MAYO CLINIC
Mayo Medical Laboratories

TEST DEFINITION

12/6/2011

NAME/ORDER CODE CROSS-REFERENCE

CODE	NAME
2D6TO	CYP2D6 TAMOXIFEN GENOTYPE, SALIVA

TEST SETUP INFORMATION

ORDER CODE	RESULT CODE	TITLE	CHECKING NORMALS	PRINT NORMALS	PERFORM SITE *
2D6TO		CYP2D6 TAMOXIFEN GENOTYPE, SALIVA			MCR

TRANSPORT TEMP : AMBIENT

32947	2D6 TAMOXIFEN GENOTYPE STAR ALLELES	UNITS:
32948	2D6T DUPLICATION	UNITS:
32949	2D6T DELETION	UNITS:
32950	2D6T -1584C>G (*2A)	UNITS:
32951	2D6T 100C>T (*10)	UNITS:
32952	2D6T 124G>A (*12)	UNITS:
32953	2D6T 138INST (*15)	UNITS:
32954	2D6T 883G>C (*11)	UNITS:
32955	2D6T 1023C>T (*17)	UNITS:
32956	2D6T 1707TDEL (*6)	UNITS:
32957	2D6T 1758G>T/A (*8/*14)	UNITS:
32958	2D6T 1846G>A (*4)	UNITS:
32959	2D6T 2549ADEL (*3)	UNITS:
32960	2D6T 2613AGADEL (*9)	UNITS:

ORDER CODE	RESULT CODE	TITLE	CHECKING NORMALS	PRINT NORMALS	PERFORM SITE *
2D6TO (CONTINUED...)					
	32961	2D6T 2850C>T (*2)			
			UNITS:		
	32962	2D6T 2935A>C (*7)			
			UNITS:		
	32963	2D6T 2988G>A (*41)			
			UNITS:		
	32964	2D6 TAMOXIFEN GENOTYPE INTERP			
			UNITS:		
	32965	2D6 TAMOXIFEN REVIEWED BY			
			UNITS:		
	32966	2D6 TAMOXIFEN PHENOTYPE INTERP			
			UNITS:		

TEST CODE ALWAYS MESSAGE - [NP0089]

NP0089 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 PERCENT 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 INHIBITED BY A VARIETY OF MEDICATIONS, OR THEIR METABOLITES. BECAUSE ANTIDEPRESSANTS ARE OFTEN PRESCRIBED TO ALLEVIATE THE HOT FLASHES THAT ACCOMPANY TAMOXIFEN THERAPY, IT IS PARTICULARLY IMPORTANT TO UTILIZE AN ANTIDEPRESSANT THAT DOES NOT COMPROMISE CYP2D6 ACTIVITY, WHICH COULD REDUCE TAMOXIFEN'S EFFICACY. THE FOLLOWING IS A PARTIAL LISTING OF DRUGS KNOWN TO AFFECT CYP2D6 ACTIVITY AS OF THE DATE OF THIS REPORT.

DRUGS THAT INHIBIT CYP2D6 SIGNIFICANTLY: AMIODARONE,

BUPROPION, CIMETIDINE, CINACALCET, COCAINE, DEXMEDETOMIDINE,

TEST SETUP INFORMATION

ORDER CODE	RESULT CODE	TITLE	CHECKING NORMALS	PRINT NORMALS	PERFORM SITE *
2D6TO (CONTINUED...)					

DULOXETINE, FLUOXETINE, PAROXETINE, PERAZINE, PERPHENAZINE,
PERGOLIDE, PIMOZIDE, QUINIDINE, SERTRALINE, TERBINAFINE,
THIORIDAZINE, AND TICLOPIDINE.

ADDITIONAL FACTORS MAY MODULATE THE TREATMENT RESPONSE TO TAMOXIFEN. EXPRESSION OF ESTROGEN RECEPTOR BETA BY THE TUMOR, IN THE ABSENCE OF ESTROGEN RECEPTOR ALPHA, IS A FAVORABLE PROGNOSTIC INDICATOR FOR TAMOXIFEN THERAPY. HOWEVER, EXPRESSION OF THE HER2 RECEPTOR BY THE TUMOR MAY INDICATE TAMOXIFEN RESISTANCE EVEN IF THE CYP2D6 GENOTYPE INDICATES ACTIVATION OF SUFFICIENT ENDOXIFEN.

TEST CODE ALWAYS MESSAGE - [TZASR]

TZASR ANALYTE SPECIFIC REAGENT. THIS TEST WAS DEVELOPED AND ITS PERFORMANCE CHARACTERISTICS DETERMINED BY MAYO CLINIC. IT HAS NOT BEEN CLEARED OR APPROVED BY THE U.S. FOOD AND DRUG ADMINISTRATION.

*PERFORMING SITE LEGEND

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*** END OF REPORT ***