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Getting a Correct Diagnosis Is Vital for Treating Sarcoma, a Rare Cancer

By Julie Grisham, Tuesday, July 31, 2018



Pathologist Cristina Antonescu has focused exclusively on sarcoma for more than 20 years.

Summary

Sarcomas make up only about 1% of all cancers, yet there are more than 50 different types.

Receiving a diagnosis of **sarcoma** can be overwhelming. But it can be especially confusing because many people have never heard of sarcoma and many doctors have never treated it. It's so rare that it makes up only about 1% of all cancers.

Unlike tumors that occur in a particular organ, sarcomas can appear almost anywhere in the body. There are more than 50 different kinds. The differences arise from their location, their tissue of origin, and the genetic changes that drive them. These factors mean making an accurate diagnosis can be a challenge.

Memorial Sloan Kettering pathologist Cristina Antonescu's work has focused exclusively on sarcoma for more than 20 years. "In this new era of personalized medicine, it's important to know what's driving a tumor to identify the best treatment approach," she says.

Determining a sarcoma tumor's type based on its appearance alone can be difficult.

Soft tissue sarcoma arises in connective tissues, which include fat, muscle, tendons, blood vessels, and cartilage. It's diagnosed in about 13,000 people in the United States every year. Additionally, about 3,500 **bone sarcomas** are diagnosed annually in the United States.

Members of MSK's sarcoma team have expertise not only in diagnosing but also in treating all different types and subtypes of sarcoma, some of which are potentially deadly. Some sarcomas can be treated with surgery alone. Others may require a more wide-ranging approach to offer the best chance of fully eliminating the tumor and **preventing it from coming back**. Treatments may include chemotherapy, radiation therapy, immunotherapy, and targeted drugs.

In recognition of MSK's expertise in treating sarcoma, the team recently received a SPORE (Specialized Programs of Research Excellence) grant from the National Institutes of Health. MSK is the only single institution in the country to receive SPORE funding for sarcoma research. The project is led by surgeon-scientist **Samuel Singer**.

Specialized Diagnostic Approaches in Sarcoma

One of the first steps in any cancer diagnosis is preparing tissue taken from a biopsy to look at under a microscope. But unlike with many other cancers, determining a sarcoma tumor's type based on its appearance alone can be difficult, especially for pathologists who don't see them regularly. "Sometimes different kinds of sarcoma resemble each other," says MSK Surgical Pathology Service Chief **Meera Hameed**, who specializes in sarcoma. "Other times, there may be cells that look very different from each other found within the same sarcoma tumor."

For this reason, Dr. Hameed explains, looking at tumors under the microscope often doesn't provide enough information alone to make a diagnosis. This makes molecular pathology a vitally important piece of the sarcoma puzzle. Some of these tests are done using immunohistochemistry, a type of analysis that enables a pathologist to identify the presence of certain proteins in a tumor sample. But genomic sequencing and other types of molecular analysis are often the best way to pinpoint a sarcoma's unique characteristics.

MSK geneticist and pathologist **Marc Ladanyi**, Chief of the Molecular Diagnostics Service, is an internationally recognized leader in this field. Dr. Ladanyi has developed molecular diagnostic tests not just for sarcoma but for many other types of cancer as well. In addition, in his research lab, he has identified many of the genetic abnormalities that are known to drive sarcoma growth.

Diverse Class of Tumors with Varied Underlying Causes

More than one-third of all sarcomas are caused by a type of genetic abnormality called a fusion gene. These genes are created when a piece of a chromosome breaks off and is transferred to an unrelated gene, which causes the formation of a protein that drives uncontrolled cell growth.

MSK doctors use a special technology to detect gene fusions. Dr. Ladanyi and colleagues helped develop and refine that test, and the work is now setting the stage for diagnosis of cancers driven by fusion genes worldwide.

"Our ultimate goal is to be able to eventually find the Achilles' heel for every type of sarcoma."





Marc Ladanyi
physician-scientist

The other two-thirds of sarcomas are triggered by individual gene mutations, which can be detected with **MSK-IMPACT™**. This test, developed by the Molecular Diagnostics Service, looks for mutations in more than 400 genes that are known to play a role in cancer.

Applying Research to Sarcoma Patient Care

One of the essential contributors to MSK's expertise in sarcoma is a database of more than 10,000 people treated for sarcoma. It was started more than 20 years ago by pioneering sarcoma surgeon **Murray Brennan**, who led MSK's Department of Surgery for many years.

The database includes clinical records and medical history, images of pathology samples, and — more recently — details about the genetic and other molecular changes driving tumor growth. “Gaining a better understanding of what drives these tumors can help us come up with new ways to target them,” Dr. Ladanyi says. “Our ultimate goal is to be able to eventually find the Achilles' heel for every type of sarcoma.”

“This is an exciting time to be doing sarcoma research,” Dr. Antonescu concludes. “It's moving so quickly. We are able to take discoveries that we make about gene fusions and other mutations and immediately translate them into new diagnostic tests that can guide treatments. That's the beauty of working at MSK.”

Comments

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Ellen Mirtz

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Over 22 yrs I have had six surgeries and about five different chemos. My first tumor was buried inside my fibroid and found when I had a hysterectomy because it was growing. No one suspected a Leiomyosarcoma. It was a nightmare. The second one came back after about 7-8 years and was the size of a football. The third came back several years later and I had my surgery at MSK with Dr. Yoon. He had to remove