Ubiquinol, designated as an orphan drug for treatment of primary coenzyme Q_{10} deficiency

> ***Primary coenzyme Q_{10} deficiency is a serious rare disease which may affect the muscular, nervous or renal systems.***

> ***Ubiquinol, the reduced form of coenzyme Q_{10} , helps cells to improve their energy synthesis capacity and anti-oxidant defences. This means they can be expected to relieve part of the symptoms and clinical signs of the disease.***

> ***The application for this to be classified as an orphan drug was led by a research group from the Universidad Pablo de Olavide in cooperation with another team from the Hospital Sant Joan de Déu de Barcelona, both belonging to the CIBERER and the collaboration of a group form the University of Padova.***

Seville/Madrid, 2nd November 2016. The European Medicines Agency (EMA) has designated Ubiquinol as an orphan drug for the treatment of primary coenzyme Q_{10} deficiency, a serious rare disease which produces muscular, nervous system and kidney disorders and can cause failures of other organs.

Studies on the possible application of this orphan drug for the treatment of primary coenzyme Q_{10} deficiency have been directed by the research group led by Plácido Navas at the Universidad Pablo de Olavide in Seville belonging to the Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER). The research group at the Hospital Sant Joan de Déu de Barcelona led by Rafael Artuch, also from the CIBERER, and companies PharmaNord and Kaneka also took part in the clinical side. This orphan drug is sponsored by the CIBER. Dr. Leonardo Salviati from the University of Padova has also participated actively.

Primary coenzyme Q_{10} deficiency syndrome is a hereditary disease caused by mutations in the genes acting in the biosynthesis of coenzyme Q_{10}, an essential compound in producing energy and in the anti-oxidant defence system, which are biological processes necessary for the cells to work properly.

Ubiquinol is the reduced form of coenzyme Q_{10}. This drug is expected to complement coenzyme Q_{10}, which is naturally found in the organism, helping the cells to produce more energy y and thus relieve the symptoms of this disease. The product has been evaluated on experimental models, and at the present time there is already a clinical trial with patients.

CoQ_{10} is a natural product that is currently available on the international market as a food complement, but in different formulations, which often cannot be properly absorbed in the organism. "Our research over the last ten years has consisted in finding the most effective formulation for recovering from the symptoms in patients. We have worked on animal models of the disease and on preliminary clinical trials. This has been the result of cooperative endeavours between several laboratories and companies. We would like to thank the families of affected persons who have cooperated with us, without whom we would never have managed this ", asserts Plácido Navas.

On primary coenzyme Q_{10} deficiency

Primary coenzyme Q_{10} deficiency causes damage to organs that are particularly dependent on energy such as the brain, the muscles, the liver or the kidneys. This can consequently result in muscular weakness, kidney failure, ataxia, hearing loss and epilepsy, amongst other clinical signs. Bioenergetics inefficiency leads to the death of a large number of cells, making the organs collapse. In the long term this illness is weakening and sometimes ends in death, particularly through kidney failure, muscular limitations and encephalopathy. This illness has a prevalence of roughly 1 in 100,000 persons, which is equivalent to around 5000 citizens in the whole European Union.

Orphan drugs sponsored by the CIBER

Ubiquinol is the fifth EMA orphan drug that has been sponsored by the CIBER. These orphan drugs are recommended for the following rare diseases: Fanconi anaemia type A, pyruvate kinase deficiency, adrenoleukodystrophy, leukocyte

adhesion deficiency type I and primary coenzyme $Q_{10.}$ deficiency.

Benefits of designation as an orphan drug

Designation as an orphan drug by the EMA entails such advantages as receiving an authorisation for marketing for ten years in which similar products cannot be marketed, being able to make use of assistance protocols and free or low-cost scientific advice and exemption of payments for the designation. The organisations which develop orphan drugs also have access to specific EU subsidies and to the programmes of its member states.

About the CIBERER

The Centro de Investigación Biomédica en Red (CIBER) is a consortium dependent on the Instituto de Salud Carlos III (Ministry of the Economy and Competitiveness) and cofinanced with ERDF funds. The CIBER, in its thematic Area of Rare Diseases (CIBERER) is the benchmark centre in Spain in research into rare diseases. Its main aim is to coordinate and promote basic, clinical and epidemiological research, as well as to ensure that the research work done in laboratories reaches the patient, and gives scientific answers to the questions arising from the interaction between doctors and patients. The CIBERER's human resources are made up of a team of over 700 professionals and integrates 62 research groups. It also has 20 associated clinical groups.

Further information

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