Galactosemia FAQs

What is AT-007?
AT-007 is an oral investigational drug in development by Applied Therapeutics to potentially prevent complications resulting from Galactosemia. An investigational drug is a drug that is being studied in a particular disease – it is not an approved drug.

What does AT-007 do?
AT-007 blocks an enzyme called aldose reductase, which may play a role in complications of Galactosemia. So far, AT-007 has been studied in an animal model of Classic Galactosemia (rats without GALT activity). In this model, AT-007 shows a beneficial effect on characteristic abnormalities associated with Galactosemia. No study in humans has been conducted to date.

What is known so far about AT-007?
So far, we know that AT-007 prevents complications of Galactosemia in a rat model of disease. These rats were treated from newborn stage up to 5 months of age, which spans the newborn, childhood and adult phases of rat development. The drug safety has also been tested in two animal species (rats and dogs) over one month of treatment, as well as several other safety studies that are required by the FDA before a drug is allowed to move into human clinical trials. Because the drug has not been tested in humans, we don't have any information on safety or effectiveness in humans.

Is treatment with AT-007 efficacious?
AT-007 has been studied only in animals. In an animal model of Galactosemia treatment with AT-007 produced multiple beneficial effects, including prevention of cataracts and prevention of central nervous system dysfunction (cognitive, memory, motor problems). However, no human study has been conducted to date.

Is treatment with AT-007 safe?
AT-007 has been studied only in animals. The doses that were studied in animal models supports studying the safety and tolerability in humans. No human study has been conducted to date.

Galactosemia FAQs

What is Orphan Designation?
Orphan Designation is a status granted by the FDA for a drug in development to treat a rare disease. Under Orphan Designation, a rare disease is defined as a disease affecting less than 200,000 people in the US. Applied Therapeutics was granted Orphan Designation in May 2019 for AT-007 in Galactosemia. While it is not a surprise to those of us in the Galactosemia community that Galactosemia qualifies as a rare disease, the formal designation signals that the process for drug development has started, and that Galactosemia is recognized as a rare disease by the FDA.

When will the human clinical trial start?
We understand the sense of urgency that many patients and family members feel to develop treatments for Galactosemia, and are moving as quickly as we can within the safe and responsible guidelines of clinical development. The first clinical trial will test safety and drug levels in adult healthy volunteers (without Galactosemia). We will then test safety, drug levels, biomarkers such as galactose and galactitol levels, as well as other outcomes in adults with Galactosemia (patients over the age of 18). After completing the adult Galactosemia study and understanding the safety profile in adults, we hope to expand into clinical trials in children with Galactosemia, if the safety profile is supportive of this.

Currently, we are recruiting adults (age 18 and older) with Classic Galactosemia and a small number of GALKD patients. If you would like to sign up to receive information on clinical studies, please visit:
https://www.appliedtherapeutics.com/galactosemia/
or email galactosemia@appliedtherapeutics.com

The Galactosemia Foundation kindly requests that patients and families do not contact them regarding the study, as they are not staffed to respond to multiple inquiries. Please reach out to Applied Therapeutics directly regarding participation in the clinical trial or any future studies.