

Molecular Diagnostics Laboratories Informed Consent for DNA Testing for patients

My physician/ genetic counselor _____ suggested that an attempt be made to assess the probability that I have an inherited gene for one or more of the following: (1) increased clotting risk, (2) taking or about to take the drug warfarin (Coumadin), (3) inability to metabolize the anticancer drug 5-fluorouracil (5FU) and some of its analogues, (4) hyperlipidemia, (5) hypertension, (6) emphysema and liver disease. The test to be done at Molecular Diagnostics Laboratories, Inc (MDL) will look for specific mutations or variants*. **(Circle those that apply below):**

(1) a) **Factor V Leiden**, b) **MTHFR C677T** and/or **A1298C**, c) **Glycoprotein GpIIa**, d) **Prothrombin**, e) **PAI** gene variants, f) **eNOS**, h) **Stromelysin**. Individuals having any combination of these genetic mutations typically have an increased risk of blood clotting. The absence of these specific mutations does not rule out the possibility of increased risk for clotting from other factors.

(2) **Warfarin Panel**. Genotyping of CYP2C9 and VKORC1 for metabolism status to determine proper initial dosage of warfarin to avoid bleeding events and overdose.

(3) **DPD** (DihydroPyrimidine Dehydrogenase) enzyme. A defect in the activity of this enzyme due to a mutation will result in my inability to metabolize some drugs like 5FU. The absence of this specific mutation does not rule out the inability to metabolize 5-FU or its analogues.

(4) **Apo E** has three alleles; E2, E3 and E4. The E3 allele is the most common normal allele. The E2 allele is associated with Type III hyperlipidemia. The E4 is associated with hypercholesterolemia and hyper-lipoproteinemia. Absence of the E2 or the E4 alleles does not rule out the possibility of hyperlipidemia due to some other reason.

(5) **ACE** (Angiotensin Converting Enzyme). The D allele is associated with left ventricular hypertrophy, myocardial infarction, increased plaque instability, and other cardiovascular diseases. Absence of the D allele does not rule out the possibility of cardiovascular disease due to other factors.

(6) **UGT1A1**. Camptosar (irinotecan) metabolism. To determine if treatment is toxic to patient or to modify dosage.

(A fact sheet for each test is available from MDL)

1. The test detects an abnormality, called a mutation in the gene using my DNA. The test procedure is specific to the genetic condition described above and cannot determine the complete genetic makeup of an individual.

2. The DNA for this test will be obtained from a blood sample that will be taken by inserting a needle in my vein. This may cause pain, soreness, bruising and rarely infection. Experienced individuals will perform this procedure.

3. The test is approximately 99% accurate for detection of the specific mutation although it is not an FDA-approved test.

4. Given the limitations in current technology of this type there is a possibility exists that the test will not work properly or that an error will occur. (currently estimated to be 1 in 1000).

5. No other tests (other than those suggested by my physician/counselor and authorized by me) shall be performed in my samples (unless I consent to additional tests herein). I understand that MDL (the "Laboratory") is not a DNA banking facility and the unused sample will be destroyed after completion of test (within 60 days). I understand that my sample may be used for research only after all identifiers have been removed from the sample.

Patient's Initials

6. The results of my test will be reported to me only through a physician or genetic counselor that I designate. I understand that a) a negative test result does not mean that I am not susceptible for the disease I am being tested for and b) a positive test result is an indication that I am predisposed to or have the specific disease or condition tested for, and I may wish to consider further independent testing, consult with my physician or pursue genetic counseling.

I hereby designate the following individual to receive this information:

(physician or counselor)

7. The Laboratory will maintain my test results in confidentiality and will release them only to other medical professionals, insurance companies or other parties with my written consent. The test results may become part of my medical record if my physician records the results therein.

8. I understand that participation in DNA testing is completely voluntary and I may withdraw my consent at any time and request destruction of DNA sample without penalty.

All of the above has been discussed with me, and my questions answered by my physician/counselor

Patient Name (Print)

Patient Signature

Witness

Date

Date

Physician's/Counselor's Statement: I have explained DNA testing to this individual, addressed the limitation outlined above, and answered this person's questions.

Signature: _____ Date: _____

*Note to physician: The specific gene mutations or variants tested, when requested, are:
Factor V Leiden mutation Prothrombin G20210A
MTHFR: C677T and/or A1298C PAI: 4G vs. 5G variant in the promotor region
A-1-AT: S and Z mutations Apo E: E2, E3 and E4 alleles
ACE: D and I variants CBS: T833C and the G919A mutations
Glycoprotein IIIa: P1A1 vs. P1A2 variants DPD: gt to at mutation in intron 13
Thrombospondin-4 (A387P) & Thrombospondin 1 (N700S) Mutations
Stromelysin-1 5A/6A eNOS T-786C

Additional Abbreviations: MTHFR; MethyleneTetraHydroFolate Reductase; PAI; Plasminogen Activator Inhibitor CBS; Cystathionine-b-Synthase,

eNOS; Endothelial Nitric Oxide Synthase

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