COWDEN SYNDROME - REPORT OF TWO CASES

V Usha, T V Gopalakrishnan Nair, K Yogirajan

Two case of Cowden syndrome with typical features are presented. In addition to the cutaneous lesions like facial trichilemmomas, oral mucosal fibromas, acrokeratosis verruciformis and palmo plantar hyperkeratosis, the first patient had follicular carcinoma of thyroid, fibrocystic disease of the breasts and ovarian teratoma and second patient had a cerebellar hamartoma (Lhermitte Duclos syndrome) which is a rare entity.

Key words : Cowden syndrome, Lhermitte Duclos syndrome

Introduction

Cowden syndrome (CS) is characterized by multiple hamartomatous lesions of ectodermal, endodermal and mesodermal origin and associated with a predisposition to malignant tumours of the breast, thyroid and other organs.[1] More than 130 cases have been reported till 1995.[2] The disorder is probably more common but often missed because the clinical findings are subtle. We report 2 classical cases of CS.

Case Reports

Case 1

A 32- year-old lady was seen for evaluation of skin lesion on her face and extremities which were present since childhood. She had follicular carcinoma of thyroid 12 years back and was treated with surgery and radiation. She was treated with surgery for fibrocystic disease of the breasts, 1 year back (Fig.1).

The multiple skin-coloured follicular facial papules were confirmed as trichilemmoma on biopsy. The lesions on the extremities were hyperkeratotic verrucous papules and were typical of acrokeratosis verruciformis

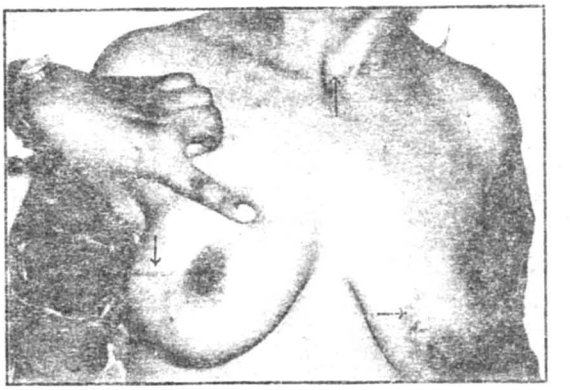

Fig. 1. Bilateral fibrocystic disease of the breast and surgical scar of follicular carcinoma thyroid.

on histopathology. Her gingival mucosae showed multiple firm whitish papules which coalesced to give a cobble stone appearance suggestive of mucosal fibromas. In addition, she had palmo plantar punctate keratosis with central depression and cutaneous horn on the nape of the neck. Diagnosis of CS was made because of the multiple hamartomas on the skin associated with carcinoma thyroid

From the Department of Dermatology and Venereology, Medical college Hospital, Trivandrum - 695011, India.

Address correspondence to:

Dr. T. V. Gopalakrishnan Nair

and fibrocystic disease of the breast. She was further evaluated in detail and USS pelvis revealed an asymptomatic left ovarian dermoid of 8X6X5 cms size.

Case 2

A 55 - year - old man was seen for skin lesions on face and extremities which were present since childhood. He had follicular facial papules, gingival fibromas, acrokeratosis verruciformis on the dorsa of hands and feet, multiple lipomas on the forearm and cutaneous horn on the left thigh.

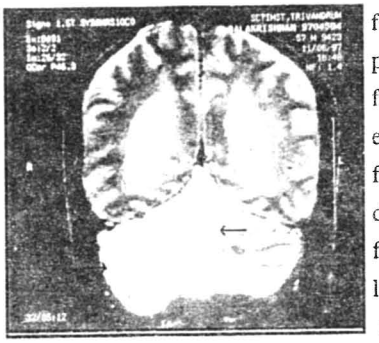

Fig 2: MPI picture showing right cerebellar hamartoma.

He had undergone surgery for a cerebellar hamartoma (Fig.2) 1 month back. He did not have any evidence of internal malignancy on screening. The cutaneous features were consistent with the diagnosis of Cowden syndrome.

Discussion

CS (multiple hamartoma syndrome) was named after Rachel Cowden, a 20-year-old patient who died of breast carcinoma with the syndrome. It is characterized by mucosal and cutaneous papillomatosis and fibromatosis with various benign and malignant neoplasms. The primary defect is thought to be an abnormality in the regulation of cell proliferation especially of tissues like epidermis, oral gastro intestinal mucosa, thyroid and breast epithelium. Salem and Steck have proposed the following diagnostic criteria.[3]

Major

1) Cutaneous facial papules, 2) Oral mucosal papillomatosis

Minor

1) acral keratoses , 2) Palmoplantar keratoses

A diagnosis of CS may be made with any of the following

1. 2 major criteria. 2. 1 major + 1 minor criteria

3. 1 major+positive family history of CS. 4. 2 minor+ positive family history of CS.

Skin lesions are present in 99-100% of cases. Age of onset is usually 2nd to 3rd decade. It is associated with carcinoma breast in 25% of female patients, thyroid swelling, multiple gastro-intestinal polyps, ovarian cysts, leiomyomas and skeletal anomalies.

Our first patient satisfied all the major and minor criteria and second patient had 2 major and 1 minor criteria. The cerebellar hamartoma in this patient is a rare association with Cowden syndrome and only 5 cases are reported in the world literature.[2] It is a dysplastic enlargement of cerebellar cortex with a classical 'inverted cortex appearance' on microscopy. This is due to atrophy of the central white matter and hypermyelination of the superficial layer. It may also occur sporadically.[4]

References

1. Harper J. Genetics and genodermatoses. In : Champion R H, Burton J L and Ebling F J G, editors. Rook A , Wilkinson D S, Ebling FJG. Textbook of Dermatology. Oxford : Blackwell, 1992 :332 -33.

2. Susan BM. Cowden syndrome (Multiple hamartoma syndrome) Dermatol Clin 1995 ; 13 : 27 -31.

3. Lee HR, Moon YS, Yeom CH, et al. Cowden's disease : a report on the first case in Korea and literature review. J Korean Med Sci 1997; 12 : 70 -75.

4. Vinchon M, Blond S, Lejeune JP, et al. Association of Lhermitte-Duclos and Cowden disease : report of a new case and review of the literature. J Neurol Neurosurg Psychiat 1994 ; 57 : 699 - 704.