Background: Neurofollicular hamartoma (NH) is a rare benign neoplasm, with mesenchymal follicular differentiation and histological characteristics that resemble a normal hair follicle, or recapitulate part of its embryological development.

Observation: A 51-year-old woman presented with a 10-year history of a normochromic, asymptomatic nodule on the mandibular region, which slowly grew. There was no history of trauma. Physical examination disclosed a 1.0 × 1.5-cm, hard, painless, mobile and normochromic nodule on the anterior right mandibular region that corresponded with the clinical diagnosis of epidermoid cyst. The lesion was excised and histological sections exhibited follicular-sebaceous hyperplasia with infundibulo-cystic dilation containing rudimentary sebaceous structures adhered to the wall. The adnexal lesion involves loose proliferation of fusocellular cells, with undulated nuclei without atypia, within a fibromyxoid stroma, which emulates a neurofibroma-like appearance. Mitosis were not identified. Immunohistochemical reaction with anti-S-100 protein antibody revealed strong nuclear and cytoplasmic positivity in spindle cells, confirming the interpretation of probable neural differentiation. Therefore, based on the histopathological findings of the benign hamartomatous pilosebaceous adnexal lesion in the midst of neural stroma without atypia, it is possible to confirm the diagnosis of neurofollicular hamartoma (NH).

Key message: The pathogenesis of NH is still unknown. Due to the histological characteristics, it was suggested to position NH and trichodiscoma / fibrofolliculoma within the same hamartomatous spectrum. However, NH presents itself with a prominent spindle cell stroma positive for S-100 immunohistochemical reaction. Clinically, it expresses itself as a painless, solitary papule or nodule on the face and not exclusively on the nose; as showed in this case report. In addition, NH has been described as part of the Generalized Basaloid Follicular Hamartoma Syndrome, an autosomal-dominantly inherited disorder with presence of several adnexal tumors.