

Gene test information

**PHARMACOGENETICS OF TAMOXIFEN
(CYP2D6 GENE TEST)**

- **Background**

Tamoxifen, a molecule essentially prescribed for the treatment of breast cancer, is a pro-drug which must be activated by the enzyme CYP2D6 before becoming pharmacologically active. In post-menopausal women, CYP2D6 can become an indirect indicator of the after-effects of primary breast cancer being treated with tamoxifen.

- **CYP2D6 genotypes**

| Class | Frequency | Genotypes (examples) | Commentary |
|----------------------------|-----------|----------------------|------------------------------------|
| Extensive metabolizer (EM) | 92% | *1*1, *1*4, *1*3 | No sign of reduced CYP2D6 activity |
| Poor metabolizer (PM) | 8% | *4*4, *4*6, *3*4 | Strongly reduced CYP2D6 activity |

The gene tests includes the most important non-functional CYP2D6 alleles (*3, *4, *6).

- **Indications for testing**

Individuals starting tamoxifen therapy.

References:

Punglia RS, Burstein HJ, Winer EP, Weeks JC. Pharmacogenomic variation of CYP2D6 and the choice of optimal adjuvant endocrine therapy for postmenopausal breast cancer: a modeling analysis. *J Natl Cancer Inst.* 2008;100:642-8.

Goetz MP, Kamal A, Ames MM. Tamoxifen pharmacogenomics: the role of CYP2D6 as a predictor of drug response. *Clin Pharmacol Ther.* 2008;83:160-6.