Condition: Tangier disease

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in *ABC1* gene encoding ATP-binding cassette-1 gene.

Frequency:
Rare

Clinical features:
Tangier disease presents with enlarged, orange tonsils, hepatosplenomegaly, enlarged lymph nodes, hypocholesterolemia, and low plasma high density lipoproteins. There is early onset of coronary artery disease. The pathophysiology involves inability to transport cholesterol to the cell surface required to synthesize HDL.

Management:
Supportive care.

Genetic counseling:
Found mostly in specific isolated populations; autosomal recessive transmission.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.