Soft Tissues, Tough Battles

Learn How to Support Patients with Rhabdomyosarcoma and Their Families

By Brian Justice

Rhabdomyosarcoma (RMS) is an extremely rare type of cancer. Up to 500 new cases are reported every year in the United States,[1] making RMS the most common soft-tissue cancer in children,[2] accounting for approximately 3% of all childhood cancers.[1]

Most cases occur in children under the age of 10, and boys are at slightly higher risk than girls. While RMS is primarily a childhood cancer, it can also affect teenagers and adults. In those age groups, survival rates are considerably lower.[1]

"Rhabdomyosarcoma, or sarcomas in general, are cancers of the soft and connective tissues in the body," explains Michael Ferguson, MD, associate professor of pediatrics and medical director of the Solid Tumor Program at Riley Hospital for Children in Indianapolis, Indiana. "These are specific muscle-related tumors. There are muscles, such as in the arms and legs, but also smooth muscles in the bowels and gut associated with blood vessels and other parts of the body. So, this is a cancer of a muscle origin."

RMS presents in two primary forms[2]:

- **Embryonal RMS**, which commonly occurs in the head and neck area, genitals, or urinary tract, often affects children under the age of 6. Embryonal RMS generally responds well to treatment.
- **Alveolar RMS** is more prevalent in teenagers and frequently found in the arms, legs, chest, or abdomen. Alveolar RMS is harder to treat and requires more intensive therapy.

Causes

Researchers are still trying to determine what causes RMS. The simplest explanation is that while the human body includes tumor suppressor genes that stop cells from growing too fast and turning into tumors, RMS may hinder those suppressors, causing cells to grow and form tumors.[3] People with certain inherited gene mutations, like Noonan syndrome, which affects one out of every 1,000 to 2,500 people,[4] and neurofibromatosis type-1, which occurs in one out of every 3,000 to 4,000 people,[5] seem somewhat more likely to develop RMS.

A gene called p53 stops cells from dividing too much. However, cells continue dividing and create a tumor if a mutation occurs. This inherited gene is specific to embryonal RMS, notes Dr. Ferguson. Meanwhile, Dr. Ferguson adds, the PAX/FOX01 fusion gene (i.e., when portions of an individual's DNA switch between chromosomes), which leads to uncontrolled cell division and tumor formation,[3] is specific to alveolar RMS.

Symptoms

RMS symptoms vary depending on the tumor's location. In the urinary tract, RMS may cause difficulties with urination, blood in the urine, or a vaginal, testicular, or abdominal mass. In the head or neck, it causes problems in the sinuses or nasal

passages, headaches, bleeding or growth in the ear canal, or a painful bulging eye if behind the eye socket. In an extremity, it may present as a mass that may or may not be painful. If the cancer is within the trunk, individuals may experience abdominal pain, constipation, and vomiting.[3]

"Cancers of the muscles, or that affect skeletal muscle, are very painful," explains Danielle Leonardo, MD, a medical oncology consultant at the Medical City Ortigas in Pasig City, Philippines. "These patients also tend to have disfigurements because of large tumors, and that can be devastating."

Treatment

Various factors determine the prognosis and treatment strategies, such as the type of RMS, the tumor's location and size, surgical outcomes, and whether the cancer has metastasized. Children between the ages of 1 and 9 generally have a more favorable outlook than infants, older children, or adults.[1]

The primary methods for treating RMS are chemotherapy, surgery, and radiotherapy, often in combination. Chemotherapy is used to shrink the tumor before surgery or remove remaining cancer cells.[3] The type and stage of the disease determine the specific drugs administered and the duration of treatment.[2] If surgery is unfeasible, a combination of chemotherapy and radiotherapy is used, with high-energy rays directed to the area of origin to destroy cancer cells while minimizing harm to the surrounding healthy ones.[2]

Up to 90% of children with low-risk RMS survive five years or more after diagnosis. For those with intermediate risk, that drops to about 50% to 70%. Among children with high risk, only up to 30% survive five years or longer. These statistics emphasize the need for early diagnosis and treatment.[3]

"It's not always fatal—especially in younger kids, because they tend to get the embryonal subtype, and we are able to achieve a cure around 70% of the time," explains Dr. Ferguson. "And when the tumor occurs in other locations, like behind the eye, we see cure rates of almost 90%."

Notably, adults with RMS have a five-year survival rate of 47%, according to Dr. Ferguson. Factors contributing to this disparity include the cancer's rapid growth and the fact that it can manifest in challenging-to-treat anatomical locations.[3]

Residual Effects of Childhood Cancer

The effects of childhood cancer are long-lasting and extend beyond full recovery. In a recent review[7] of 73 studies, researchers discovered that childhood cancer survivors remain at risk for other long-term health challenges. The vast majority (95%) develop issues related to their cancer or treatment by age 45, including hormone issues, reproductive health challenges, musculoskeletal problems, and cognitive impairment.

Approximately one-third of childhood cancer survivors experience severe or potentially life-threatening chronic issues, with endocrine disorders, neoplasms, and cardiovascular disease being the most common. Radiation fields on the chest, brain, neck, abdomen, or pelvis pose other specific risks, possibly leading to other cancers, especially after treatment with higher radiation doses. Mental health is also impacted, with depression rates ranging from 2% to 41%, far exceeding the national average of 10% of adults who report experiencing depression.

Given these ongoing risks, researchers advocate for survivors to receive lifelong care focused on awareness of potential health issues—physical and mental. Health care providers are encouraged to inform patients about the potential long-term effects of treatment, emphasizing the need for regular physicals and an elevated level of preventive care for adult survivors of childhood cancer.

Parents and Families

Any serious childhood illness profoundly affects families, and cancer presents unique challenges.

Research shows that parents of children with cancer face multiple unmet needs. The chronic nature of childhood cancer necessitates continuous care that impacts the family's personal, social, and professional lives and makes them more vulnerable to emotional and physical stresses. Recognizing and addressing their needs helps them focus on their child's care.[6]

Families require clear, understandable information to fully participate in health care decisions. Robust collaboration between health care providers and parents is vital to providing the support and assistance they need to deal with their family's new levels of responsibility and stress.[6]

"The best way to interact with children and their families is to give them the utmost compassion," says Jessica Blessinger, CMA (AAMA), clinical preceptor lead for Hancock Health in Greenfield, Indiana. "Remember that there are no questions that should not be asked." ✦

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