

Ataxia with vitamin E deficiency (AVED) is a rare inherited neurodegenerative disorder characterized by impaired ability to coordinate voluntary movements (ataxia) and disease of the peripheral nervous system (peripheral neuropathy). AVED is a progressive disorder that can affect many different systems of the body (multisystem disorder). Specific symptoms vary from case to case. In addition to neurological symptoms, affected individuals may experience eye abnormalities, disorders affecting the heart muscles (cardiomyopathy), and abnormal curvature of the spine (scoliosis). AVED is extremely similar to a more common disorder known as Friedreich's ataxia. AVED is inherited as an autosomal recessive trait. AVED affects males and females in equal numbers. The disorder is estimated to occur in fewer than 1 in 1 million people. In Tunisia, the disorder is estimated to occur in 1 in 100,000 people. The onset of AVED may occur during childhood or adulthood with cases reported in children as young as two and adults as old as 52. The disorder was first described in the medical literature in 1981. A diagnosis of AVED is made based upon a thorough clinical evaluation, a detailed patient history and a variety of tests and characteristic findings (e.g. low levels of vitamin E with normal levels of lipoproteins and lipids and no evidence of fat malabsorption).