

# Discovery of gene involved in fatty liver disease

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An international research consortium led by CIC bioGUNE has discovered the involvement of a gene in the development of non-alcoholic fatty liver disease (NAFLD). Although the gene concerned (SLC2A1) had never previously been linked to liver disease, it appears to be involved in the development of this disorder. NAFLD is currently one of the leading causes of chronic liver disease in the world.

It was already known that, as well as environmental aspects (lifestyle, diet, etc.), the likelihood of developing NAFLD was also affected by [genetic factors](#), although the [genes](#) concerned are poorly defined. The aim of this study was to identify genetic variants associated with this disease in order to gain a better understanding of the genetics behind its development. The final goal of the study was therefore to try identifying new targets for the diagnosis and treatment of this disease.

This work, which was published recently in the leading scientific journal *Hepatology*, was undertaken by 20 institutions from Spain, France, Germany, Denmark and the USA, with Basque institutions playing a key role. Thus, as well as CIC bioGUNE, the company OWL Genomics, the Physiology Department at the UPV/EHU School of Medicine and the Galdakao Hospital all took part in this research.

## The research

The initial phase of the study involved searching for variations in the

DNA sequence of 92 genes, which had shown to be likely candidates by previous studies. DNA from 69 patients diagnosed with NAFLD and 217 health individuals was used. Various genes that exhibited significant differences in their sequence were identified and studied again in a different cohort of 451 NAFLD patients and 304 healthy individuals.

The correlation of NAFLD and high frequency of changes in the sequence for SLC2A1 was found at this stage. Once this relationship had been found, it was studied whether the presence of this gene at an RNA level was the same in the livers of healthy individuals and NAFLD patients. This study showed that [RNA](#) from the SLC2A1 gene was present at much lower levels in NAFLD patients.

Finally, an in vitro test with hepatocytes (one of the main cell types in the liver) showed that inhibition of SLC2A1 gene expression resulted in much greater accumulation of lipids than in cells with normal levels of this gene, as well as greater oxidative damage. These are the most representative characteristics of liver status in NAFLD patients.

According to the CIC bioGUNE researcher Ana M. Aransay, who headed the project, "involvement of the SLC2A1 gene in NAFLD still needs to be confirmed in other patient populations from other countries", adding that "as NAFLD is a complex disease, our findings must be interpreted in light of the results of other recent studies".

"Thus", continued Aransay, "an understanding of the development of NAFLD must take into account all the causes of this disease, especially genetic and environmental factors, as well as the interaction between them. An interpretation of the interactions that arise will help us to describe the reality of many complex diseases".

## **NAFLD**

NAFLD is a progressive disease that ranges from fatty liver build-up (steatosis) to non-alcoholic steatohepatitis (NASH), which is inflammation around the fats. Non-alcoholic [fatty liver disease](#) is the most common [liver disease](#) in Europe and the USA, and its prevalence is currently increasing in many other regions of the world, especially Asia and India.

The prevalence of steatosis and NASH in adults in the Western countries is around 30% and 3%, respectively. NASH is the most serious manifestation of NAFLD as around 20% of NASH patients go on to develop cirrhosis within 10 years, with over a quarter of these patients subsequently developing hepatocellular carcinoma (HCC), or liver cancer, after around 10 years.

**More information:** Vazquez-Chantada, M. et al. Solute Carrier Family 2 Member1 Is Involved in the Development of Nonalcoholic Fatty Liver Disease, *Hepatology* Volume 57, Issue 2, pages 505–514, February 2013.

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