Giant Vascular Cylindroma in a Case of Brooke-Spiegler Syndrome

Dear Editor,

A 68-year-old Japanese woman was referred to our hospital complaining of multiple skin-colored eruptions on the face. The patient stated that her mother, sisters, and children had small nodules similar to those on her face. Physical examination revealed a number of papulonodules on the nasal ala and around the eyes, which were first noticed approximately 50 years earlier [Figure 1a]. In addition, there was a 10 cm sized giant nodule on the occipital region, which was partially blue in color [Figure 1b], as well as multiple 10–13 mm sized nodules on the parietal and left temporal regions. The occipital scalp lesion initially appeared 10 years previously, and gradually increased in size. A magnetic resonance imaging examination revealed that the inside of the tumor was full of liquid components with no adhesion between the tumor and the skull. Biopsy from the face revealed many lobules of basaloid tumor nests containing horn cysts with sebaceous differentiation, and the fibrous stroma closely associated with the epithelial components [Figure 1c]. There were no atypical cells. The occipital nodule was surgically removed, and histological findings revealed a large cyst with a blood-filled vascular space, and a solid basophilic tumor mass around the cyst [Figure 1d]. The basophilic tumor mass was composed of irregularly shaped islands of epithelial cells that fit together like pieces of a jigsaw puzzle [Figure 1e]. There were no atypical cells. The parietal and left temporal nodules were also surgically removed, and were found to share similar histological features. Genetic examination revealed a heterozygous mutation, c. 1681_1682delTT, p.Leu561Serfs*8 in exon 11 of the cylindromatosis tumor suppressor *(CLYD)* gene [Figure 1f]. Genetic testing of other family members was not performed.

The patient was diagnosed as having Brooke-Spiegler syndrome (BSS) based on the clinical and histological examination results, familial history, and genetic analysis. BSS is an autosomal dominant hereditary disease,[1] in which multiple cylindromas and trichoepitheliomas develop concomitantly in the same patient. Mutation of the *CYLD* gene, a tumor suppressor located on chromosome 16, is responsible for BSS, and several mutations have been identified.[2] The *CYLD* gene is also responsible for familial cylindromas and multiple familial trichoepitheliomas.[3] In the present case, the patient's cylindroma over the occipital scalp was as large as 10 cm in diameter. To our knowledge, there has been only one case report of a cylindroma larger than 10 cm, occurring in a young woman after a minor trauma.[4] Histological examination showed predominantly eccrine spiradenoma with vasodilatation, as well as features of cylindroma and follicular epithelioma. Malignant cylindromas tend to be larger than benign ones[5]; however, both the current case and the previously reported case[4] were benign, even though they were larger than 10 cm in size. Our patient had not experienced any trauma to the scalp, and enlargement of the nodule had been gradual. The enlargement of cylindroma in the present case is considered to be attributed to vasodilatation within the tumor, which may be originated from the highly

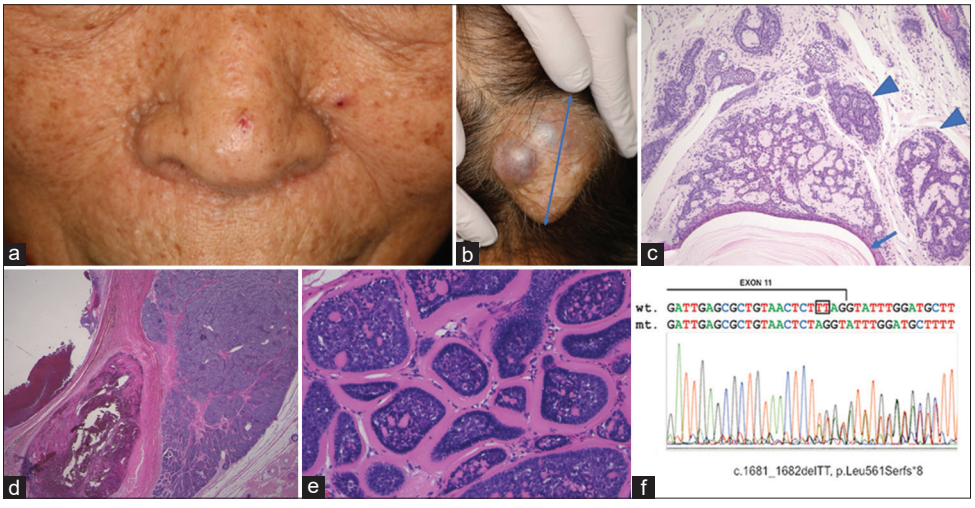

Figure 1: (a) The patient had multiple nodules of normal skin color on the nasal ala. (b) The occipital tumor was about 10 cm in diameter (arrow), and was soft and elastic. (c) Histological findings of the nasal alar nodules showed many lobules of basaloid tumor nests (arrowhead) containing horn cysts (arrow) with sebaceous differentiation, and the fibrous stroma closely associated with the epithelial components (H&E 40x). (d) Histological findings of the occipital nodule revealed a large cyst with a blood-filled vascular space, and a solid basophilic tumor mass around the cyst (Hematoxylin-eosin × 4). (e) The basophilic tumor mass was composed of irregularly shaped islands of epithelial cells that fit together like pieces of a jigsaw puzzle. The hyaline sheath surrounding the tumor islands was thick and pink in color (Hematoxylin-eosin × 20). (f) Mutation analysis of the patient identified a heterozygous mutation c. 1681_1682delTT in exon 11 of the *CYLD* gene

vascular region of the sweat glands, or may result from a degeneration of the tumor stroma during the aging process. BSS is rarely complicated with cylindromas with malignant transformation or basal cell carcinoma, therefore, careful follow-up is necessary.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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