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.

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