## **Quick Reference Guide for Using the Test/Laboratory Data Table**

We offer this guide to facilitate a discussion with your oncologist or genetic counselor <u>before you begin</u> chemotherapy with fluoropyrimidines. Here we list different types of tests to determine the possibility of DPD deficiency so that your oncologist may use the results to guide treatment.

The list identifies different labs, their tests, information about the tests, and who may order the test. Unfortunately, our research has been unable to obtain much useful information concerning cost.

Disclaimer: we do not prefer or recommend any lab over another; we simply recommend you get the best test available that suits you.

Below briefly serves to describe each column on the spreadsheet:

- <u>DPYD variants</u>: this shows which known variants of the DPYD gene are tested (see Table below for the variants and aliases). Generally, the more the better unless the test is performed using "sequencing" which may identify even more variants that could put you at risk.
- <u>Lab Name</u>: self-explanatory.
- <u>Test Name:</u> self-explanatory.
- <u>Test Indication:</u> this identifies how the lab will use your sample to identify your potential risk of DPD deficiency. Labs may use different methods to screen:
  - Gene/condition specific: the lab will look for predetermined DPYD variants as identified in the first column.
  - o PGx Panel: the DPYD test may be included in a screening of other genes.
  - Sequencing: this will assess and report your full genetic sequence of the DPYD gene, regardless
    of whether it matches a clinically significant genotype.
- # DPYD Variants Tested: this shows the # of variants tested for DPYD gene specific tests unless it is a sequence indicated test which will look for all variants of the DPYD gene.
- Turn Around Time: time to process test results.
- Cost: of the lab to process and report results; excludes any cost your oncologists office may add.
- <u>Physician or Patient Initiated:</u> identifies who may order the test.
- How to Order: self-explanatory.
- Sample Type: identifies what is collected from the patient for the assessment.
- Links: a web link to the lab.
- Other Notes: identifies certifications of the lab. The CLIA program ensures quality lab testing; the Centers for Medicare & Medicaid Services manage this certification program.

## Table of DPYD Alleles and Aliases

| * Allele | rsID  | Aliases                                     |
|----------|---|---|
| *2A      | rs3918290   | IVS14+1G>A, c.1905+1G>A                     |
| *13      | rs55886062  | c.1679T>G, p.1560S                          |
| NA       | rs67376798  | c.2846A>T, p.D949V                          |
| NA       | rs56038477 (Linkage<br>Disequilibrium w/rs75017182) | 1236G>A, p.E412E (LD w/1129-5923C>G, HapB3) |
| NA       | rs115232898   | c.557A>G, p.Y186C                           |