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Committee for Orphan Medicinal Products

Public summary of opinion on orphan designation

Omigapil maleate for the treatment of congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy)

On 8 May 2008, orphan designation (EU/3/08/540) was granted by the European Commission to Santhera Pharmaceuticals (Deutschland) GmbH, Germany, for omigapil maleate for the treatment of congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy).

The name of the sponsor changed to Santhera Pharmaceuticals (Deutschland) GmbH in September 2010.

What is congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy)?

Congenital muscular dystrophies (CMD) are a group of hereditary disorders, frequently presenting at birth or within the first six months of life. There are many different forms of CMD and each form is caused by a specific defect in a gene. All forms of CMD share some symptoms and signs; such as weakness and degeneration of muscles, contractures and joint deformities. Usually CMD leads to difficulty in movement, skeletal deformation (scoliosis) and respiratory failure. Mental retardation is sometimes present.

Congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy) is caused by a defect in one of the collagen VI genes. Collagen is the main protein of connective (supporting) tissue in the body and provides support for the muscle cells. The exact mechanisms how these genetic defects lead to the disease are not fully characterized but the muscle cells of the patients are more sensitive to cell death and there might be a defect in the energy supplying parts of the cells called mitochondria. Bethlem myopathy represents a milder variant of Ullrich Syndrome. Congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy) is a chronically debilitating and life-threatening disease.

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

At the time of designation, congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy) affected approximately 0.03 in 10,000 people in the European

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Union (EU)[*]. This is equivalent to a total of around 1,500 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 exist that were authorised at the time of application.

How is this medicine expected to work?

The exact mechanism of action of omigapil maleate is not known. However, it is thought that the product interacts with a protein called glyceraldehyde 3-phosphate dehydrogenase, which is involved in cell death. By interacting with this protein, omigapil maleate may protect muscle cells from dying.

What is the stage of development of this medicine?

The effects of omigapil maleate were evaluated in experimental models.

At the time of submission of the application for orphan designation, no clinical trials in patients with congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy) were initiated.

Omigapil maleate was not authorised anywhere worldwide for congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy) or designated as orphan medicinal product elsewhere for this condition, at the time of submission.

In accordance with Regulation (EC) No 141/2000 of 16 December 1999, the COMP adopted a positive opinion on 4 March 2008 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.

[*]Disclaimer: For the purpose of the designation, the number of patients affected by the condition is estimated and assessed based on data from the European Union (EU 27), Norway, Iceland and Lichtenstein. This represents a population of 502,282,000 (Eurostat 2008). This estimate is based on available information and calculations presented by the sponsor at the time of the application.

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	Omigapil maleate	Treatment of congenital muscular dystrophy with collagen VI deficiency (Ullrich Syndrome and Bethlem Myopathy)
Bulgarian	Омигапил малеат	Лечение на вродена мускулна дистрофия с дефицит на колаген VI (синдром на Ullrich и миопатия на Bethlem)
Czech	Omigapil maleát	Léčba kongenitální muskulární dystrofie s deficiencí kolagenu VI (Ullrichův syndrom a Bethlemova moypatie)
Danish	Omigapil-maleat	Behandling af kongenit muskeldystrofi med kollagen VI-mangel (Ullrich syndrom og Bethlem myopati)
Dutch	Omigapil maleaat	Behandeling van congenitale spierdystrofie met collageen-VI-deficiëntie (Ullrich-syndroom en Bethlem-myopathie)
Estonian	Omigapiilmaleaat	Kollageen VI vaegusega kaasasündinud lihasdüstroofia (Ullrichi sündroom ja Bethlemi müopaatia) ravi
Finnish	Omigapiilimaleaatti	Kollageeni VI:n puutteesta johtuvan synnynnäisen lihasdystrofian (Ullrichin lihasdystrofian ja Bethlemin myopatian) hoito
French	Maléate d'omigapil	Traitement de la dystrophie musculaire congénitale avec déficit en collagène VI (syndrome d'Ullrich et myopathie de Bethlem)
German	Omigapilmaleat	Behandlung der kongenitalen Muskeldystrophie mit Kollagen VI-Defizienz (Ullrich-Syndrom und Bethlem-Myopathie)
Greek	Μηλεϊνική ομιγαπίλη	Θεραπεία της συγγενούς μυϊκής δυστροφίας με ανεπάρκεια κολλαγόνου IV (σύνδρομο Ullrich και μυοπάθεια Bethlem)
Hungarian	Omigapil maleát	VI. típusú kollagén hiányos congenitalis izomdystrophiák (Ullrich szindróma és Bethlem myopathia) kezelése
Italian	Omigapil maleato	Trattamento della distrofia muscolare congenita da deficit di collagene VI (sindrome di Ullrich e miopatia di Bethlem)
Latvian	Omigapila maleāts	Iedzimtas muskuļu distrofijas ar kolagēna VI trūkumu (Ullricha sindroms un Bethlema miopātija) ārstēšana
Lithuanian	Omigapilio maleatas	Įgimtos raumenų distrofijos, sąlygotos kolageno VI nepakankamumo, gydymas (Ullrich sindromo ir Bethlem miopatijos)
Maltese	Omigapil maleate	Kura tad-distrofija muskolari konġenitali b'nuqqas ta' kollagene VI (Sindrome ta' Ullrich u Mijopatija ta' Bethlem)
Polish	Maleinian omigapilu	Leczenie wrodzonej dystrofii mięśni z niedoborem kolagenu VI (zespół Ullricha i miopatia Bethlema)

[1] At the time of designation

Language	Active Ingredient	Indication
Portuguese	Maleato de Omigapil	Tratamento de distrofia muscular congénita com deficiência de colagénio VI (síndrome de Ullrich e miopatia de Bethlem)
Romanian	Omigapil maleat	Tratamentul distrofiei musculare congenitale cu deficit de colagen VI (sindromul Ullrich şi miopatia Bethlem)
Slovak	Omigapilmaleát	Liečba vrodenej svalovej dystrofie s nedostatkom kolagénu VI (Ullrichov syndróm a Bethlemova myopatia)
Slovenian	Omigapilov maleat	Zdravljenje prirojene mišične distrofije s pomanjkanjem kolagena VI (Ullrichov sindrom in Bethlem miopatija)
Spanish	Maleato de omigapilo	Tratamiento de la distrofia muscular congénita con deficiencia de colágeno VI (síndrome de Ullrich y miopatía de Bethlem)
Swedish	Omigapil maleat	Behandling av kongenital muskeldystrofi med kollagen VI-brist (Ullrichs syndrom och Bethlems myopati)
Norwegian	Omigapilmaleat	Behandling av kongenital muskulær dystrofi med kollagenVI-mangel (Ullrich syndrom og Bethlem myopati)
Icelandic	Ómigapíl maleat	Meðferð við meðfæddri vöðvarýrnun með kollagen-VI skorti (Ullrich heilkenni og Bethlem vöðvakvilli)