulfaat	LIPIN1 haiguse ravi
Finnish	Hydroksiklorokiinisulfaatti	LIPIN1 puutoksen hoito
French	Hydroxychloroquine Sulfate	Traitement du deficit en LIPIN1
German	Hydroxychloroquinsulfat	Behandlung von LIPIN1 Erkrankung
Greek	Υδροξυχλωροκίνη Θειική	Θεραπεία της ανεπάρκειας LIPIN1
Hungarian	Hidroxi-klorokin-szulfát	LIPIN1 betegség kezelése
Italian	Idrossiclorochina solfato	Trattamento del deficit di Lipina-1
Latvian	Hidroksihlorokvīna sulfāts	LIPIN1 slimības ārstēšana
Lithuanian	Hidroksichlorokvino sulfatas	LIPIN1 ligos gydymas
Maltese	Sulfat idroksiklorokina	Kura tad-defiċit f'LIPIN1
Polish	Siarczan hydroksychlorochiny	Leczenie deficytu LIPIN1
Portuguese	Sulfato de hidroxicloroquina	Tratamento do déficit em LIPIN1
Romanian	Sulfat de hidroxiclorochină	Tratamentul bolii LIPIN1
Slovak	Hydroxychlorochín sulfát	Liečba deficitu v LIPIN1
Slovenian	Hidroksiklorokvin sulfat	zdravljenje pomanjkanja LIPIN1
Spanish	Sulfato de hidroxicloroquina	Tratamiento del déficit en LIPIN1
Swedish	Hydroklorkokin-sulfat	Behandling av LIPIN1-underskott
Norwegian	Hydroksyklorokin sulfat	Behandling av underskuddet i LIPIN1 sykdom
Icelandic	Hýdroxýklórókín Súlfat	Meðhöndlun hallans á LIPIN1

[1] At the time of designation