

# Breast Cancer in Cowden Syndrome: Manifestation of a Familial Cancer Syndrome

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Cowden syndrome is a familial cancer predisposition syndrome associated with an increased risk for breast, thyroid, and endometrial cancers and benign manifestations. With early detection and appropriate therapeutic treatment, many of its associated tumors and cancers are treatable. By better understanding mutations such as Cowden syndrome, additional targeted therapies can be developed and delivered.

Healthcare providers in oncology often see families with a variety of sporadic cancers or individuals with multiple cancers at early ages. In such cases, concern about whether genetic factors are implicated is important. Innovations in genetic and genomic technology and increased emphasis on genetic counseling and mapping of familial cancers have begun to improve the identification of familial cancer syndromes, such as Cowden syndrome.

Cowden syndrome is an often overlooked cancer predisposition syndrome associated with an increased risk for breast, thyroid, and endometrial cancers as well as benign manifestations (Gustafson, Zbuk, Scacheri, & Eng, 2007) (see Figure 1). Cowden syndrome is among a spectrum of disorders caused by autosomal dominant germline mutations of the phosphatase and tensin homolog tumor suppressor gene (*PTEN*) (Agrawal & Eng, 2006). *PTEN* is mutated in 85% of Cowden syndrome cases, meaning that other syndromes are allelic with Cowden syndrome or variants of the same gene and cause similar neoplasms and fibromas. The gene mutations are related to the deregulation of cellular growth, proliferation, apoptosis, oncogenesis, and angiogenesis (Agrawal & Eng, 2006). If suspected, Cowden syndrome is diagnosed with DNA and molecular genetic testing or by using an operational set of diagnostic criteria (for more information, visit [www.nccn.org](http://www.nccn.org)).

## Case Study

Mrs. O is 39 years old with stage II, multifocal ductal carcinoma in situ, hormone receptor-positive breast cancer. Her treatment has included mastectomy of the affected breast with prophylactic mastectomy of the second breast, followed by reconstructive surgery. She returned to the clinic to discuss additional evaluation and treatment.

Mrs. O is a college graduate with two children. She is employed part-time as a computer programmer. Her surgical history is significant for two prior procedures to remove lipomas and liver mass (benign). Mrs. O has had recurrent bronchitis and eczema.

Mrs. O's genogram revealed multiple, sporadic cancers in her family (see Figure 2). Her paternal grandfather died of colon cancer, as did many of his siblings. In addition, Mrs. O's mother and maternal great-grandmother had breast cancer, a paternal uncle (deceased) had renal cancer, an uncle and a sibling (whose son has macrocephaly) have thyroid cancer, another sibling (whose son has autism, seizures, macrocephaly and café au-lait spots) has breast cancer and lipomas, a brother has cutaneous lipomas, and two cousins have

lipomas. The live generations represent ages ranging from 60–70 years; the next generation ranges from 30–40 years, and the youngest generation ranges from infancy to 30 years.

Focused physical examination found Mrs. O to be alert with no acute distress. She is 5'2" and weighs 120 lbs, with 120/62 blood pressure and a heart rate of 72 beats per minute. She has multiple simple and filiform papular lesions on the center of her face. Her neck has no thyromegaly or palpable nodules. Mrs. O's surgical incisions are well healed without overgrowth of tissue at incision sites. Her abdomen is soft and nontender.

## Treatment Options

To date, few specific treatment options exist for Cowden syndrome. The main goal is the treatment of the presenting tumor, benign or cancerous, through recognized therapies that target the specific pathologic and clinical features of the primary tumor. With early detection and appropriate therapeutic treatment, many associated tumors and cancers are treatable. However, one must be vigilant and continue surveillance for signs and symptoms of additional growths because at least 40%

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