**Condition:** Cowden syndrome (PTEN hamartoma syndrome)

**Inheritance:**
Autosomal Dominant

**Genetic etiology:**
The *PTEN* hamartoma syndrome is a group of related disorders, including Cowden syndrome and Bannayan-Riley-Ruvalcaba syndrome due to mutations in the *PTEN* gene (encodes phosphatidylinositol-3,4,5-trisphosphate 3-phosphatase). Most pathogenic mutations involve loss of function.

**Frequency:**
1/200,000 (may be underdiagnosed).

**Clinical features:**
The *PTEN* hamartoma syndromes are a group of closely related hamartoneoplastic disorders. Features of Cowden syndrome include a set of lesions on the skin and mucous membranes, including facial tricholemmomas, acral keratoses, and papillomas; macrocephaly; intestinal polyps; breast, thyroid, and endometrial cancers. Bannayan-Riley-Ruvalcaba syndrome includes macrocephaly, large birth weight; gastrointestinal polyps; lipomas; developmental delay.

**Management:**
Symptomatic treatment and surveillance for development of cancer.

**Genetic counseling:**
Recurrence risk of Cowden or Bannayan-Riley-Ruvalcaba syndrome is according to autosomal dominant inheritance. Cases occur both within families and sporadically. Genetic testing for *PTEN* mutation is available.