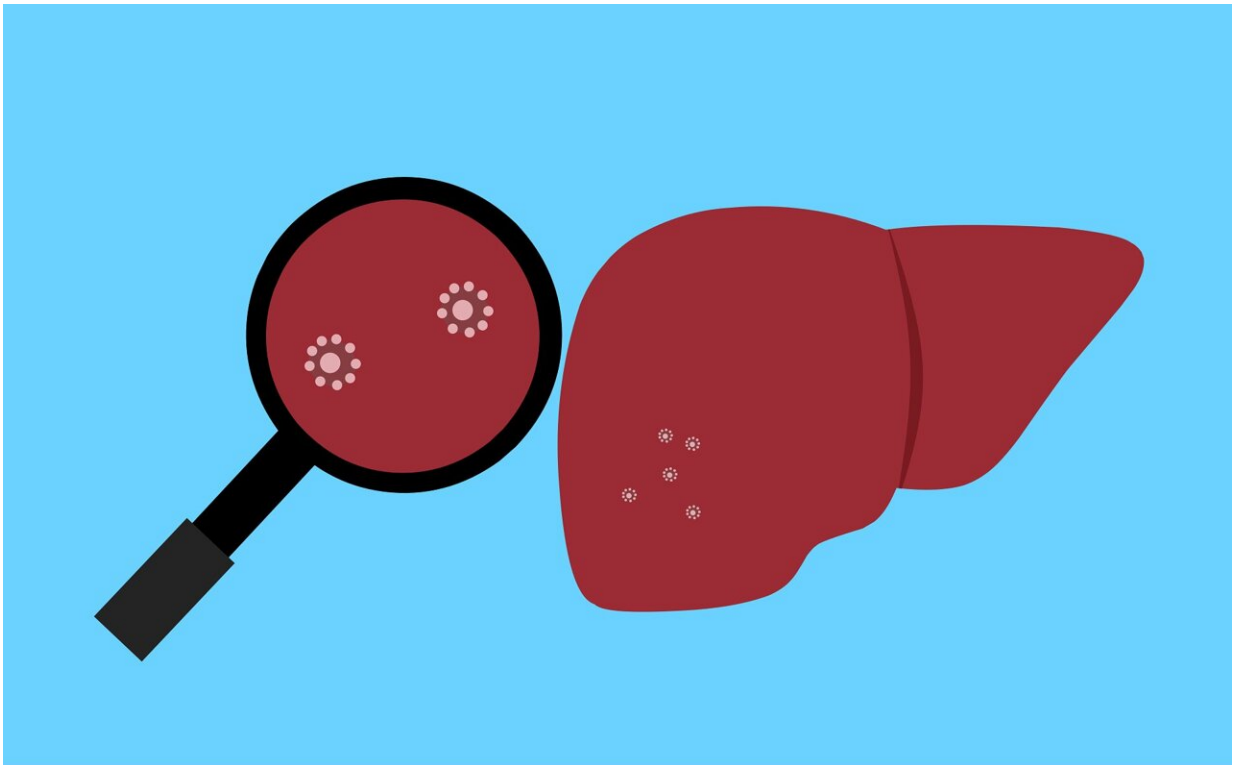


# Q&A: New thesis on the acute porphyrias, a group of rare inherited metabolic diseases

October 16 2023

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Mattias Lissing from the Gastroenterology and Rheumatology Unit, at the Department of Medicine, Huddinge (MedH) is defending his thesis: "Liver transplantation, cancer risk and comorbidity in acute porphyria"

on 20 October 2023. The main supervisor is Staffan Wahlin (MedH).

## **What is the main focus of your thesis?**

Our research focuses on a group of rare inherited metabolic diseases—the acute porphyrias. We have explored the outcomes in patients who have had a [liver transplantation](#) as treatment for recurrent porphyria attacks. Furthermore, we have studied the risk of [liver cancer](#), [cardiovascular disease](#), other (non-hepatic) cancers, kidney disease and other late complications in acute porphyria.

## **Which are the most important results?**

We found that the risk of liver cancer is 38 times higher among patients with acute porphyria compared to the general population. The risk is primarily associated to the biochemical activity in acute porphyria and not as much to symptoms, sex or type of genetic variant. We also found that despite the high prevalence of high blood pressure and kidney disease in patients with acute porphyria, the risk of other cardiovascular diseases was not increased. Liver transplantation cures acute attacks and provides a gradual improvement in neurological symptoms.

## **How can this new knowledge contribute to the improvement of people's health?**

In recent decades, the focus in management of patients with acute porphyria has shifted from first reducing mortality (which was high), to prevention, symptom reduction and managing acute complications during acute attacks, to now increasingly dealing with long-term complications such as liver cancer, [high blood pressure](#) and [kidney disease](#). With the results from our studies, clinicians can improve the follow-up of patients at risk, introduce monitoring and implement

appropriate measures in time.

## What are your future ambitions?

Knowledge gaps are common in rare diseases, also in the porphyrias. I want to continue building and further develop the collaboration between the unit of hepatology and our colleagues at Porphyria center Sweden at the Center for Inherited Metabolic Diseases (CMMS) and involve experts from other research areas in our future projects.

**More information:** Liver transplantation, cancer risk and comorbidity in acute porphyria. [openarchive.ki.se/xmlui/bitstr ...  
quence=2&isAllowed=y](https://openarchive.ki.se/xmlui/bitstream/handle/11524/153811/1/thesis-acute-porphyrias-group.html)

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Citation: Q&A: New thesis on the acute porphyrias, a group of rare inherited metabolic diseases (2023, October 16) retrieved 11 May 2024 from <https://medicalxpress.com/news/2023-10-qa-thesis-acute-porphyrias-group.html>

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