Frequently Asked Questions

RP-L301- Gene Therapy Clinical Trial for Pyruvate Kinase Deficiency

What is Pyruvate Kinase Deficiency?

PKD is a rare, inherited blood disorder. PKD is caused by a defect in the *PKLR* gene, leading to red blood cell (RBC) break down, which causes anemia, jaundice, and chronic fatigue.

Who is eligible to participate in the gene therapy clinical trial?

Individuals who:

- Have a PKD diagnosis with a confirmed *PKLR* mutation;
- Are 8 through 55 years of age;
- Have severe, transfusion-dependent anemia after removal of the spleen;
- Are not receiving other experimental therapies; and
- Have no evidence of severe organ damage

What does participation in the PKD gene therapy clinical trial involve?

About 3 days

Screening period

Before participants join the clinical trial, they will have to undergo several medical tests at the study center to determine whether they are eligible to join the trial.

About 1 week

Stem cell collection

A participant will receive medications over several days to cause stem cells to enter the blood from the bone marrow. These stem cells have the potential to make different blood cells for the remainder of a participant's life. The stem cells will be collected from the body through a catheter temporarily placed in a vein. This procedure is called apheresis. Participants will be discharged after stem cell collection. After apheresis, a participant's stem cells will be genetically modified in a laboratory to introduce the correct copy of the PKLR gene, which takes about 12 weeks.

About 6 weeks

Infusion of investigational gene therapy

Before receiving the new cells, the participants will receive a chemotherapy medication called busulfan to remove the existing bone marrow cells to make room for the genetically modified cells. The participant will then receive an intravenous infusion of the genetically modified cells through the catheter, similar to having a transfusion. Participants will remain hospitalized and be evaluated daily until the infused cells start to make new blood cells, known as engraftment. Participants may require hospitalization for up to 6 weeks following gene therapy infusion.

2 Years

On-site follow-up period

Participants will need to return for follow-up visits, including blood and bone marrow tests, over the next 2 years. In addition, participants may expect to have long-term follow-ups, which will be discussed with the study center.

What is the impact of the investigational gene therapy on future fertility?

Study participants, both adult and parent(s)/guardian(s) of pediatric participants, will be informed by the study doctor of the risks of infertility/impaired fertility during the consent process. Rocket Pharma covers fertility preservation procedures for study participants who are at risk of their fertility being impaired due to participation in the investigational gene therapy clinical trial.

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