



Living Liver Donation: Medical Toolkit

CHAPTER • 11

Liver donation if you have a family history of progressive familial intrahepatic cholestasis (PFIC)

What is progressive familial intrahepatic cholestasis (PFIC)?

Progressive familial intrahepatic cholestasis (PFIC) is a disease that causes liver damage, which gets worse over time. Here's what happens:

1. In PFIC, your liver cells are less able to get rid of bile. Bile is a fluid your liver makes that helps break down fats in food
2. The bile builds up in your liver cells and your blood
3. This causes liver damage, which can lead to liver failure (your liver stops working), and can cause jaundice, which is a buildup of a substance in your blood that causes yellow skin and yellow in the whites of the eyes

Most people with PFIC need a liver transplant during childhood. Often, the recipient's parent is the donor.

What causes PFIC?

PFIC is a genetic condition passed down from your parents. It is caused by a change (mutation) in a gene that tells liver cells how to get rid of bile.

Everyone gets 2 copies of the gene – one from each parent. To have PFIC, you need to have 2 changed genes – one from your mother and one from your father.

If you have one changed gene and one normal gene, you are a “carrier” for PFIC, but do not have PFIC. Carriers can have liver cells that are less able to get rid of bile in certain situations, such as during pregnancy or when taking certain medicines or can have no medical issues at all.

How will I know if I have PFIC?

As part of the living liver donor evaluation, your doctor will ask about your family history of liver disease. If you have a family history, you will give a blood sample for genetic tests to see if you have PFIC or are a carrier.

Can I donate if I have a family history of PFIC?

If you have PFIC, you will not be able to donate.

If you are a carrier of PFIC, you will still be able to donate. Research has shown that living liver donation from a carrier of PFIC is safe for both the donor and the recipient. And donating does not raise the chance of liver problems for you.

For example, it's safe for a parent who is a carrier of PFIC to donate to their child with PFIC.

REFERENCES

1. Harmanci O et al. Late-Onset Drug-Induced Cholestasis in a Living-Related Liver Transplant Donor with Progressive Familial Intrahepatic Cholestasis. *Experimental and Clinical Transplantation*. 2015 Nov;13 Suppl 3:107-109.
2. Cutillo L et al. Safety of living-related liver transplantation for progressive family intrahepatic cholestasis. *Pediatric Transplantation*. 2006 Aug;10(5):570-4.
3. Trotter JF and Kam I. Living Donor Transplantation: Evaluation and Selection in Adults. *Transplantation of the Liver*. 3rd Edition. Ronald W Busuttil and Goran BG Klintmalm. Elsevier: Philadelphia, 2015.

Note: This information is the opinion of the Living Donor Community of Practice (LDCOP) of the American Society of Transplantation. The LDCOP is a group of health care professionals and researchers who specialize in living donation. The LDCOP's recommendations are meant to offer you helpful information, but you may find opinions from other groups or organizations that are helpful to you, too.