COWDEN'S DISEASE

M L Khatri, M Shafi, N K Sen

A 24-year-old male patient developed multiple lesions of keratoacanthoma in the epidermal verrucous naevus. He also had multiple papillomatous lesions on the lips, buccal mucosa, gingiva and tongue, with positive family history of similar lesions. He also had multiple skin tags and patchy palmoplantar keratoderma and minimal kyphoscoliosis.

Key Words: Cowden's disease, Keratoacanthoma

Introduction

Cowden's disease was named after the patient in whom it was first described in 1963 by Lloyds and Dennis.[1] It is a complex hereditary syndrome, appears to be determined by autosomal dominant gene with high penetrance and moderate variation in symptoms.[2] It is characterized by hamartomas of ectodermal, endodermal, and mesodermal origin affecting multiple organs and organ systems. Mucocutaneous lesions like facial tricholemmomas, oral papillomatosis and acral keratoses are the characteristic features. In addition, they may have hamartomas and tumours of other organ systems like benign and malignant thyroid tumours, fibrocystic condition of the breast, carcinoma of the breast, female genital tract neoplasms, and gastrointestinal polyposis.[3]

About 100 cases have been reported.[1-6] We describe a case with mainly mucocutaneous lesions.

Case Report

A 24-year-old Libyan male patient came with the complaint of gradually developing asymptomatic growths of different types on various sites of the body. A verrucous epidermal naevus on the left leg and thigh was first noticed in early childhood which increased in size an extent at the age of 14 years. He also had gradually increasing, soft to firm multiple papular lesions (1-4 mm) on the lips (Fig.1), buccal mucosa, gingiva and tongue since the age of 8 years. There was also a smooth dome-shaped skin-coloured soft papular lesion (0.7 cm) on the upper lip since the age of 10 years. At the same time he also developed multiple nodular lesions over the verrucous naevus on the gluteal region and some near the medial maleolus, which gradually increased in size ranging from

Fig. 1. Oral papular lesions with high arched palate.

From the Departments of Dermatology and Pathology, Faculty of Medicine, A1-Fateh University of Medical Sciences, Tripoli, Libya.

Address correspondence to : Dr M L Khatri 860 Harrison Avenue, Apt 1009, Boston, MA 02118, USA.

1 to 5 cm with development of cutaneous horns in most of them (Fig.2). There were

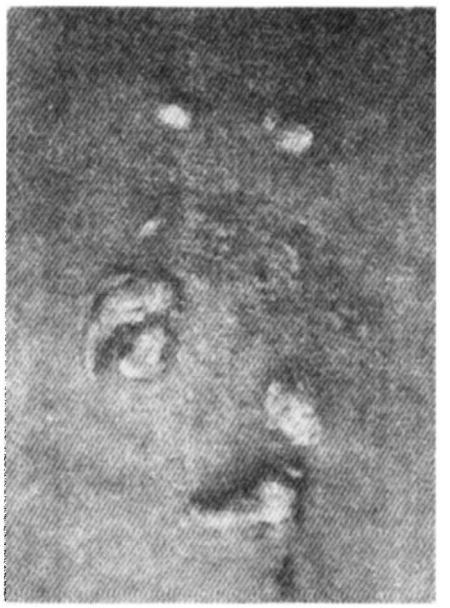

Fig. 2. Multiple lesions of keratoacanthona with cutaneous horns over the verrucous epidermal naevus on the gluteal region.

multiple skin tags on the sides of the neck. He also had patchy keratoderma of palms and soles since early childhood. His palate was high-arched. He has also been suffering from rheumatic heart disease since the age of 13 years.

His father, uncle and elder brother also have papillomatous lesions on the lips, buccal mucosa and tongue.

Histopathologic studies of lip lesions revealed changes of squamous papilloma and that of nodular lesions of the gluteal region showed picture of keratoacanthomas with cutaneous horns, and epidermal cysts.

Routine investigations on blood, urine and stools did not reveal any significant abnormality. X-ray of spine showed minimal kyphoscoliosis in the thorasic region. Barium meal and anema studies did not reveal any tumour in the gastrointestinal tract. Ultrasound abdomen also did not reveal any abormality.

Discussion

Presence of oral lesions in patient's father, uncle and elder brother favours the autosomal dominant mode of inheritance. Although various genetic studies including chromosomal analysis and DNA repair studies have been done in the past but the genetic cause of this disease remains elusive.[1,2,6]

Salem and Steck[4] have proposed certain diagnostic criteria for Cowden's disease: 1. Major clinical criteria- (a) cutaneous facial papules and (b) oral mucosal papillomatosis; 2. Minor clinical criteria - (a) acral keratoses and (b) palmoplantar keratoses; and 3. Family history of Cowden's disease. Our patient fulfilled majority of the criteria but he did not have facial tricholemmomas. These lesions were present in 84% of 83 patients studied in the past.[2]

Gastrointestinal lesions and thyroid involvement have been recorded in 71% and 67% respectively in the previously reported cases.[6] Our patient did not have such an involvement at the time of assessment. This patient developed lesions of keratoacanthoma, so frequent follow-up will be essential to check any malignant change in these lesions.

References

1. Lloyd KM II, Dennis M. Cowden's disease: A possible new symptom complex with system involvement. Ann Intern Med 1963; 58: 136-42.
2. Starink TM, Van Der Veen JPW, Arwert F, et al. The Cowden syndrome: a Clinical and genetic study in 21 patients. Clin Genet 1986; 29: 222-33.

3. Marra G, Armelao F, Vecchio FM, et al. Cowden's disease with extensive gastrointestinal polyposis. J Clin Gastroenterol 1994; 18: 42-7.

4. Salem OS, Steck WD. Cowden's disease (multiple hamartoma and neoplasia syndrome). J Am Acad Dermatol 1983; 8: 686-96.

5. Khatri ML, Shafi M. Cowden's disease: a case report. In: Penconesi E, ed. Dermatology in Europe: Proceedings of the 1st Congress of the European Academy of Dermatology and Venereology. 1st edn. Oxford: Blackwell Scientific Publications; 1991; 627-9.

6. Williard W, Borgen P, Bol R, et al. Cowden's disease - a case report with analysis at the molecular level. Cancer 1992; 69: 2969-74.