Medical Case Reports and Short Reviews

CASE REPORT

Sclerotic Fibroma in Patient with Chiari Malformation of Cerebellum, a Possible Cowden Disease

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Abstract

Sclerotic fibroma is an uncommon, benign skin neoplasm that can occur sporadically or in the context of Cowden's disease. Chiari malformation is a congenital abnormality affects the structural relationships between the cerebellum, brainstem, the upper cervical cord, and the bony cranial base. Few patients of Cowden disease have been reported with cerebellar tonsillar herniation. We report a 46-year-old female who presented with both sclerotic fibroma and Chiari malformation.

Introduction

Sclerotic Fibroma (SF) is a well-defined dermal hypocellular lesion composed predominantly of sclerotic thick collagen bundles. It is an uncommon, benign skin neoplasm that can occur sporadically in otherwise healthy individuals or as a solitary or multiple, discrete skin nodule in the context of Cowden's disease [1]. Chiari malformation is a congenital hindbrain abnormality affects the structural relationships between the cerebellum, brainstem, the upper cervical cord, and the bony cranial base [2].

Herein we present a case of solitary sclerotic fibroma associated with Chiari malformation. To date, this is the first case report that describes a possible association between these two conditions.

Case Report

A 46-year-old Saudi female, without notable medical history has presented to the emergency department complaining of headache, dizziness, and nausea for one month. These symptoms persisted for two days and have disturbed her sleep pattern. At physical examination, she was vitally stable, and looked comfortable. Complete CNS examination including motor, sensory and gait assessment was intact.

Incidentally, she had a solitary skin colored nodule, measuring 1 cm in diameter with firm consistency and non-painful on palpation located at the left palm surface.

Basic laboratory investigations have been done and all were within normal range. MRI brain revealed Chiari malformation with cerebellar tonsils herniation associated with moderate hydrocephalus.

Excisional biopsy of the palm nodule showed sharply circumscribed hypocellualr dermal spindle cells with strikingly uniform storiform pattern of sclerotic collagen bundles separated by clefts arranged in a plywood-like pattern. The pathological diagnosis was consistent with sclerotic fibroma (Figure. 1, 2)

After diagnosis of sclerotic fibroma with Chiari malformation, Cowden disease was suspected, and further investigations advised to exclude internal malignancies. Thyroid Ultrasound showed diffuse asymmetrical enlargement of the right thyroid lobe. Complete abdominal ultrasound has not revealed any gross abnormalities.

Discussion

PTEN hamartoma tumor syndrome (PHTS) refers to a spectrum of disorders caused by mutations in the phophatase and tensin homolog (PTEN) gene, a tumor suppressor gene, on arm 10q [3].

Diagnostic criteria for Cowden disease, the main PTEN-related disorder, were first established in 1996. These criteria were based on clinical experience and case reports in the existing literature. Cutaneous involvements occur in more than 80% of patients and the characteristic mucocutaneous features including trichilimmomas, oral mucosal papillomatosis, acral keratosis, and palmoplantar keratosis that usually begin to appear during the second to third decades of life. Multiple sclerotic fibromas of the skin are relatively specific cutaneous marker of Cowden disease [4]. This disease is associated with an elevated risk for tumors of the breast, thyroid, prostate, brain, endometrium, and skin [5].

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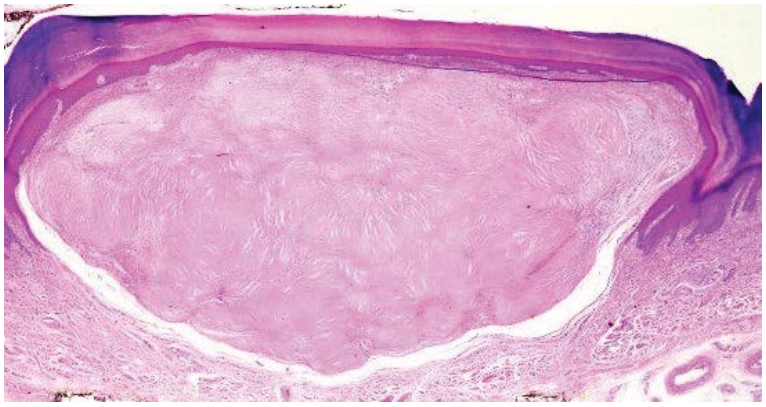

Figure 1: Excisional biopsy of the dermal nodule reveals a well circumscribed hypocellular fibrotic lesion (hematoxylin and eosin stain X20).

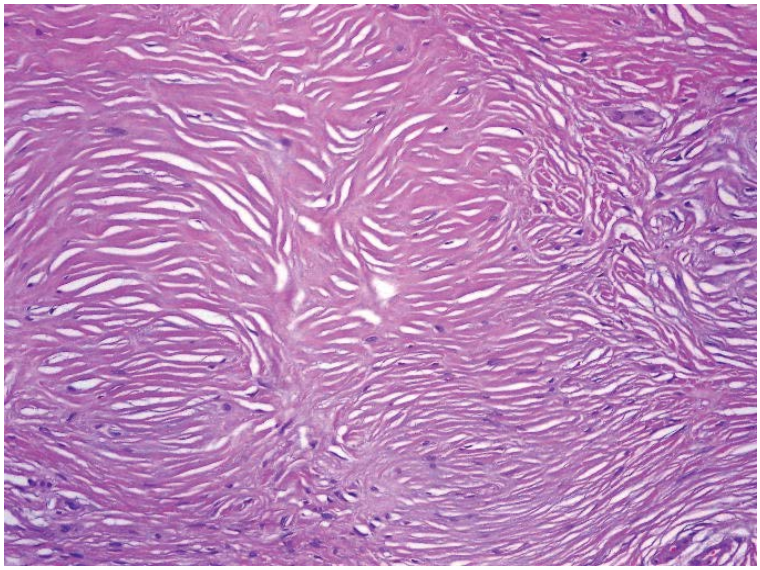

Figure 2: High power demonstrates the storiform pattern of sclerotic collagen bundles separated by clefts arranged in a plywood-like pattern (hematoxylin and eosin stain X400).

Sclerotic fibroma was first reported in a biopsy of a lesion from the tongue of a patient with Cowden's disease [6]. In 1989, Rapini and Golitz have described 11 cases of solitary sclerotic fibroma in the absence of Cowden's disease [7]. Clinically, sclerotic fibroma presents as asymptomatic, flesh-colored papule or nodule that can occur on any location. It is a benign lesion and no treatment is usually required. Microscopically, a non-encapsulated, well-circumscribed, hypocellular dermal nodule comprising hyalinized thick bundles of collagen is noted. A characteristic histological feature is thick collagen bundles that are arranged in a plywood-like pattern with prominent clefts between collagen bundles [8]. Immunohistochemically, this tumor usually stains positive for CD34, but negative for neurofilaments, S-100, and smooth muscle actin [7].

Chiari malformations are abnormalities of the posterior fossa that affect the cerebellum, brainstem, and the spinal cord. It can be a congenital or acquired in which the cerebellar tonsils protrude through the foramen magnum, and the disorder has been defined as downward herniation of the tonsils of 5 mm or more [9]. Three patients of Cowden disease with Lhermitte-Duclos have been reported with tonsillar herniation. [10, 11] Lhermitte-Duclos disease (dysplastic gangliocytoma of the cerebellum) is a rare slowly enlarging cerebellar hamartoma [12, 13]. It is believed to be a pathognomonic feature of Cowden disease [14].

In conclusion, we have presented the first case in which sclerotic fibroma was diagnosed in a patient with previous medical history of Chiari malformation of the cerebellum.

This association and their relevance with Cowden disease need more studies and case series.

Permission from the patient was obtained to conduct the diagnoses on her tissue specimens for the case report. In addition, the patient was told that she would remain anonymous and she was assured that the individual data would not be made available to another third party. To meet this promise, the authors keep any identifying information out of published reports.

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