Cowden syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cowden syndrome. ORPHA:201

Cowden syndrome (CS) is a difficult to recognize, under-diagnosed genodermatosis characterized by the presence of multiple hamartomas in various tissues and an increased risk for malignancies of the breast, thyroid, endometrium, kidney and colorectum. When CS is accompanied by germline PTEN mutations, it belongs to the PTEN hamartoma tumor syndrome (PHTS; see this term) group.