

Imagine the following office encounter: your patient with type 2 diabetes, and benign multinodular goiter who has come for a routine visit is asking about a new genetic test she heard about in direct to consumer advertisements. The test which costs around \$ 500 promises to screen her genome for variants that confer risk for “heart attack” and “cancer”. Since she is worried about her risk for coronary events and thyroid cancer she wants to do the test and asks your opinion. This scenario is not at all unlikely given the genomics revolution that happened in the past few years resulting in the availability of many genetic and genomic tests, some offered direct to consumer. Therefore, knowledge of the strengths and pitfalls of personalized medicine is essential for all practicing physicians.

The basic premise of personalized medicine is that “one size fits all” medicine does not work well. Traditionally medicine tried to group diseases and their treatments into unified categories in order to facilitate rationale diagnostic and treatment decisions. However, the same disease in different individuals will progress differently, develop complications differently, and will respond to medications in varying ways. Therefore, there is a need to individualize therapy for patients. While individualization of therapy encompasses many factors such as environmental exposures, diet, treatment adherence, and patient’s culture and beliefs, personalized medicine specifically focuses on individualizing therapy based on molecular and genomic markers. For example, the response of breast cancer to chemotherapy is well-known to be influenced by the genes expressed by the tumor (estrogen receptor, HER2). Incorporating molecular biomarkers and genetic information into clinical practice is already happening and some genetic tests have already received FDA approval. To name a few recent examples, variants in the TPMT gene are tested to predict bone marrow aplasia in patients who are treated with azathioprine and 6-mercaptopurine; variants in the VKORC1 and CYP2C9 genes can predict response to Warfarin, and many more genetic variants are now in different stages of testing. Therefore, it is clear that we are at the beginning of a new age in medicine where personalized medicine will be integrated into routine care.

Genetic and molecular biomarker testing could offer critical information for patient care decisions. They could offer information about disease susceptibility, which is at the essence of preventive care. They could provide information about disease progression and likelihood of complications, which is critical when deciding how aggressive therapy should be, for example in thyroid cancer. And they could offer critical information about response to medications. Incorporating genetic and biomarker information into predicting positive and adverse responses to drug therapy is the basis of the new field of pharmacogenomics. Pharmacogenomics will transform medicine as it will enable physicians to screen patients for genetic variants that will make them respond to a medication, will help determine the dose, and will also identify patients that will likely develop adverse effects. Such a test would be of tremendous help, for example when deciding on thionamide therapy for Graves’ disease, or when giving new kinase inhibitors for thyroid cancer.

The ATA Spring Symposium aims to introduce personalized medicine to practicing thyroidologists. The meeting will emphasize both general concepts in personalized medicine as well as specific issues relating to thyroid disease.