

## How is pharmacogenomic testing different from “traditional” molecular diagnostic testing?

*The GeneSight® test is a pharmacogenomic test - not a diagnostic test.*

Diagnostic testing and pharmacogenomic testing are two different types of genetic tests. In some ways they are similar; both may analyze DNA to identify changes that may alter structure, function, and expression of proteins. However, there are meaningful differences between the two.

### What is the purpose of each type of test?

**Diagnostic testing:** Looks at changes in an individual's DNA that are associated with a disease state (e.g. testing for Huntington's disease). A mutation may be diagnostic of a disease, and lack of the mutation may help rule out the disease.

**Pharmacogenomic testing:** Looks at changes in an individual's DNA that influence how that person may metabolize (pharmacokinetic markers) or respond (pharmacodynamic markers) to medications. Pharmacogenomic testing can help inform medication selection and dosing, but it cannot identify which medications or doses will definitely work for a patient. It also does not diagnose the patient with any disease.

### Who are the target populations for each type of test?

**Diagnostic testing:** Individuals who are suspected of having, or are at risk to develop, a specific genetic disease. Depending on the disease pattern of inheritance, family members of these individuals may also be candidates for molecular diagnostic testing.

**Pharmacogenomic testing:** Individuals who are being considered for medications, changes in medications, or dose adjustments.

### How does each type of test fit into the psychiatric space?

**Diagnostic testing for psychiatric illness is not currently possible.** There is no single genetic marker or group of markers that are able to reliably diagnose any particular psychiatric illness. Psychiatric disease is multifactorial, which means that multiple genes as well as environmental factors are involved. While some genes have been implicated in the risk of developing certain conditions such as schizophrenia and bipolar disorder<sup>1-3</sup>, they contribute only a small amount to the overall disease risk. Without a proper understanding of the effect of these variants, this type of genetic testing may lead to overestimation of disease risk and could cause undue patient anxiety regarding the development of serious conditions.

**Pharmacogenomic testing for psychiatric medications can help inform medication selection and dosing.** Pharmacogenomic testing may provide benefit by supporting providers during the medication selection process. Testing pharmacokinetic and pharmacodynamic markers may help patients get on the right medication faster. This may help patients feel empowered by their genetic knowledge and its role in their treatment. While there are many pharmacogenomic tests on the market, many of them lack proof of efficacy. Therefore, it is important to learn whether a given test has clinical proof of efficacy. Multiple peer-reviewed published studies have demonstrated the clinical validity, clinical utility, and economic utility of the GeneSight test.<sup>4-11</sup>

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