

**CYTOCHROME P450 2D6 GENOTYPE**  
**#83180**

**EXPLANATION:** This assay has been enhanced by an addition of several polymorphisms (SNPs) to the list of potential findings. Also, effective with the detection of the additional SNPs, the individual SNP results will be reported along with the current interpretation field. This will require file definition changes.

**NOTE:** Specimen requirements and fees will remain unchanged.

**CURRENT CPT CODE:**

"Cytochrome P450 2D6 Genotype for Tamoxifen Hormonal Therapy"

83892/Enzymatic digestion

83896/x14 Nucleic acid probe, each

83900/Amplification, target, multiplex, first 2 nucleic acid sequences

83912/Interpretation and report

"Rapid DNA Extraction"

83890

**NEW CPT CODE:**

"Cytochrome P450 2D6 Genotype for Tamoxifen Hormonal Therapy"

83892/Enzymatic digestion

83900/Amplification, target, multiplex, first 2 nucleic acid sequences

83901/x2 Amplification, target, multiplex, each additional nucleic acid

83912/Interpretation and report

83914/x17 Mutation identification by enzymatic ligation or primer

"Rapid DNA Extraction"

83890

**QUESTIONS:** Contact your Mayo Medical Laboratories' Regional Manager  
Shirley Pokorski, Mayo Medical Laboratories' Technologist Support  
Telephone: 800-533-1710



# TEST DEFINITION

5/15/2009

CODE NAME  
 -----  
 83180 CYP2D6 GENOTYPE  
 81769 RAPID DNA EXTRACTION

| ORDER CODE | EFF DATE  | TC    | TITLE                                                                        | CHECKING NORMALS | PRINT NORMALS (# CODED) | PERFORM SITE * |
|------------|-----------|-------|------------------------------------------------------------------------------|------------------|-------------------------|----------------|
| 81769      | 5/23/2007 |       | RAPID DNA EXTRACTION                                                         |                  |                         | MCR            |
|            |           | 28357 | TRANSPORT TEMP : AMBIENT\FROZEN OK\REFRIG OK<br>COMMENT                      |                  |                         |                |
| 83180      | 4/7/2009  |       | CYP2D6 GENOTYPE                                                              |                  |                         | MCR            |
|            |           | 45518 | TRANSPORT TEMP : AMBIENT\FROZEN OK\REFRIG OK<br>2D6 PHENOTYPE INTERPRETATION |                  |                         |                |
|            |           | 84294 | 2D6 GENOTYPE STAR ALLELES                                                    |                  |                         |                |
|            |           | 23649 | 2D6 DUPLICATION                                                              |                  |                         |                |
|            |           | 23648 | 2D6 DELETION                                                                 |                  |                         |                |
|            |           | 23650 | 2D6 -1584c>g (*2A)                                                           |                  |                         |                |
|            |           | 23651 | 2D6 100c>t (*10)                                                             |                  |                         |                |
|            |           | 23652 | 2D6 124g>a (*12)                                                             |                  |                         |                |
|            |           | 30796 | 2D6 138INST (*15)                                                            |                  |                         |                |
|            |           | 23653 | 2D6 883g>c (*11)                                                             |                  |                         |                |
|            |           | 23654 | 2D6 1023c>t (*17)                                                            |                  |                         |                |
|            |           | 23655 | 2D6 1707TDEL (*6)                                                            |                  |                         |                |
|            |           | 23656 | 2D6 1758g>t/a (*8/*14)                                                       |                  |                         |                |

- - - - -  
23657 2D6 1846G>A (\*4)  
- - - - -  
23658 2D6 2549ADEL (\*3)  
- - - - -  
23659 2D6 2613AGADEL (\*9)  
- - - - -  
23660 2D6 2850C>T (\*2)  
- - - - -  
23661 2D6 2935A>C (\*7)  
- - - - -  
26373 2D6 2988G>A (\*41)  
- - - - -  
45519 2D6 GENOTYPE INTERPRETATION  
- - - - -  
23662 2D6 REVIEWED BY

POSSIBLE RESULT VALUES INCLUDE : DENNIS J. O'KANE, PH.D., JOHN L. BLACK, M.D., LINNEA M. BAUDHUIN, PH.D., LORALIE J. LANG.

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\*PERFORMING SITE LEGEND

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MCR MAYO CLINIC DPT OF LAB MED & PATHOLOGY  
200 FIRST STREET SW  
ROCHESTER, MN 55905

LAB DIRECTOR: FRANKLIN R. COCKERILL, III, M.D.

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## LABORATORY SERVICE REPORT

1-800-533-1710

|                                                                                                  |                              |                                    |             |                                        |                       |                         |
|--------------------------------------------------------------------------------------------------|------------------------------|------------------------------------|-------------|----------------------------------------|-----------------------|-------------------------|
| PATIENT NAME<br>TESTING, JAMIE                                                                   |                              | PATIENT NUMBER                     |             | AGE<br>29                              | SEX<br>F              | ACCESSION #<br>G9127015 |
| ORDERING PHYSICIAN                                                                               |                              | CLIENT ORDER #                     |             |                                        | ACCOUNT #<br>LIAISONS |                         |
| COLLECTION<br>05/14/09 09:45 A                                                                   | RECEIVED<br>05/14/09 09:45 A | REPORT PRINTED<br>05/15/09 02:57 P |             | SPECIMEN INFORMATION<br>DATE OF BIRTH: |                       |                         |
| <b>DATE</b> <b>TIME</b>                                                                          | <b>DATE</b> <b>TIME</b>      | <b>DATE</b>                        | <b>TIME</b> |                                        |                       |                         |
| Test Client<br>Attn: Mayo Liaisons<br>200 First Street SW<br>Rochester, MN 55905<br>507-284-8202 |                              |                                    |             |                                        |                       |                         |

| TEST REQUESTED | HI<br>LO | REF RANGE | PERFORM SITE * |
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**Rapid DNA Extraction**

Comment

Genomic DNA was  
extracted.

MCR

**CYP2D6 Genotype****2D6 Phenotype**

Interpretation

Predicted intermediate metabolizer

This patient has a genotype associated with reduced enzyme activity. This patient will have reduced enzyme activity as compared to individuals with the normal genotype. Some caution should be exercised when treating with drugs metabolized by CYP2D6. There is a small residual risk of having an additional rare, undetected polymorphism which may result in poor metabolizer status. This should be considered if the predicted phenotype is discordant with clinical findings.

**2D6 Genotype Star**

1/4

MCR

Alleles

See <http://www.cypalleles.ki.se/cyp2d6.htm> for a full description of CYP2D6 alleles.

**2D6 Duplication**

NO DUP

MCR

**2D6 Deletion**

NO DEL

MCR

**2D6 -1584c>g (\*2A)**

C/C

MCR

**2D6 100c>t (\*10)**

C/T

MCR

**2D6 124g>a (\*12)**

G/G

MCR

**2D6 138inst (\*15)**

WT

MCR

**2D6 883g>c (\*11)**

G/G

MCR

**2D6 1023c>t (\*17)**

C/C

MCR

**2D6 1707tdel (\*6)**

T/T

MCR

**2D6 1758g>t/a (\*8/\*14)**

G/G

MCR

**2D6 1846g>a (\*4)**

G/A

MCR

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|--------------------------------|-----------------------|----------------------------------------------|
| PATIENT NAME<br>TESTING, JAMIE | ORDER STATUS<br>Final | COLLECTION DATE AND TIME<br>05/14/09 09:45 A |
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| TEST REQUESTED      | HI<br>LO | REF RANGE | PERFORM SITE * |
|---------------------|----------|-----------|----------------|
| 2D6 2549adel (*3)   | A/A      |           | MCR            |
| 2D6 2613agadel (*9) | WT       |           | MCR            |
| 2D6 2850c>t (*2)    | C/C      |           | MCR            |
| 2D6 2935a>c (*7)    | A/A      |           | MCR            |
| 2D6 2988g>a (*41)   | G/G      |           | MCR            |
| 2D6 Genotype        |          |           | MCR            |

**Interpretation**

This patient has one copy of a null allele and one copy of an allele with normal CYP2D6 activity. Additional descriptions of the effects of the star alleles on CYP2D6 function are found in the Mayo Test Catalog (<http://www.mayomedicallaboratories.com/test-catalog/>).

**2D6 Reviewed by**

Dennis J. O'Kane, Ph.D.

MCR

Direct polymorphism analysis for -1584C>G, 100C>T, 124G>A, 138insT, 883G>C, 1023C>T, 1707T>del, 1758G>T, 1758G>A, 1846G>A, 2549A>del, 2613delAGA, 2850C>T, 2935A>C, 2988G>A, CYP2D6 gene deletion, and gene duplication is performed following PCR amplification. Direct DNA testing will not detect all the known mutations that result in decreased or inactive CYP2D6. Absence of a detectable gene mutation or polymorphism does not rule out the possibility that a patient has an intermediate or poor metabolizer phenotype. Based on the test sensitivity and currently available CYP2D6 polymorphism carrier frequencies, persons of Caucasian descent who tested negative for the above polymorphisms would be estimated to have a less than 1.4% residual risk for carrying one or more copies of an undetected poor metabolizer allele. This residual risk may be higher or lower in other ethnic groups. The frequency of polymorphisms causing poor metabolism is highest in the Caucasian population and lower in African Americans and Asians. Patients with an extensive (normal) or intermediate metabolizer genotype may have CYP2D6 enzyme activity

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inhibited by a variety of medications, or their metabolites. The following is a partial listing of drugs known to affect CYP2D6 activity as of the date of this report.

## Drugs known to increase CYP2D6 activity:

## Dexamethasone

Co-administration of this drug will increase the rate of excretion of CYP2D6 metabolized drugs, reducing that drug's effectiveness.

## Drugs known to decrease CYP2D6 activity:

Amiodarone, bupropion, celecoxib, chlomipramine, chlorpheniramine, chlorpromazine, cimetidine, citalopram, cocaine, doxorubicin, duloxetine, fluoxetine, haloperidol, indinavir, methadone, metochlopramide, paroxetine, quinidine, ranitidine, ritonavir, sertraline, and triclopidine.

Co-administration will decrease the rate of metabolism of CYP2D6 metabolized drugs, increasing the possibility of toxicity.

## Drugs that undergo metabolism by CYP2D6:

Alprenolol, amitriptyline, amphetamine, aripiprazole, atomoxetine, chlorpromazine, clomipramine, codeine, desipramine, dextromethorphan, diltiazem, disopyramide, duloxetine, encainide, felbamate, flecainide, fluoxetine, fluvoxamine, haloperidol, iloperidone, imipramine, labetalol, lidocaine, mephobarbital, metoprolol, mexilitine, mirtazapine, nortriptyline, oxycodone, paroxetine, perhenazine, phenformin, propafenone, propranolol, respirdone, sertraline, tamoxifen, thioridazine, timolol, tranadol, and venlafaxine.

Co-administration may decrease the rate of elimination of other drugs metabolized by CYP2D6.

## Investigational Use Only

## \* PERFORMING SITE

|     |                                        |                                                |
|-----|----------------------------------------|------------------------------------------------|
| MCR | Mayo Clinic Dpt of Lab Med & Pathology | Lab Director: Franklin R. Cockerill, III, M.D. |
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| 200 First Street SW Rochester, MN 55905 |          |           |                |

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