

Ataxia telangiectasia (AT) is a complex genetic neurodegenerative disorder that may become apparent during infancy or early childhood. The disorder is characterized by progressively impaired coordination of voluntary movements (ataxia), the development of reddish lesions of the skin and mucous membranes due to permanent widening of groups of blood vessels (telangiectasia), and impaired functioning of the immune system (i.e., cellular and humoral immunodeficiency), resulting in increased susceptibility to upper and lower respiratory infections (sinopulmonary infections). Individuals with AT also have an increased risk of developing certain malignancies, particularly of the lymphatic system (lymphomas), the blood-forming organs (e.g., leukemia), and the brain. Ataxia telangiectasia usually begins during infancy (between one and three years of age) and often affects more than one child in a family. Males and females may be affected in equal numbers. In the United States, the prevalence is approximately one in 40,000-100,000 live births.