

Table S1 Diagnostic criteria

A. Clinical manifestations

1. Cutaneous or tendon xanthomas
2. Premature coronary artery disease (male <45 years, female <55 years)

B. Laboratory testing

1. Serum sitosterol elevated

C. Differential diagnosis exclude familial hypercholesterolemia and cerebrotendinous xanthomatosis

D. Genetic analysis

Pathogenic mutations in *ABCG5* or *ABCG8* gene

Definite: fulfills A-1, B-1, C, and D. Probable: fulfills A-1, B-1, and C. Possible: fulfills A-1, A-2, and B-1.