Genetic Risk Assessment: Frequently Asked Questions

What is a genetic risk assessment?
A genetic risk assessment is a service that assesses your personal history, family history and determines whether you are at an increased risk for any specific genetic (inherited) conditions. Genetic testing options will be discussed and if testing is performed, a follow up consultation will discuss results and next steps.

Who might benefit from a genetic risk assessment?
Genetic assessments can be helpful at many different points of your life. Some people consider genetic testing before they have a baby, or during pregnancy, to help assess potential risks to a future baby. Genetic assessments and testing can be helpful for children or adults who have multiple medical features that could have an underlying genetic cause(s). Genetic assessments and testing can also be helpful for individuals with a family history of a specific condition in multiple family members, particularly at a younger-than-expected age.

Is there anything I should do to prepare for my genetic risk assessment appointment?
It can be helpful to reach out to family members to obtain medical history and in particular to learn about whether any specific diagnoses or genetic tests have been performed on any of your close biologic family members (parents, siblings, aunts/uncles, grandparents). We will focus on conditions occurring in multiple generations; those that are known to be genetic (with or without a genetic test to document it within the family) and those common adult disorders that are more common in your family, particularly at < 50 years old. If genetic testing has been performed on a relative, it’s helpful to have a copy of their report, or at least know the specific mutation information to facilitate testing in yourself.

What kinds of genetic tests might be recommended?
Diagnostic or predictive genetic tests will be recommended on the basis of your personal and family history. In some cases we recommend initially testing an affected relative for the most accurate genetic test interpretation. In other cases we may offer you a test for a single gene or panel of related genes. Other types of genetic testing include carrier testing (most useful when thinking about childbearing) and pharmacogenomics testing (which can help inform specific prescriptions and dosing). We do not currently offer Direct-To-Consumer testing or interpretation, ancestry testing, or recommend whole exome/genome testing on otherwise healthy individuals.

How is genetic testing performed?
Typically, genetic testing entails a blood test, which can often be obtained at the same time as other blood that is being drawn for other testing purposes. Some genetic testing companies have started offering testing from a “buccal (cheek) swab” but this is very limited at this time.

How much does genetic testing cost?
There is a wide range of costs for genetic testing (several hundred dollars to several thousand), and it is often, but not always covered by medical insurance. You can choose to submit your genetic testing to insurance, but we will not perform a pre-authorization so you would be responsible for charges (ranging up to several thousand dollars) if not covered.
Should I be worried about discrimination on the basis of genetic testing? The 2008 Genetic Information Nondiscrimination Act provides federal protection against health insurance and employment discrimination on the basis of predictive genetic information, and many states have supplemental laws to protect genetic privacy. You can find more information about it at [http://www.genome.gov/10002077](http://www.genome.gov/10002077) but you should note that it does not provide protection against discrimination in life insurance, disability or long term care insurance, and it does not apply to companies with fewer than 15 employees.