



UNDERSTANDING AND COPING WITH WISKOTT-ALDRICH SYNDROME (WAS)

By Deena Marie Biengardo

Patients with this rare disease and their families now have renewed hope of locating information and a support network to help them cope and survive.

Wiskott-Aldrich syndrome (WAS) is a rare, devastating disease that occurs in only four out of every one million live male births — which means only approximately 500 young men in the United States are afflicted with the disease. Because it is so rare, there is very little awareness, and diagnoses and information are hard to come by. But, with the creation of a new website, WAS patients now have a place to turn to for information and support.



What Is WAS?

WAS is an immune deficiency disorder caused when not enough immunoglobulin is produced by the body. WAS patients suffer from low numbers of blood platelets that are small and do not function properly. The disorder is associated with a defective gene on the X (female) chromosome called the WAS gene. Females tend to be carriers of the gene, while males with the gene develop symptoms.

While WAS is generally symptomatic in children, individuals of all ethnic backgrounds in all geographic regions can be affected by it.

What Are the Symptoms of WAS?

Symptoms experienced by WAS patients include recurrent serious infections such as pneumonia, meningitis and sepsis; bloody diarrhea; prolonged bleeding; and unusual



Sumathi Iyengar, a pediatrician who developed a website for WAS patients and their families, has a son who was diagnosed with WAS at 1 year of age.

bruising. Additionally, patients have a genetic tendency to develop common allergic diseases such as eczema (itchy red or purple spots) caused by minor hemorrhage, and they are at significantly higher risk for developing autoimmune diseases and malignancies, such as lymphoma and leukemia.

How Is WAS Diagnosed?

WAS is diagnosed based on a blood film and low immunoglobulin levels. Often, leukemia is first suspected because of low platelets and serious, recurrent infections. Decreased levels of the WAS protein and confirmation of a causative mutation provide the most definitive diagnosis. However, there are different levels of mutation that are not only difficult to diagnose but also complicated to treat.

At the time of diagnosis, the patient may not demonstrate all of the typical WAS symptoms. The disease can present itself at varied levels of severity. A milder form of WAS is XLT (X-linked thrombocytopenia), where the patient presents mostly with a low platelet count. Another milder form of WAS is XLN (X-linked neutropenia), where the patient presents mostly with a low neutrophil count. Patients with XLT are often considered ITP (immune thrombocytopenic purpura), which means they have a low platelet count for no known cause. According

to Dr. Hans Ochs of Seattle Children's Research Institute, a physician with experience will diagnose a classic case of WAS within the first few months of life. Inexperienced physicians, on the other hand, often

WAS Treatment and Transplant Centers

Patients with WAS should be able to find assistance at most large Children's Hospitals. According to Dr. Hans Ochs, the best places to seek treatment are where stem cell transplants can be performed. In the West, transplants can be performed in Vancouver, B.C., Seattle, Portland, San Francisco and at the University of California, Los Angeles, and the Children's Hospital in Los Angeles. In the middle states, transplants can be performed in Denver, Dallas, Houston, Chicago (Northwestern University), St. Louis, Cincinnati, Minneapolis and New Orleans. In the East, they can be performed in Boston, New York, Washington, D.C., Duke University in North Carolina, Emory University in Atlanta, Ga., and Birmingham, Ala. In Canada, Sick Children centers in Toronto, Ont., can provide help to WAS patients.

WAS is diagnosed based on a blood film and low immunoglobulin levels.

make incorrect diagnoses. Therefore, many patients are not diagnosed as having a mutation of the Wiskott-Aldrich protein for years, and some patients are never diagnosed.

How Is WAS Treated?

Because WAS is primarily a disorder of the blood-forming tissues, a hematopoietic (blood-forming) stem cell transplant done through cord blood or a bone marrow transplant offers the only hope for a cure. So far, with stem cell transplantation, the life expectancy of a WAS patient is normal. Transplant is not always recommended, however. Symptomatic treatment can be provided to patients with milder forms of WAS, and with this, their average life expectancy is 15 to 30 years. Without any treatment, a patient's life expectancy is very short.

Symptomatic treatments for patients suffering from WAS are limited. Often, children wear a helmet to protect them in case they fall and bump their heads. This is meant to protect their brain from an injury that could cause a bleed. For severely low platelet counts and serious bleeds, patients may require platelet transfusions. A controversial treatment is splenectomy, or removal of the spleen. Splenectomy can raise platelet counts, but it is reserved for patients who are not transplant candidates, and it is currently not recommended unless the patient is having serious bleeding problems. Patients who undergo splenectomy have to be on prophylactic antibiotics every day afterward to prevent sepsis and other life-threatening infections. Anemic patients may even require iron supplementation or a blood transfusion. Intravenous immunoglobulin (IVIG), given to boost the immune system, is another option for patients with frequent infections.

Information and Support for WAS

Dealing with a diagnosis of WAS is devastating for families. It is especially perplexing to attempt to navigate through the maze of doctors, medications and new terminologies. So, a new website designed to bring the WAS community — patients, doctors and families — together may help to improve understanding, provide support and spur significant improvements in the care and cure of this disease.

Developed by Sumathi Iyengar, a pediatrician whose son was diagnosed with WAS at 1 year of age, the site includes a number of sections. The "find a doctor" feature allows patients to find local doctors who are capable of treating WAS, as well as other experts in the field. Doctors' contact information is listed, and links to their websites are provided. A "resources" page has links to medical literature, immunodeficiency foundations, and financial and transplant resources. And, a "how to cope" section provides information for families to better deal with the emotional aspects of dealing with this disease. Last, a link to a support forum where families can connect, exchange experiences and support one another is provided.

Raising Awareness of WAS

Because WAS affects a very small number of people, it has received little attention. Raising awareness about WAS among physicians, patients and their families will help to pave the way for early diagnosis, more research in the field and more funding. ■

WAS Resources

To access the Wiskott-Aldrich website, visit sites.google.com/site/athreyi
(Note: The WAS site is under construction and will be accessible in fall 2009.)

To access the WAS support forum, visit www.primaryimmune.org/forum/forum_intro.htm

DEENA MARIE BIENGARDO is a writer who lives in New York and is currently pursuing a degree in cultural studies.