Condition: Holt-Oram syndrome

Inheritance:
Autosomal dominant.

Genetic etiology:
Mutation in *TBX5* gene, encoding T-box transcription factor.

Frequency:
Approximately 1/100,000.

Clinical features:
Holt-Oram syndrome includes malformations of the upper extremity (abnormal carpal bones, radial ray anomalies, phocomelia, radial anomalies, etc.) and congenital heart malformations (ASD, VSD), and abnormalities of cardiac conduction.

Management:
Surgical treatment of limb and cardiac malformations; monitoring and management of disturbances of cardiac rhythm.

Genetic counseling:
Autosomal dominant transmission with variable expression; genetic testing is available.

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