

The WAS-related disorders are a spectrum of conditions affecting the immune system that are caused by mutations in the WAS gene. These disorders include Wiskott-Aldrich syndrome, X-linked thrombocytopenia and X-linked congenital neutropenia. The WAS gene abnormality results in a deficiency in the WASP protein that leads to a low platelet count (thrombocytopenia). WAS-related disorders usually present in infancy and are characterized by bloody diarrhea, recurrent infections, scaling, itchy, skin rashes (eczema), and the appearance of small purple spots on the skin (petechia). The development of *Pneumocystis carinii* pneumonia (PCP) and intracranial bleeding are possible early, life-threatening complications. Later potential complications include destruction of red blood cells (hemolytic anemia), arthritis, vasculitis and kidney and liver damage. Affected individuals have an increased risk of developing lymphomas, especially after exposure to Epstein-Barr virus. WAS-related disorders are extremely variable, even in individuals in the same family. The estimated prevalence of WAS-related disorders is 3/1,000,000 males. This condition has been described in many ethnic groups and in many countries. A WAS-related disorder is suspected based on clinical features and laboratory testing. Testing shows a low platelet count and small platelet size, and sometimes shows abnormal levels of serum immunoglobulins: low IgM, elevated IgA and IgE, decreased absolute numbers of CD8+ T cells and decreased function of natural killer cells. Decreased or absent WASP protein in blood cells strengthens the diagnosis. Molecular genetic testing for the WAS gene is available to confirm the diagnosis.