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Research Lends Hope for Patients With Rare Swelling Disease

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(PhysOrg.com) -- The results of two recent clinical trials offer promising treatments for sufferers of the rare genetic disease hereditary angiodema, says Jonathan Bernstein, MD, an internationally recognized allergy specialist and researcher at the University of Cincinnati.

"This is an orphan disease that until now hasn't been provided good treatments," says Bernstein, who co-authored two of three research articles on medicinal treatments for the disease that were published in the Aug. 5, 2010 issue of the *New England Journal of Medicine*.

Hereditary angioedema (HAE) is the spontaneous swelling of various body parts, including the hands, feet, face, stomach and airway, which is the most dangerous because it can lead to asphyxiation. According to the U.S. Hereditary Angioedema Association (HAEA), the disease occurs in about 1 in 10,000 to 1 in 50,000 people.

HAE is usually caused by a deficiency in the C1 esterase inhibitor enzyme.

"They either don't make enough of the C1 esterase inhibitor or it doesn't function properly," Bernstein explains, adding that research on the two treatments in which he participated provides evidence of prevention or symptom relief.

One of the trials Bernstein co-authored was sponsored by the pharmaceutical company Viropharma. This trial studied the intravenous

injection of the C1 esterase inhibitor enzyme, harvested from the plasma of healthy donors. This replacement therapy showed beneficial effect in preventing swelling attacks, says Bernstein.

The second trial, which he says proved effective as well but is approved currently only in Europe, used a chemical receptor blocker to prevent the swelling, much like an antihistamine works to block histamine. This trial was sponsored by the pharmaceutical company Shire.

Historically, he says, the disease has been treated with anabolic steroids, the same steroidal treatment used by body builders to enhance muscle mass and performance, as these steroids have been shown to increase production of C1 esterase inhibitor in the liver. This treatment, he says, is effective for many patients but comes with potentially severe side effects such as liver toxicity, weight gain, lipid abnormalities, masculinization in women and increased risk for liver cancer.

"These findings provide a whole new avenue of treatment for a small patient population who suffer greatly," says Bernstein.

Provided by University of Cincinnati

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