

استغفر الله العظيم



# **Accurate DNA allele assay (rs738409) in patients with non-alcoholic fatty liver disease (NAFLD)**

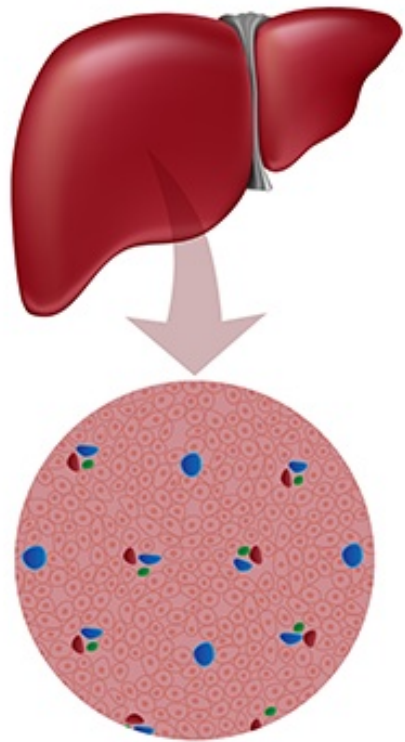
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# Introduction:

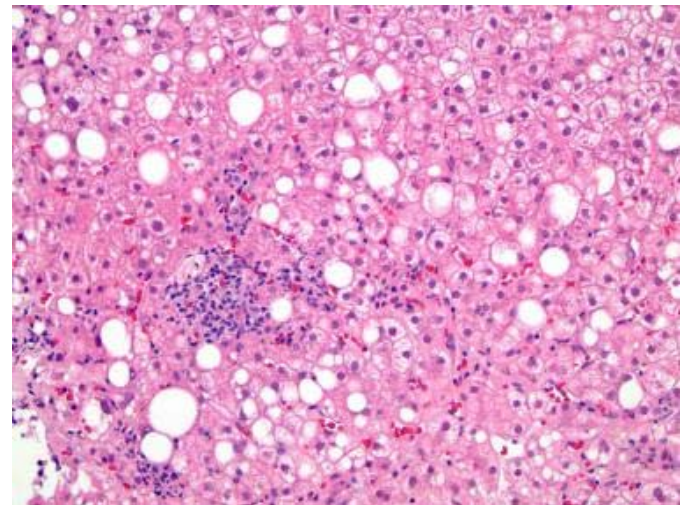
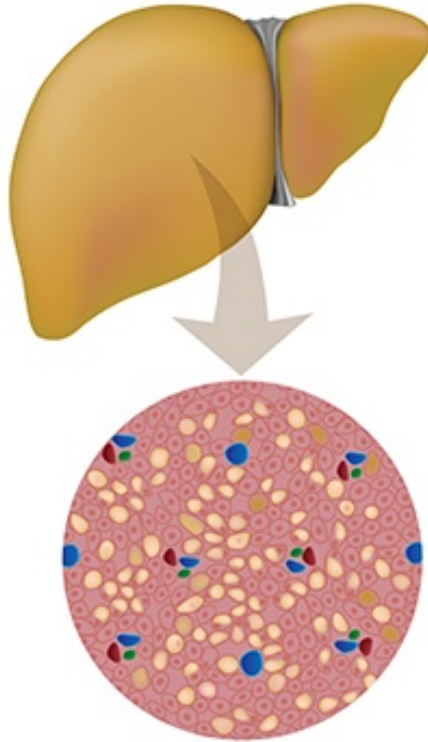


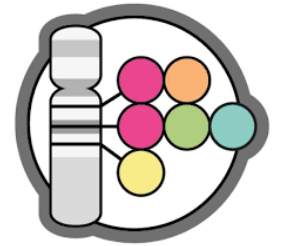
- Non-alcoholic fatty liver disease is considered nowadays a **major public health** problem worldwide.
- Most often is a mix of different causes. is exposed in the presence of other risk factors, such as **severe obesity**, **visceral adiposity**, increased intake of **sugars** or **omega-6 poly-unsaturated fatty acids**, and other genetic **factors**.
- Obesity, **type II diabetes** and **dyslipidemia**, in the absence of alcohol consumption, are the most important risk factors for non-alcoholic fatty liver disease.
- The natural course of NAFLD is variable and is possible influenced by genetic factors. Polymorphisms in **specific genes** and **histocompatibility complex genes** have been studied in patients with NAFLD.

Healthy liver



Fatty liver

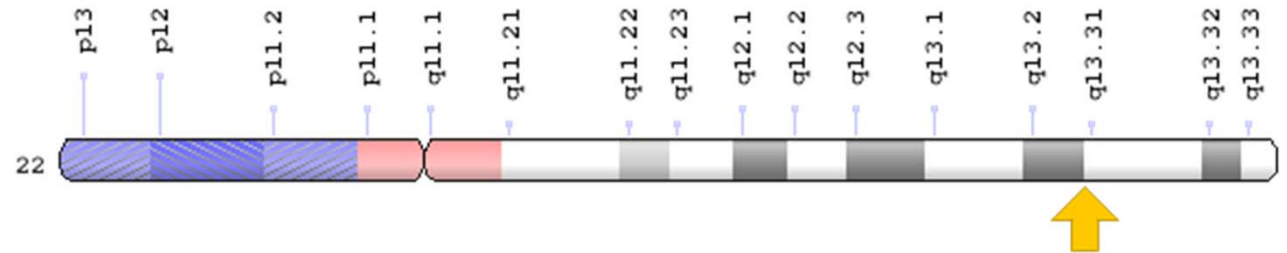




# Genome-wide association studies

- The interaction of genetic and environmental factors play an important role in the determination of the phenotype of the disease and its progression.
- Consistent with these data, a common variant in **patatin-like phospholipase domain containing protein 3** (PNPLA3) (adiponutrin) gene has been associated with hepatic triglyceride content and with the development of hepatic steatosis.
- Gene rs738409 polymorphism is associated with an increased risk for fatty liver, with the minor allele [G] as steatosis risk allele It is located on **22 chromosome** and encodes a protein linked to the main hydrolase of adipose tissue-triglyceridlipase.
- As a result of the substitution of isoleucine with methionine at 148 position, patients carrying the risk variant will have increased hepatic fat content .

# rs738409



Geno	Mag	Summary
<u>(C;C)</u>	3.1	higher odds of alcoholic liver disease, increased liver fat
<u>(C;G)</u>	2	increased liver fat, odds of alcoholic liver disease
<u>(G;G)</u>	2.1	most common genotype; slightly less damage from alcohol

# Material and Methods:

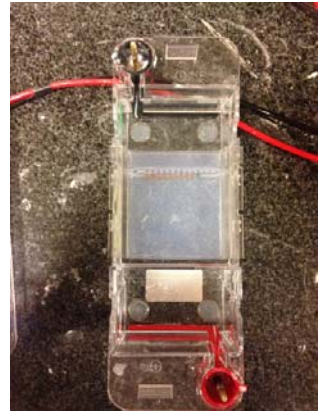


- This study aimed to investigate the relationship single polymorphism PNPLA3I148M in patients with NAFLD and compared the results with those reported in other parts of the world sets.
- DNA was extracted from the peripheral blood of 85 patients with non-alcoholic fatty liver disease and 120 normal persons.
- 356 bp gene fragment was amplified by the polymerase chain reaction (PCR). subsequently, the mutation was screened for random fragment length polymorphism analysis (RFLP).

# Allele assay

- DNA Extraction
- Purification of genomic DNA
- PCR-RFLP
- The restriction endonuclease **BtsCI**
- electrophoresis in a 2% agarose gel

5'... GGATGNN<sup>▼</sup>... 3'  
3'... CCTACNN<sup>▲</sup>... 5'





# Results & Discussion :



- This study assessed the influence of the rs738409 polymorphism of the PNPLA3 gene on NAFLD Patients.
- Statistical analysis showed that the risk allele (G-allele) frequency of rs738409 was **40.07 %** in the control subjects and **80.12 %** in patients with NAFLD.
- patients with the **GG** genotype presented a higher risk of steatosis than patients with the **CC** genotype
- this shows a strong association PNPLA3I148M polymorphism with the non-alcoholic fatty liver disease in our population (P-value <0.001 OR = 3.651, CI: 2.123 - 4.471).

A green rectangular sign with rounded corners and a white border, mounted on a wooden post. The sign features the word "Challenges" in a large, white, sans-serif font. The background is a bright blue sky with scattered white clouds. The sign is tilted slightly to the right.

Challenges

# محبان محمد (ع) او خواهد آمد

قلبهای ما جایگاه مشیت الهی است، پس هر گاه او  
بخواهد ما هم می خواهیم. (بهار انوار، ج ۵۲، ص ۵۱)

