WHAT WE DO

Identify and fund new, medically viable treatments for rare childhood brain tumors.

Provide message forums for families of children with cancer to talk with others fighting the same battle and to learn more about these rare types of tumors. Our forums promote open dialogue between parents, doctors, and researchers worldwide in an effort to find a cure.

Develop and distribute educational material to children's hospitals nationwide to be provided to parents of newly diagnosed children.

Compile and store information supplied to us voluntarily by parents of children battling rare cancers pertaining to their child's diagnosis, treatment protocols, results, and other relevant data. Our goal is to isolate trends and patterns in the data in hopes of helping other parents and doctors make the best choices for their children and patients.

HOW WE ARE DIFFERENT (WHY HELP US?)

Efficiency – More of your money goes toward research and education as we pay no salaries, rent, utilities, or lobbyists. Our services and equipment are donated, so expenses are few.

Focus – Our programs focus on the rare types of pediatric tumors that go largely overlooked by the other organizations.

Motivation – We are fresh and energetic! You can take as large or small of a role as you'd like with us. We value your input, and we need YOUR help to make a difference!

CHILDREN'S BRAIN TUMOR RESEARCH FOUNDATION

Children's Brain Tumor Research Foundation is an IRS 501(c)(3) not-for-profit organization dedicated to promoting and funding research, furthering education, and building awareness about rare, malignant, childhood brain tumors. CBTRF consists of and reaches out to parents of children diagnosed with these rare cancers, doctors, researcher, caregivers, and concerned friends.

We specifically aim to identify, promote, and support research targeted at delivering therapy directly to the affected areas rather than to the entire body, minimizing collateral damage to healthy tissue and organs, and improving the quality of life for survivors.

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CHILDREN'S BRAIN TUMOR RESEARCH FOUNDATION

Rhabdoid Cancer

Frequently Asked Questions

What is a Rhabdoid Tumor?

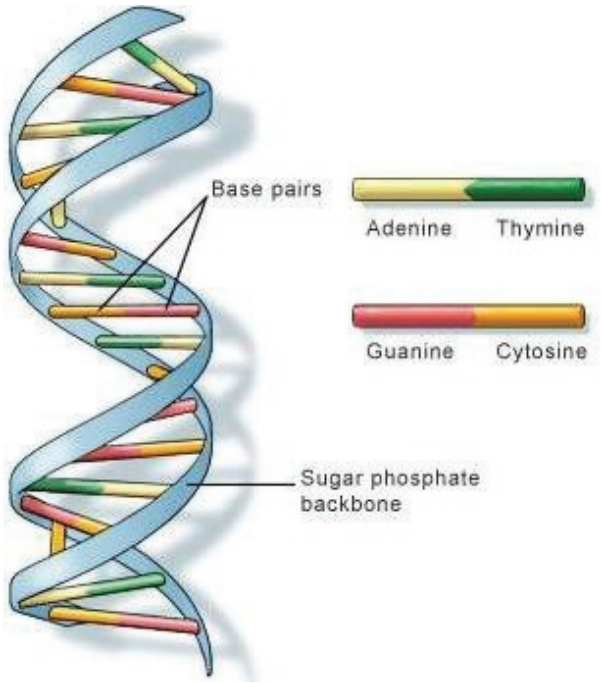
A rhabdoid tumor is a rare, malignant cancer, usually diagnosed in the early childhood years. Rhabdoid tumors may be found in the brain or spinal cord, where they are referred to as atypical teratoid rhabdoid tumors (AT/RT), or in any of the soft tissue in the body, including the kidneys, lungs, liver, skin, pelvic cavity, or stomach. Rhabdoid tumors may be referred to as malignant rhabdoid tumor (MRT), rhabdoid tumor of the kidney (RTK), or by various other acronyms.

What Causes Rhabdoid Tumors?

Rhabdoid cancer is thought to occur, at least in part, due to mutation or deletion of the INI1 / SMARCB1 gene on chromosome 22 (22q11.2).

A chromosome is comprised of two strands of DNA, each made up of four types of miniscule building blocks (adenine, thymine, guanine, and cytosine – or A, T, G, and C) known as nucleotides. These two long strands fit together like two sides of zipper, but there's a rule involved: adenine only pairs with thymine, and guanine only pairs with cytosine. So each rung in the ladder is a pair of nucleotides (base pair), and each pair is either an A stuck to a T or a G stuck to a C.

You've got six billion pairs of nucleotides in each of your cells, and amongst them are roughly 30,000 genes, or distinct stretches of DNA that determine something about who you are (your height, eye color, hair color, etc). When one or more nucleotides are altered, and cannot pair correctly, it is called a mutation. A large piece of DNA may also be gained (duplication) or lost (deletion) which can also change one or more genes on the chromosome. A damaged gene may not be able to perform its intended function, and in some cases can lead to disease, including cancer. The INI1/ SMARCB1 gene is thought to act as a tumor suppressor, so when it is damaged, there is no longer a gene to prevent rhabdoid cancer from occurring.



So Rhabdoid is Genetic?

Rhabdoid cancer is genetic, meaning that it occurs due to DNA damage, but it may or may not have been inherited from a biological parent. In fact, in the majority of cases, the DNA damage was not passed genetically from a parent, rather it occurred during the child's development. The only way to determine this is by genetic testing.

How Do I Have Testing Performed?

Speak with your child's oncologist and ask them to get in touch with Dr. Jaclyn Biegel at The Children's Hospital of Philadelphia, the foremost rhabdoid cancer genetics expert in the United States. Dr. Biegel can be reached by email at biegel@mail.med.upenn.edu .

What Are the Possible Testing Outcomes?

First, you will find out if your child's mutation or deletion is constitutional, meaning that it occurs in every cell in their body, or if it is sporadic, meaning that it only occurs in the cells within the tumor. It is theorized that a finding of sporadic damage lends to a better prognosis, but there is simply not yet enough data to support this hypothesis. If the damage is found to be constitutional, the researchers may test the parents' blood samples to determine if the genetic condition was inherited or happened during the child's prenatal development (*de novo*).

Are My Other Children at Risk?

Genetic testing is <u>always</u> recommended with rhabdoid cancer, but is especially recommended if there are other current or planned siblings. If the gene mutation was inherited from a biological parent, all current and future siblings sharing the same biological parents have a fifty percent (50%) chance of also having the genetic disorder. If the mutation was found to be *de novo*, there is still a slight chance (around 1%) that other children may be at risk, due to mosaicism, which means that only the parent's sex cells (sperm or egg) carry the mutation, making it undetectable by standard blood tests. Your geneticist can explain these possibilities further.

Where Can I Find More Information?

You may visit the Children's Brain Tumor Research Foundation website at www.cbtrf.org for additional information. CBTRF maintains the largest list of current children with rhabdoid cancer in the world, and helpful resources for parents, including information regarding treatments (both standard and trial), financial assistance, grief counseling, and other pertinent resources. In addition, CBTRF operates the largest online community (message forum) for discussion of rhabdoid cancer in the world, with over 200 parents of children with rhabdoid cancers signed up as members to offer information and support to families of newly diagnosed children.