MAYO CLINIC LABORATORIES

1-800-533-1710

# 1A2Q

Cytochrome P450 1A2 Genotype, Varies

| Patient ID | Patient Name | | Birth Date | Gender | Age |
|---|---|---|---|---|---|
| SA00143969 | TESTINGRNV, 1A2Q ABNORMAL | | 1981-09-04 | M | 39 |
| **Order Number** | **Client Order Number** | **Ordering Physician** | **Report Notes** | | |
| SA00143969 | SA00143969 | CLIENT,CLIENT | | | |
| **Account Information** | | **Collected** | | | |
| C7028846 DLMP Rochester | | 06 Apr 2021 00:00 | | | |

## Results

| Analyte | Result | Performing Site |
|---|---|---|
| *CYP1A2* Genotype | *1/*1F | MCR |
| CYP1A2 Phenotype | ⚠ Rapid metabolizer | MCR |

## Interpretation

MCR

This individual is expected to metabolize CYP1A2 substrates at a normal rate, or at a higher than normal rate if CYP1A2 is induced. This individual has a CYP1A2 variant which leads to increased inducibility when exposed to certain substances such as tobacco smoke (see other inducers in the pharmacogenomic associations tables). If CYP1A2 inducers are stopped or started, a change in phenotype is possible. Caution should be exercised when using CYP1A2 substrates because blood levels may be lower than expected when CYP1A2 is induced.

### Method

MCR

Genotyping is performed using a PCR-based 5'-nuclease assay. Fluorescently labeled detection probes anneal to the target DNA. PCR is used to amplify the segment of DNA that contains the polymorphism. If the detection probe is an exact match to the target DNA, the 5'-nuclease polymerase degrades the probe, the reporter dye is released from the effects of the quencher dye, and a fluorescent signal is detected. Genotypes are assigned based on the allele-specific fluorescent signals that are detected. (TaqMan(r) SNP Genotyping Assays User Guide, Applied Biosystems)

### Disclaimer

1 MCR

Targeted variant analysis was used to test for the presence or absence of the following CYP1A2 variant alleles: *1F (c.–9–154C>A), *1K (c.–10+113C>T), *6 (c.1291C>T), and *7 (c.1253+1G>A), based on GRCh37, NM_000761.4.

This method will not detect all variants that result in altered CYP1A2 activity. In the absence of detectable CYP1A2 variants, a presumed *1 genotype is reported. Therefore, absence of a detectable gene variant does not rule out the possibility that a patient has an altered CYP1A2 metabolism status. Furthermore, when two or more gene variants are identified, the cis-/trans-status (whether the variants are on the same or opposite chromosomes) is not always known.

The frequency of variants that cause altered CYP1A2 metabolism has not been fully characterized in all ethnic groups. CYP1A2 enzyme activity may be inhibited or induced by a variety of substances, medications, or their metabolites.

### 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 an allogeneic hematopoietic stem cell transplant. 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 received an allogeneic hematopoietic stem cell transplant, a pre-transplant DNA specimen is recommended for testing.

CYP1A2 genetic test results in patients who have undergone liver transplantation may not accurately reflect the patient's CYP1A2 status.

Test Environment PGx Template

**Performing Site Legend**

| Code | Laboratory | Address | Lab Director | CLIA Certificate |
|---|---|---|---|---|
| MCR | Mayo Clinic Laboratories - Rochester Main Campus | 200 First Street SW, Rochester, MN 55905 | William G. Morice M.D. Ph.D | 24D0404292 |

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**Reviewed by** MCR

Ann Marie Moyer, M.D., Ph.D.

**Received:** 07 Apr 2021 09:32 **Reported:** 07 Apr 2021 16:09

Test Environment PGX Template

## 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.

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