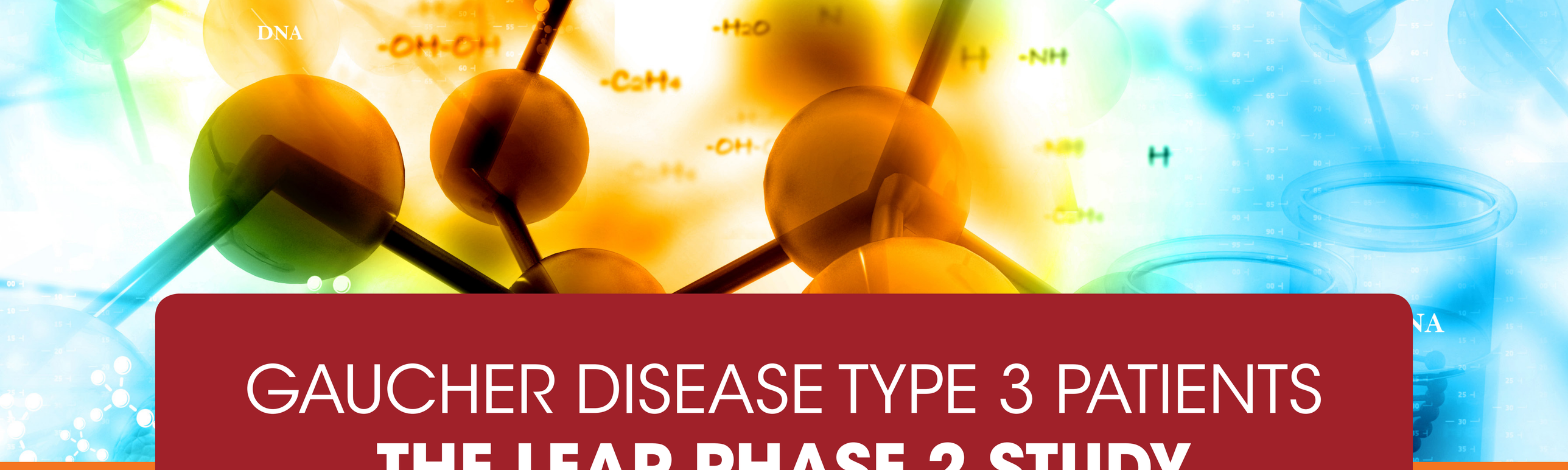

GAUCHER DISEASE TYPE 3 PATIENTS
THE LEAP PHASE 2 STUDY

Gaucher disease (GD) is a progressive, genetic condition that causes a fatty substance called glucosylceramide (commonly known as GL-1) to build up in cells of certain organs and bones. A specific enzyme called acid ß-glucosidase (also known as **glucocerebrosidase**) helps the body break down that fatty substance. People with Gaucher disease do not have enough of this enzyme. As a result, GL-1 fills up certain cells. These swollen cells are called **Gaucher cells**. Gaucher cells mainly build up in the **spleen**, the **liver**, and **bone marrow**. This build-up can cause symptoms such as: enlarged spleen and liver, anaemia (caused by low haemoglobin levels), easy bruising or bleeding (caused by low platelet levels), bone disease.

Clinically, patients with the classically described neurological manifestations are distinguished as having GD type 2, the acute, infantile rapidly progressive form, or type 3, the chronic neurological form of the disease. While effects of enzyme replacement therapy (ERT) on nonneurological manifestations of GD have been established, there are no approved treatments for the neurological aspects of the disease.

WHAT IS THE LEAP STUDY?

The LEAP study is a 52-week, two-part, open-label, multicenter, multinational study of the safety, tolerability, the effect the drug has to the body, efficacy of venglustat in combination with Cerezyme® in adult Gaucher disease type 3 (GD3) patients.

WHY IS THE LEAP STUDY BEING CONDUCTED?

Information collected during the LEAP trial will help investigating doctors understand if venglustat is effective for the treatment of GD3.

WHAT IS THE STUDY DRUG?

Venglustat is being investigated as a potential oral substrate reduction therapy (SRT) for GD3. Preclinical studies indicate the novel oral treatment is a glucosylceramide synthase inhibitor that reduces the synthesis of GL-1. The pill has shown that in mouse models it was effective in reducing neuropathologic and behaviour manifestations of neuronopathic GD[1].

WHAT IS THE STUDY DESIGN?

This study consists of two parts and may last up to 61 weeks. Part 1 consists of a measurable indicator of the severity of the disease evaluation phase in adult GD3 and adult GD1 patients, as well as a screening phase for adult GD3 patients prior to entering Part 2. Part 2 consists of a venglustat 52-week treatment phase in adult GD3 patients to investigate the safety, tolerability and efficacy of venglustat in combination with Cerezyme® infusions in adult patients with GD3.

ELIGIBILITY CRITERIA:

- 18 years of age or older (and ≤40 years for GD1 cohort)
- Confirmed diagnosis of GD1 or GD3 as applicable to part 1 and 2 of the study
- Patient has received treatment with enzyme replacement therapy for at least 3 years and received Cerezyme® at a stable monthly dose for the past 6 months
- Patient has reached Gaucher disease therapeutic goals

More information is available at:
www.clinicaltrials.gov
www.clinicaltrialsregister.eu

Site recruiting patients:

UK

Royal Free Hospital,
London Salford Royal,
Manchester
Addenbrooke's Hospital, Cambridge

For more information please contact the Gauchers Association

www.gaucher.org.uk