MAYO CLINIC LABORATORIES

1-800-533-1710

NAT2Q

N-Acetyltransferase 2 (NAT2) Genotype, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
SA00151655	TESTING, NAT2Q ABNORMAL		1985-03-20	M	37
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00151655	SA00151655	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		22 Mar 2022 08:30			

Results

Analyte	Result	Performing Site
NAT2 Genotype	*5/*5	MCR
NAT2 Phenotype	⚠ Slow Acetylator	MCR

Interpretation

MCR

This individual has two copies of the NAT2 gene encoding enzyme with decreased or no activity. This NAT2 genotype is referred to as a slow acetylator phenotype. This individual is expected to have reduced enzyme activity as compared to individuals with the rapid (normal) acetylator phenotype. Medications metabolized by NAT2, such as isoniazid and amifampridine, should be used with caution.

Method

MCR

Targeted variant analysis was performed to test for the presence of specific NAT2 variants: c.190C>T, c.191G>A, c.341T>C, c.364G>A, c.434A>C, c.499G>A, c.590G>A, and c.857G>A, based on GRCh37 NM_000015.2. Genotyping was performed using a polymerase chain reaction (PCR)-based 5'-nuclease assay. After amplification of the target DNA, fluorescently labeled detection probes were used to determine the nucleotide(s) present. The detection probe(s) that match the target DNA are degraded by 5'-nuclease polymerase, releasing the reporter dye from the effects of the quencher dye, which generates a fluorescent signal. Genotypes are assigned based on the allele-specific fluorescent signals that are detected. (TaqMan SNP Genotyping Assays User Guide, Applied Biosystems)

Disclaimer

[1] MCR

This test will not detect all NAT2 genetic variants. If no detectable NAT2 variant is found, a presumed *4 allele is assigned. Therefore, absence of a detectable variant does not rule out the possibility that a patient has altered NAT2 metabolism due to other NAT2 variants that cannot be detected with this method. Furthermore, when two or more variants are identified, the cis-trans status (whether the variants are on the same or opposite chromosomes) is not known. Therefore, in some cases, multiple potential haplotypes are reported.

CAUTIONS:

Rare variants may be present that could lead to false negative or positive results. If results obtained do not match the clinical findings (phenotype), additional testing should be considered.

Samples may contain donor DNA if obtained from patients who have recently received non-leukoreduced blood transfusions or allogeneic hematopoietic stem cell transplantation (AHSCT). Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have had AHSCT, a pre-transplant DNA specimen is recommended for testing. NAT2 genetic test results in patients who have undergone liver transplantation may not accurately reflect the patient's NAT2 status.

Reviewed by

MCR

BENJAMIN VANSTEINBURG

Received: 22 Mar 2022 15:32 **Reported:** 23 Mar 2022 08:54

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
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