

The First Wiskott - Aldrich syndrome Families and Investigators Conference
July 30, 2010 - August 1, 2010
Hilton Chicago Indian Lakes Resort and Conference Center, Bloomingdale, Illinois

Summary

The First Wiskott Aldrich Syndrome family conference took place from the 30th July to the 1st August at the Hilton Chicago Indian Lakes Resort and Conference Centre in Bloomingdale, Illinois

Wiskott - Aldrich syndrome (WAS) is a rare primary immune deficiency disease affecting 4 per million live male births. Patients with WAS are prone to significant bleeding due to low platelets and are at an increased risk of serious infections because they are immunodeficient. Most of these patients have mild to severe eczema. They are also at a higher risk of developing autoimmune disorders and malignancies such as lymphoma. WAS varies in the severity of its symptoms, with a little more than half of the patients having Classic WAS with severe manifestations and the other half have a milder form with varying degrees of immunodeficiency. This makes the management of WAS variable, ranging from bone marrow transplants, splenectomy, IVIG infusions and antibiotics to just watchful waiting.

In total, 160 people from all around the world attended the conference, including 50 children from 30 families. The conference included presentations from leading experts from the US and Europe. The conference began with a welcome dinner that included an ice breaker to help attendees relax and get to know each other. A number of families also spoke about how their life and family were affected by WAS.

Saturday morning, the presentations began with an overview of WAS, from its discovery to current challenges in management. They also included the spectrum of the clinical presentation of WAS, and the frequency with which it is misdiagnosed. The WAS gene and its role in the immune and blood systems, was also explained in detail. This highlighted for families the complexity of the disease, and the difficulties with its management, such as the variation in clinical condition of people with the same genetic mutations.

The afternoon sessions covered the complexities of Hematopoietic Stem Cell Transplants which at present are the only cure for WAS. There was an intense discussion between the panel of experts and the audience about quality of life issues and the definition of "cured" after having a transplant.

Two experimental medications - Interleukin 2 therapy to help preserve immune function and eltrombopag (promacta) to boost platelets were also presented. These medications may improve quality of life for patients with WAS.

Saturday afternoon finished with results of the gene therapy studies which have been carried out in Germany and Italy, and are due to get underway in the UK. Results from the retroviral gene therapy study in Germany have been very positive to date, with almost all the candidates doing well. Gene therapy is a very exciting development for Classic WAS patients. It appears to have less of the side effects associated with a HSCT.

Sunday focused on social, behavioural and quality of life issues. There was also an open discussion on all aspects of WAS. It was clear that there are many controversies in terms of management of the XLT patients, while management decisions are more straightforward for a classic WAS patient.