**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.