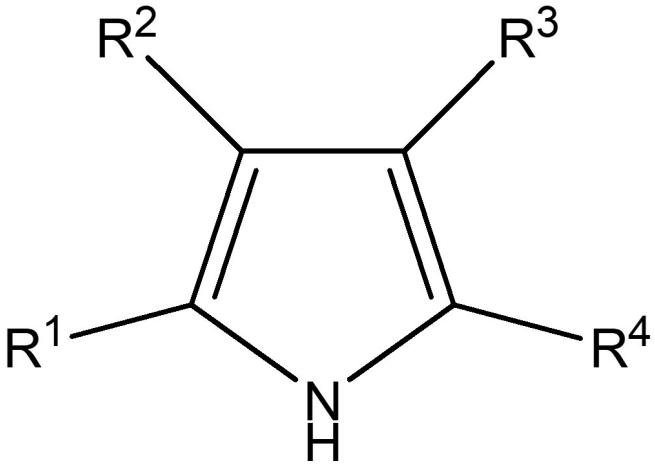
Use of inhibitors of porphobilinogen deaminase in the treatment of congenital erythropoietic porphyria

Background
Congenital erythropoietic porphyria (CEP) also known as Günther's disease is a hereditary disease and the least frequent of the porphyrias. This is a consequence of a malfunction in the uroporphyrinogen III synthase, leading to accumulation of Uroporphyrinogen I in bags below the eyes and in the limbs. Depending on its severity, other common symptoms of the disease are an extreme sensitivity to sunlight bone and cartilage destruction, erythrodontia, anemia, splenomegaly, hypertrichosis, etc.
Current Options. The treatment of CEP is only limited to attempting to prevent or relieve its symptoms, such as for example, by using sunscreens to prevent skin and eye scars, the continuous administration of heme derivatives or transfusing blood to counteract heme group deficiency. Splenectomy has also been performed to decrease severe haemolytic anaemia, however, the results of this surgical procedure have been variable and the benefit is often short-lived. Allogeneic haematopoietic stem cell transplant has also been used for CEP where few patient were treated and all experienced post-transplant acute complications.

Unmet Medical Need
CEP classifies as a rare disease with a prevalence of 2-5 case every 100,000 inhabitants. Europe and USA are the major predominant markets. Better therapeutic options are needed with the following characteristics: 1) Better Safety Profile, 2) Faster Onset and 3) Effective for Prophylaxis.

Technology
Out technology use of compounds having inhibitory capacity to inhibit the catalytic activity of porphobilinogen deaminase, the previous enzyme in the haeme biosynthetic pathway. The compound has been modified to allow improving of the pharmacological properties of the compound without significantly altering the inhibitory capacity of it.



Application
Class of compounds for the treatment and/or prevention of CEP.

Advantages
Some of the compounds protected under current patent are natural metabolites or are very chemically similar with natural metabolites. Most relevant characteristic of our compounds is low toxicity and especially important in CEP since the low number of patients, limits or makes impossible, the development of the clinical phases of the medical products/drugs.

Patent Status.
Priority Date: 5 Oct 2010; **Title**: Use of inhibitors of porphobilinogen deaminase in the treatment of congenital erythropoietic porphyria. WO2011EP63559; PCT/EP2011/063559. **Inventor**: Oscar Millet AGUILAR-GALINDO, Arantza SANZ-PARRA, Pedro Urquiza, Ana LAÍN, Juan Manuel FALCÓN-PEREZ.
Coverage: US, IT, GE, GB, FR, ES, CA (pending).

State of the Technology.
Outlicensed. ODD designation

Need.
Investors Series A Funding. Clinical trials

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