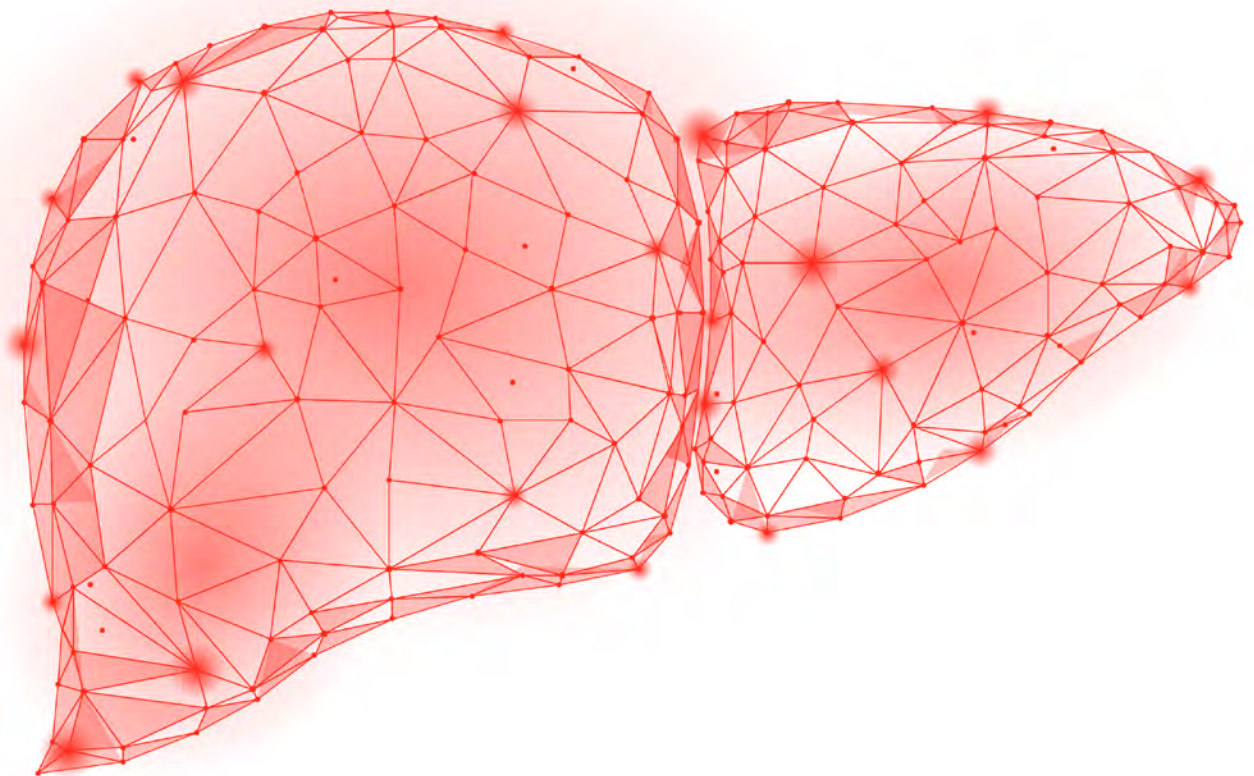


PNPLA3 Genotyping Kit

Enabling insights into MASH/MASLD biology



Highlights

- Analytically Robust
- Highly Accurate
- Simple Workflow
- Reduced Hands-On Lab Time



Steatotic Liver Disease and PNPLA3

Steatotic Liver Disease (SLD), including MASH and MASLD, is one of the most common forms of chronic liver disease, with an estimated prevalence of about 30% across many countries.¹

The I148M genotype variant of the PNPLA3 gene is associated with MASH / MASLD disease severity and fibrosis progression even in the absence of classical metabolic dysfunction (including obesity) and as such is of significant research interest to the field.

Intended Use

The Almac PNPLA3 Genotyping Kit is a qualitative polymerase chain reaction (qPCR) based kit that contains the reagents required for identification of the I148M (rs738409) genotype within the PNPLA3 gene from DNA isolated from whole blood or buccal swabs and using the QuantStudio5Dx™ instrument. (Additional instrument compatibility to follow).

The Almac PNPLA3 Genotyping Kit will be used for research use only, offering accurate genotyping of the PNPLA3 gene to provide researchers with better insights into MASH / MASLD biology.

For Research Use Only. Not for use in diagnostic procedures.

Assay Overview

Almac Diagnostic Services has developed the PNPLA3 Genotyping Kit for the Intended Use described above. The Almac PNPLA3 Genotyping kit contains the required reagents to enable allelic discrimination of rs738409 (C>G) within the PNPLA3 gene from 20 ng DNA derived from whole blood or buccal swab specimens. The assay kit includes 3 genotype assay controls (rs738409 homozygous C/C, rs738409 homozygous G/G and rs738409 heterozygous C/G) to ensure confidence in allelic assay calls. DNA is extracted using commercially available kits, and qPCR is performed on the QuantStudio5Dx™ instrument, using on instrument genotyping software.

Feature	Almac PNPLA3 Genotyping Kit
Target	PNPLA3 gene
Alteration	rs738409 I148M
Technique	qPCR
Sample type	Blood and Buccal swabs
Kit reaction size	24 reactions
Components	Assay Primers, Genotyping mastermix + 3 assay controls
Benefits	Simple to use and highly accurate
Performance	Repeatability $\geq 99\%$ Reproducibility $\geq 99\%$
Hands on time	<30 minutes
Input requirements	20ng of DNA
Species category	Human

Table 1 - PNPLA3 Assay Kit specifications

Assay Performance

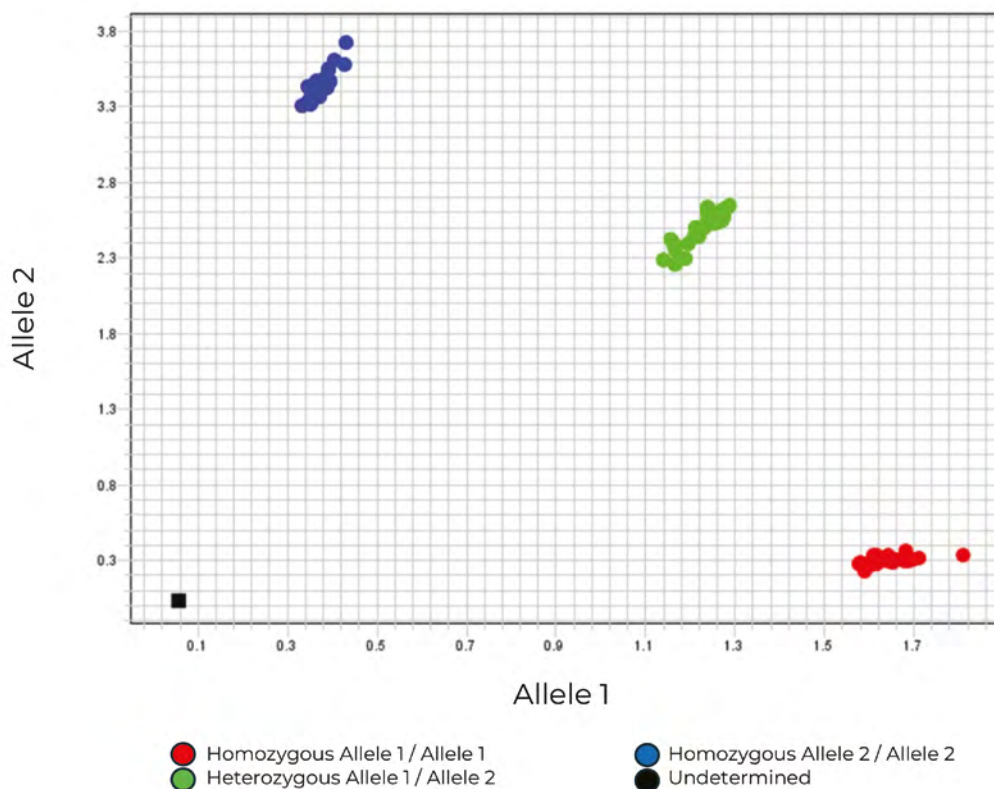


Figure 1 - DNA extracted from MASH patient blood samples and genotyped using the Almac PNPLA3 Genotyping kit

Performance Specifications	
Analytical Precision (UK)	100% [96.57, 100.00]
Analytical Precision (US)	100% [96.47, 100.00]
Analytical Accuracy	
OPA (UK and US)	100% [96.30, 100.00]
Sensitivity (Wildtype Allele)	100% [96.07, 100.00]
Specificity (Wildtype Allele)	100% [60.97, 100.00]
Sensitivity (Alternative Allele)	100% [92.59, 100.00]
Specificity (Alternative Allele)	100% [93.12, 100.00]

Table 2 - Representative assay performance results

To demonstrate the assay capabilities, DNA extracted from samples from patients with confirmed MASH / MASLD were evaluated using the Almac PNPLA3 Genotyping Kit. This demonstrated clear separation of the 3 potential genotypes (Figure 1) and high analytical performance metrics including precision, accuracy, sensitivity and specificity when compared to orthogonal methods (Table 2).

Find out more

Visit our website for more information on the Almac PNPLA3 Genotyping Kit:
almacgroup.com/diagnostics/almac-pnpla3-genotyping-kit

Ordering information

Order the Almac PNPLA3 Genotyping Kit at:

dxshop.almacgroup.com

References

¹ - The global epidemiology of nonalcoholic fatty liver disease (NAFLD) and nonalcoholic steatohepatitis (NASH): a systematic review <https://pubmed.ncbi.nlm.nih.gov/36626630/>

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