26 March 2013
EMA/COMP/92274/2013
Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Recombinant human tripeptidyl-peptidase 1 for the treatment of neuronal ceroid lipofuscinosis type 2

On 12 March 2013, orphan designation (EU/3/13/1118) was granted by the European Commission to BioMarin Europe Ltd., UK, for recombinant human tripeptidyl-peptidase 1 for the treatment of neuronal ceroid lipofuscinosis type 2.

What is neuronal ceroid lipofuscinosis type 2?

Neuronal ceroid lipofuscinosis type 2 (also known as Jansky-Bielschowsky disease) is one of a group of inherited diseases belonging to the larger family of metabolic disorders called 'lysosomal storage diseases'. Patients with this condition are unable to produce enough of the enzyme tripeptidyl-peptidase 1, which plays a role in breaking down proteins inside the cells. This results in a build-up of deposits known as lipofuscins in the cells, including nerve cells, which damages tissues and leads to progressive degeneration of the brain and retina (the light-sensitive layer at the back of the eye). Symptoms usually begin between 2 and 4 years of age, and include delayed speech, inability to coordinate muscle movements, fits, loss of vision and mental deterioration.

Neuronal ceroid lipofuscinosis type 2 is a long-term debilitating and life-threatening disease that usually leads to death between the ages of 8 and 12.

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

At the time of designation, neuronal ceroid lipofuscinosis type 2 affected approximately 0.3 in 10,000 people in the European Union (EU). This was equivalent to a total of around 15,000 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 condition is estimated and assessed on the basis of data from the European Union (EU 27), Norway, Iceland and Liechtenstein. This represents a population of 509,000,000 (Eurostat 2013).

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What treatments are available?

At the time of orphan designation there were no satisfactory treatments authorised in the EU for neuronal ceroid lipofuscinosis type 2. The disease was managed by treating the symptoms of the disease.

How is this medicine expected to work?

Tripeptidyl-peptidase 1 is the enzyme that is lacking in patients with neuronal ceroid lipofuscinosis type 2. The medicine is expected to act as a replacement for the missing enzyme, carrying out its normal function of breaking down protein in the cells, which is ultimately expected to improve the overall outcome of patients.

The enzyme in this medicine is produced by a method known as 'recombinant DNA technology': it is made by cells that have received a gene (DNA), which makes them able to produce the enzyme.

What is the stage of development of this medicine?

The effects of recombinant human tripeptidyl-peptidase 1 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 neuronal ceroid lipofuscinosis type 2 had been started.

At the time of submission, the medicine was not authorised anywhere in the EU for neuronal ceroid lipofuscinosis type 2 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 6 February 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.

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	Recombinant human tripeptidyl-peptidase 1	Treatment of neuronal ceroid lipofuscinosis type 2
Bulgarian	Рекомбинантна човешка трипептидил пептидаза 1	Лечение на невронална цероидна липофусциноза тип 2
Czech	Rekombinantní lidská tripeptidylpeptidáza 1	Léčba neuronální ceroidní lipofuscinózy 2. typu
Danish	Rekombinant humant tripeptidylpeptidase 1	Behandling af neuronal ceroid lipofuscinose type 2
Dutch	Recombinant human triPeptidyl peptidase 1	Behandeling van neuronaal ceroïd lipofuscinose type 2
Estonian	Rekombinantne inimese tripeptidüül-peptidaas 1	2. tüüpi neuronaalse tseroidse lipofustsinoosi ravi
Finnish	Rekombinantti ihmisen tripeptidyylipeptidaasi 1	Tyypin 2 neuronaalisen seroidilipofuskinoosin hoito
French	Tripeptidyl-peptidase 1 recombinante humaine	Traitement de la céroïde-lipofuscinose neuronale de type 2
German	Rekombinante humane Tripeptidy-Peptidase 1	Behandlung der neuronalen Ceroid-Lipofuszinose Typ 2
Greek	Ανασυνδυασμένη ανθρώπινη τριπεπτίδυλο πεπτιδάση 1	Θεραπεία της Νευρωνικής Κηροειδούς Λιποφουσκίνωσης τύπου 2
Hungarian	Rhu tripeptidil-peptidáz 1	2-es típusú neuronális ceroid lipofuscinosis kezelése
Italian	Tripeptidil peptidasi 1 ricombinante umane	Trattamento della ceroido lipofuscinosi neuronale tipo 2
Latvian	Rekombinanta cilvēka tripeptidilpeptidāze-1	2. tipa neironu ceroīdās lipofuscinozes ārstēšana
Lithuanian	Rekombinantinė žmogaus tripeptidil-peptidazė 1	2 tipo neuronų ceroidinės lipofuscinozės gydymas
Maltese	Tripeptidyl-peptidase 1 uman rikombinanti	Kura taċ-ċerojde lipofuxinosi newronali tat-tip 2
Polish	Rekombinowana ludzka peptydaza tripeptydylowa 1	Leczenie ceroidolipofuscynozy neuronalnej typu 2

[1] At the time of designation

Language	Active ingredient	Indication
Portuguese	Tripeptidil-Peptidase 1 humana recombinante	Tratamento da lipofuscinose ceróide neuronal de tipo 2
Romanian	Tripeptidil peptidază 1 umană recombinantă	Tratamentul lipofuscinozei ceroide neuronale de tip 2
Slovak	Rekombinantná ľudská tripeptidyl-peptidáza 1	Liečba neuronálnej ceroidnej lipofuscinózy typu 2
Slovenian	Rekombinantna humana tripeptidil peptidaza 1	Zdravljenje nevronalne ceroidne lipofuscinoze tipa 2
Spanish	Tripeptidil peptidasa 1 humana recombinante	Tratamiento de la lipofuscinosis neuronal ceroidea de tipo 2
Swedish	Rekombinant mänskligt tripeptidyl-peptidas 1	Behandling av neuronal ceroidlipofuscinos typ 2
Norwegian	Rekombinant human tripeptidyl peptidase 1	Behandling av nevronal ceroid lipofuscinose type 2
Icelandic	Raðbrigða manna Þrípeptídýl-peptíðasi 1	Meðferð við taugaceróíð lípófúscinósis af gerð 2