Cowden Syndrome: a rare disorder presenting with multiple hamartomas of the head and neck

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ABSTRACT

Background: Cowden syndrome (CS), or multiple hamartoma syndrome, is a rare autosomal dominant hereditary cancer susceptibility disorder first described in 1963. Approximately 80% of patients with CS have a germline mutation of the PTEN tumor suppressor gene, resulting in lesions arising from all three germ cell layers. The disease characteristically affects several different organ systems and is highly variable in presentation. It has an incidence of approximately one in 200,000, with a slight female predominance. This incidence is actually felt to be under-estimated due to the complexity of diagnosis. We report two cases of CS that presented to our ENT clinic.

CASE REPORTS

Case 1
A 48 year old woman was referred to us with a complicated history including a reported left-sided neurological tumor, which had been resected 9 years prior. The tumor had originally presented with a vocal cord paralysis. The patient came back to attention due to concerns that the tumor was returning. She stated she was choking again and felt like something was stuck in her throat. Incidentally, she also reported a history of many lesions in the gastrointestinal tract seen during colonoscopy. Family history was positive for 2 paternal aunts with cancer. On exam, she was found to have numerous cutaneous papules involving the right tragus, external auditory canal as well as throughout the oral cavity, nasopharynx, oropharynx, hypopharynx and larynx, see Figures 1, 2 and 3. Salivary gland biopsy of lesions on the right tragus showed verrucae vulgares. She was ultimately sent for genetic testing which showed the PTEN mutation. Further work-up revealed multiple thyroid nodules as well as a large fibroid uterus. The patient underwent a total thyroidectomy and pathology returned as benign thyroid tissue and adenomatous nodules. Due to the association of CS with endometrial cancer, dilation and curettage was performed and was negative for hyperplasia and malignancy. She underwent uterine fibroid embolization, with future plans for a hysterectomy.

More recently, extensive dermatology using the coblator was performed. Multiple lesions from the nasopharynx, oropharynx and base of tongue, see Figure 4. Pathology of these lesions showed hamartomas coexisting with CS. The patient has been referred to medical oncology for future surveillance. She also obtains regular mammograms due to the diagnosis of CS and the PTEN mutation. Six years earlier, she had undergone a thyroid lobectomy followed by completion thyroidecomy and radioactive iodine treatment for papillary thyroid cancer. In addition, she had a prior history of endometrial adenocarcinoma as well as ductal carcinoma in situ of the breast, and had undergone hysterectomy with bilateral salpingo-oophorectomy and bilateral mastectomies. Her past medical history was also positive for a shunt microadenoma as well as a mild Chari I malformation with herniation of the cerebellum. Additionally, she had numerous skin tags in the axillary and groin regions for which she followed with Dermatology. Previous biopsy of right neck lesions showed benign verrucoid keratosis. Of note, her mother had a history of Cauden's disease.

She presented to the clinic with a tender right parotid mass. Fine needle aspiration of the mass was performed and was suggestive of a metastatic malignancy. However, due to the patient’s history of several different malignancies, an MRI of the neck was ordered to further evaluate the parotid mass. MRI showed a heterogenous mass in the right superficial gland. The patient subsequently underwent a right superficial parotidectomy and pathology showed low-grade mucoepidermoid carcinoma. She was seen at the Head and Neck Multidisciplinary Tumor Board where no further intervention was recommended.

The patient has continued to follow-up with ENT, Gynecology, Endocrinology and Dermatology for cancer surveillance. She undergoes yearly colonoscopies as well as regular breast ultrasounds. More recently a cutaneous lesion of the left shoulder was biopsied and pathology revealed a dermatofibroma, see Figure 5.

Figure 1. Oral cavity papillomas (case 1)
Figure 2. Cutaneous lesions along right nasal ala (case 1).
Figure 3. A and B. Videostroboscopy showing oropharyngeal, hypopharyngeal and laryngeal hamartomas (case 1)
Figure 4, A and B. Postoperative images from videostroboscopy (case 1)

Case 2
A 23 year old woman was referred to ENT with an established diagnosis of CS and the PTEN mutation. Six years earlier, she had undergone a thyroid lobectomy followed by completion thyroidecmy and radioactive iodine treatment for papillary thyroid cancer. In addition, she had a prior history of endometrial adenocarcinoma as well as ductal carcinoma in situ of the breast, and had undergone hysterectomy with bilateral salpingo-oophorectomy and bilateral mastectomies. Her past medical history was also positive for a shunt microadenoma as well as a mild Chari I malformation with herniation of the cerebellum. Additionally, she had numerous skin tags in the axillary and groin regions for which she followed with Dermatology. Previous biopsy of right neck lesions showed benign verrucoid keratosis. Of note, her mother had a history of Cauden's disease.

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