The Basics of Atypical HUS

Atypical Hemolytic Uremic Syndrome (Atypical HUS) is a very rare and serious disease that occurs when small blood clots, called thrombotic microangiopathy or TMA, form in tiny blood vessels throughout the body. These clots damage the kidneys and other vital organs. Atypical HUS can affect adults and children and often present at a very young age.

Each case of this rare disease is quite different making it difficult to diagnose and even harder to treat. Initial symptoms can include extreme fatigue, puffiness, vomiting, paleness, fever, and often diarrhea. These symptoms are chronic in some patients and less frequent in others. Clinical complications of the disease, including kidney damage, can occur even if symptoms are not present.

A diagnosis of Atypical HUS can be devastating for a family, often leading to months of inpatient hospital care, long-term dialysis, and a drastic change of lifestyle.

THERE ARE TWO TYPES OF HUS, TYPICAL AND ATYPICAL. WHAT'S THE DIFFERENCE?

1. Typical HUS can be triggered by E coli bacteria or other bacteria or food borne pathogens. In typical HUS, most cases will not occur again after the initial onset (typically lasting 4-6 weeks). Cases can see long-term kidney damage and issues with high blood pressure.

2. Atypical HUS Atypical HUS can be triggered for a variety of reasons such as genetic mutation, pneumonia, or gastrointestinal illness—as well as unknown reasons. The biggest difference between types is that Atypical HUS is unpredictable, with frequent clinical complications caused by TMA.

WHAT HAPPENS TO THE BODY DURING THE ATTACK?

Atypical Hemloytic Uremic Syndrome starts with uncontrolled activation of complement, a series of proteins that is part of the body's immune system. This produces small blood clots that cause red blood cells to shatter and travel to the kidneys where tiny microscopic clots also are formed. These complications cause severe anemia and kidney failure. When the kidneys fail to work, the body fails to rid itself of toxins, the urine output declines, and the amount of protein in the urine will increase. This process can permanently damage the kidneys. Poorly functioning kidneys also lead to high blood pressure and swelling which can stress the heart and lungs. Blood clots also may develop somewhere else in the body, with some aHUS patients also experiencing neural events such as seizure activity, or heart problems. This process is very dangerous and can be deadly if not treated by an experienced facility.

WHO CAN TREAT THIS DISEASE?

Having knowledgeable specialists is critical for successful treatment. Typically, a nephrologist (kidney specialist) and oncologist/hematologist are the primary doctors. Other specialist such as cardiologists, immunologists, and intensive care specialists are also involved. Due to the rarity of this disease, many doctors have never had experience treating an aHUS case. Often families have to commute long distances just to receive experienced care.

WHAT ARE THE TREATMENT OPTIONS?

Currently, there is no cure for this disease. Atypical HUS does not have a standard 'treatment protocol' as is common in most other diseases. These are some of the most common interventions:

BLOOD TRANSFUSIONS: Blood Transfusions are almost always the first line of management. Typically, aHUS patients are admitted to the hospital with anemia. A patient may receive packed red blood cells, whole blood, and/or platelets in order to stabilize their current situation. These products help to return the blood to a more normal level for a brief time but do not treat the disease. During a severe clinical manifestation of TMA, an Atypical HUS patient may depend on 100's of generous blood donors to stay alive.

PLASMA THERAPIES: Plasma therapies are the most traditional way to treat patients with aHUS. Donated plasma is transfused into an aHUS patient, called Plasma Infusion. In difficult cases, plasmaphersis, a process where the body's plasma is removed and replaced with donor plasma, may be used instead.

DIALYSIS: Atypical HUS can cause a patient's kidneys to permanently or temporarily to stop functioning. If the patient's kidneys cannot perform properly, patients must receive dialysis. Dialysis can be used as an interim replacement for kidneys. Though it is necessary to sustain life, dialysis is can be dangerous and risky for young patients.

Due to the ongoing nature of the disease process and subsequent damage to the kidneys, nearly all aHUS patients are on extensive blood pressure medications as well as other medications that help regulate the body's ability to manufacture red blood cells and ability to regulate electrolytes.

TRANSPLANTS: Many Atypical HUS patients have permanent kidney failure. Transplants are not a viable option for these patients until a treatment or cure can be found that puts the disease in remission.

GENETIC TESTING

In America, the primary genetic testing facility for aHUS patients is at the University of Iowa. There are only a few other labs across the globe that can perform this testing. Approximately, 50% of patients are found to have specific genetic mutations. Understanding the genetic components to the disease helps researchers determine future potential treatment for the disease.

The Foundation for Children with Atypical HUS

SOLIRIS: A NEW HOPE FOR TREATMENT

A recent development in aHUS treatment is a set of clinical trials being conducted by Alexion Pharmaceuticals, makers of eculizumab (Soliris®). Eculizumab is an intravenous (IV) drug that is FDA-approved to prevent hemolysis (red blood cell destruction) in adult patients with the rare disease PNH. This drug continues to be studied in clinical trials for aHUS patients and is not currently FDA approved for Atypical Hemolytic Uremic Syndrome. Interim results from these trials were presented last year and were encouraging. There is a handful of aHUS patients currently being treated very successfully with this medicine. Some aHUS patients on eculizumab are seeing no evidence of clinical manifestations of aHUS long-term for the first time, as well as increased quality of life. While currently an "off label" use and NOT yet FDA approved, eculizumab use is a new option for doctors to discuss on a case-by-case basis. Due to the very expensive cost associated with this drug, approval is often very difficult.

Because the new treatment option, eculizumab, may change the course of the disease, this now gives hope that a transplant may now be a possibility for the first time.

HOW YOU CAN HELP

Although there is no cure for Atypical HUS, there is new research, treatments, and information that has come available in just the past few years. This has given families new hope that a cure is very close—possibly within the next several years. Your contribution to this Foundation provides funding for ongoing research as well as resources and training for families dealing with this disease. The families involved with The Foundation for Children with Atypical HUS thank you for your support.

Learn more about Atypical HUS families, researchers, and professionals who are working to stop this disease at: www.Atypicalhus.org
More info also at: www.Atypicalhus50megs.com

Bill Biermann, Director · One Campbell Plaza, Suite 2B, St. Louis, MO 63139