Brian Shuch, MD, uses genetic profiling to guide the management of renal tumors. Among the many roles that physicians assume is that of interpreter. Doctors must be adept at interpreting a patient’s symptoms and test results to reach the right diagnosis. Then they must translate that diagnosis into terms that a patient can understand.

In his cutting-edge work treating patients with kidney cancers, Brian Shuch, MD, Assistant Professor of Urology and of Radiology and Biomedical Imaging, has proven himself a gifted interpreter. By evaluating tumors not by shape and size—as has been standard protocol for decades—but by genetic profile, Dr. Shuch has launched an entirely new dialogue about the identification and treatment of these cancers. But as his patients can attest, Dr. Shuch also has a talent for conveying the results of this incredibly complex genetic testing with simple analogies that help them clearly understand their diagnosis and treatment options.

A tumor’s genetic profile reveals crucial information: the type of kidney tumor, whether it’s benign or cancerous, what kind of kidney cancer, and how aggressive it is. “Some renal tumors may not need to have surgery immediately,” he said. “Some can be faithfully observed up to a certain size.” He considers these tumors to be “guppies”—not harmful—as compared to “sharks,” which require immediate, aggressive treatment.

Before his consult with Dr. Shuch, patient Jerome Serling, DMD had already been diagnosed with presumptive renal cell carcinoma. The traditional protocol would be to remove the lesion or kidney. But Dr. Shuch first suggested Dr. Serling undergo a renal mass biopsy with genomic profiling to determine if the lesion was, in fact, dangerous. To Dr. Serling’s great relief, his lesion was a “guppy.”

Brian Shuch's groundbreaking work on genetic profiling holds promise not only for his current patients but for their families as well. “We’ll be able to screen their family members and children to detect a tumor before it causes a problem,” he explained. “By casting a wider net and testing all the genes at once, we have a better chance of finding the cause of someone’s predisposition.”

Dr. Shuch’s use of the test was very grateful for Dr. Shuch’s use of the test and his keen clinical judgement.”

With support from an $800,000 NIH grant, Dr. Shuch is continuing his research on genetic profiling, focusing on the heterogeneity of small renal tumors. “Sometimes tumors can be a mixture of different types of cells,” he explained. “It can be like a bag of M&M’s with different colors. We know that for small tumors, when you stick your hand in a bag and find a green M&M, then most of the bag is going to be green. But for large tumors, if you pull one sample, it might be a red M&M, but in the bag, there will likely be a lot more different colors.”

The importance of this to kidney cancer research is that, if you profile one part of the tumor and get an answer that this tumor is bad or good, you want to be sure that you are giving the patient the best idea of what is in the whole bag of M&M’s,” he said. “You don’t want to base your diagnosis on an incomplete sample.”

Another aspect of integrating genetics to clinical care is how Dr. Shuch evaluates individuals with suspected hereditary forms of cancer. An estimated five to eight percent of kidney cancer patients have a strong hereditary or genetic component. Because of the rarity of these cancers, many caregivers are not familiar with the distinct symptoms associated with them. The median age of onset for hereditary kidney cancer is around 36—about 30 years younger than other kidney cancer patients. Instead of a single tumor, patients may have bilateral or multifocal tumors. A handful of hereditary kidney tumors are diagnosed before the patient has had treatment and gone through the entire diagnostic process.

Testing multiple genes at once significantly streamlines and expedites the diagnostic process. “Each of the known kidney cancer syndromes are linked to a specific gene of interest that may not be able to be identified clinically,” he explained. “By casting a wider net and testing all the genes at once, we have a better chance of finding the cause of someone’s predisposition.”

Dr. Shuch’s groundbreaking work on genetic profiling holds promise not only for his current patients but for their families as well. “We’ll be able to screen their family members and children to detect a tumor before it causes a problem,” he explained. “Because where do big problems come from? Little problems. If we can find it early and fix it, we can prevent them from having significant harm to their livelihood.”