MedicalXpress

Treatment reduces symptoms in syndrome that causes extreme light sensitivity

July 2 2015

A novel synthetic hormone that makes certain skin cells produce more melanin significantly increases pain-free sun exposure in people with erythropoietic protoporphyria, a rare, genetic disorder resulting in excruciating pain within minutes of sun exposure. Two Phase III trials, conducted in Europe and in the United States by researchers at the Icahn School of Medicine at Mount Sinai and six other U.S. sites, showed that the duration of pain-free time in the sun and quality of life were significantly improved by treatment with afamelanotide, a novel synthetic version of a melanocyte-stimulating hormone. The findings were published in the July 2 issue of the *New England Journal of Medicine*.

"For patients worldwide, hiding from the sun for fear of debilitating pain is a fact of life, and for the first time we have an effective treatment for those who suffer from this specific porphyria," said lead study author Manisha Balwani, MD, Associate Professor of Genetics and Genomics Sciences and Medicine at the Icahn School of Medicine.

Symptoms of erythropoietic protoporphyria usually often first appear in early childhood. Patients experience severe, burning pain—typically on the hands and face—with sun exposure, followed by swelling and redness. The pain can be excruciating and is often is not alleviated by pain medications. Once patients learn to recognize early symptoms including tingling, burning, and itching, they avoid further sun exposure, with major implications for their daily activities, careers and quality of life.

"The data from these two Phase III trials conducted in Europe and in the U.S. support the effectiveness of this novel photo-protective drug, offering the patients in Europe—where the drug has been approved by the European Medicines Agency—the opportunity to perform their daily functions without the pain induced by sun exposure," said Robert J. Desnick, PhD, MD, Principal Investigator of the U.S. Trials and Dean for Genetic and Genomic Medicine, Icahn School of Medicine.

Researchers at Mount Sinai and in the European Union conducted two multicenter, randomized, double-blind, placebo-controlled trials of subcutaneous implants containing afamelanotide. Researchers randomly assigned 74 patients in the European Union and 94 in the United States to receive either afamelanotide or placebo implant every 60 days. The type and duration of sun exposure, number and severity of phototoxic reactions, and adverse events were recorded over the study periods. Quality of life was assessed with the use of validated questionnaires. The primary efficacy end point was the number of hours of direct exposure to sunlight without pain.

In the both the U.S. and European studies, the duration of pain-free time in sunlight was significantly longer in the afamelanotide groups. In the European study, the number of phototoxic reactions was significantly lower in the afamelanotide group. In both trials, quality of life improved with afamelanotide therapy. Adverse events were mostly mild and unrelated to the study drug.

Provided by The Mount Sinai Hospital