Precision medicine is often used interchangeably with “personalized medicine” and “individualized medicine”. All of these terms describe an approach to healthcare that focuses on individual differences in genetics, environment, and lifestyle rather than a “one size fits all” kind of care. One aspect of precision medicine uses tests for certain biomarkers, like genetic variations, that will help to identify which groups of people will benefit from which targeted treatments and prevention plans.

Only recently has technology advanced enough to allow researchers to have the ability to test for these genetic variations. Some of these tests involve one or a few genes (genetic testing) or many or all of the genes (whole genome testing). While whole genome testing is available, the interpretation remains very complex and carries with it the potential for a lot of uncertainty.

However, the National Institutes of Health (NIH) has been charged with a long-term research endeavor called the Precision Medicine Initiative to help us learn how this kind of care can help society. With the information generated from this research we can begin to understand how individual differences in lifestyle, environment, and genetics can change disease expression.

Most of the precision medicine genetic testing involves panels of genes. This means there are several genes that may fall into different categories, but it’s important to realize that not all of these genes have been thoroughly studied yet and may not provide actionable information. Some laboratories will offer the testing by category, some will offer combinations of categories, and some will offer it as a comprehensive test covering many different health interests. Much of research in the field is very new and it may be wise to have some caution. We encourage you to discuss it more thoroughly with your care team or an expert.

As with other types of genetic testing it is important for you to consider the implications of precision medicine information. Deciding on whether or not to have any genetic test is a very personal decision. Some people choose to perform genetic testing because they feel it will empower them to make informed decisions that will lower the risk for disease. Some people do not feel that they are ready to know what could be revealed by genetic testing. This can be due to many different reasons, including feeling overwhelmed by other health concerns, feeling that the test results would not affect how they approach their medical care, or not wanting to know about risks that have no treatment options. These are all important issues to consider when deciding about precision medicine genetic testing.

Genetic Support Foundation is a resource for you - translating the science and helping you
understand what it means for you and your family. We have information on a variety of different genetic tests that are currently on the market, but as this is a rapidly changing landscape we will continue to keep you updated on many new and exciting discoveries generated by important studies like the Precision Medicine Initiative. We have broken down the different categories of precision medicine genetic testing; please keep in mind that some of these tests are for informational purposes and not recommended for altering medical decisions:

- Tumor testing
- Predictive testing
- Prognostic testing
- Direct-to-Consumer testing
- Genetic Carrier Screening
- Pharmacogenomic testing
- Ancestry testing

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

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