

Tangier disease is a rare inherited disorder characterized by significantly reduced levels of high-density lipoproteins (HDL) in the blood. HDL-cholesterol (HDL-C) is often referred to as the “good cholesterol” as it can facilitate the removal of cholesterol out of the walls of arteries, particularly the coronary (heart) arteries. Classic features of Tangier disease include fatty accumulations that present as enlarged and yellow- or orange-colored tonsils, or enlarged liver (hepatomegaly), spleen (splenomegaly), or lymph nodes. Tangier disease may also be associated with an increased risk of cardiovascular disease, moderate elevation in triglycerides (hypertriglyceridemia), nerve disturbances (neuropathy), and rarely an opaqueness in the covering of the eye (corneal clouding). This disorder was originally named after the location in which it was first discovered - Tangier Island in the Chesapeake Bay. Later, the disease was further characterized as more individuals were found to have the disease in other areas of the United States and around the globe. Symptoms of Tangier disease are variable and depend on which organs are involved and the severity of those manifestations. Tangier disease is most often characterized by enlarged orange- or yellow-colored tonsils. This discoloration is due to fatty deposits accumulating in the tonsils. Fatty deposits can also form in other organs causing enlargement of the throat, liver, spleen, or lymph nodes. Fat accumulations in nerves can cause disturbances and loss-of-sensation called peripheral neuropathy. Discoloration may also occur in the digestive system, particularly the rectum and large intestine. Cardiovascular disease has been reported in adults with Tangier disease. In rare cases, a clouding of the cornea of the eye can occur, but is generally mild and does not cause vision impairment. Tangier disease is a rare disorder with only approximately 100 cases diagnosed worldwide. Tangier disease is thought to be present at birth, but the age of diagnosis can be highly variable (from infancy through 7th decade) due to the nature of symptoms.