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.

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 and mapping of familial cancers have begun to improve the identification of familial cancer syndromes, such as Cowden syndrome.

Cowden syndrome is a familial 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 in 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).

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% 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 nodes. Mrs. O's surgical incisions are well healed without overgrowth of tissue at incision sites. Her abdomen is soft and nontender.