

## Bullous Pilomatricoma: A Rare Occurrence

Sir,

Pilomatricoma is a benign appendageal tumour originating from the hair matrix. The lesion is usually a solitary, deep dermal or subcutaneous, firm-to-hard, lobulated mass. However, patients with multiple lesions (1) and, rarely, with malignant (2, 3), bullous (4, 5) and anetodermic (6, 7) changes have been reported.

We report here on 2 patients presenting with pilomatricoma with bullous change.

### CASE REPORTS

#### Case 1

A 32-year-old woman had developed multiple, asymptomatic, hard swellings on the neck, back, buttocks and proximal extremities since the age of 7 years. The first swelling appeared on the upper back and gradually, over the years, new lesions developed at other sites. The patient noticed a gradually progressive small swelling on the back of the neck about 4–5 months earlier. There was a rapid increase in the swelling, with formation of bullae in the last week. She had no systemic complaints, nor did she have a past history of any significance. Both parents suffered from diabetes mellitus, but none of the family members had similar lesions. On cutaneous examination, 12 swellings were located (neck-1, upper back-2, arms-3, buttocks-3, thighs-3) with sizes varying from 1 cm in diameter to 6 × 3 cm, firm-to-hard, mobile, non-tender with normal overlying skin. The swelling on the neck was 6 × 3 cm in size, lobulated, hard, non-tender and with multiple tense bullae of 0.5 cm to 2 cm in diameter with haemorrhagic fluid (Fig. 1). Stretching the skin over the other swellings revealed their multifaceted surface. Systemic examination was unremarkable.

Routine investigation of blood, urine and stools, liver and renal function tests and chest X-ray were within normal limits. The excision biopsy of the neck lesion showed multiple islands of shadow cells with small foci of basophilic cells at the periphery in deep dermis, along with areas of necrosis and giant cell reaction within the tumour. The tumour was surrounded by a fibrous capsule. An increase in the connective tissue stroma of the dermis above the tumour could be seen. Large numbers of dilated lymphatic and blood vessels could be seen both within the tumour and in the dermis above the tumour. One of the lymphatic vessels was hugely dilated. Chronic inflammatory cell infiltrate was seen in the dermis. The clinical and histopathological features were those of bullous pilomatricoma.



Fig. 1. Multiple tense bullae over the swelling on the left side of the neck (case 1).

#### Case 2

A 25-year-old man had an asymptomatic swelling over the right scapular region for 4–5 months. The swelling suddenly increased in size, with formation of a bulla and pain for one week. There were no systemic complaints. Past history and family history were non-contributory. On cutaneous examination, a single, slightly tender, flaccid bulla, measuring 3.5 cm × 2 cm, was detected, with haemorrhagic fluid and striae-like extensions from the bulla into the surrounding normal skin, over the right scapular region. Within the bulla there was a single hard swelling, 0.5 cm in diameter. The skin over the bulla could be pushed deep into the dermis, which popped out with release of pressure. Systemic examination did not reveal any abnormality.

Routine investigation of blood, urine and stools, liver and renal function tests and chest X-ray were within normal limits. Excision biopsy of the lesion revealed islands of shadow cells surrounded by a capsule but no basophilic cells. In the dermis above the tumour, there was collagen degeneration, with large spaces and dilated blood vessels. Special stains for elastin did not reveal any abnormality. The clinical and histopathological findings indicated a diagnosis of bullous pilomatricoma.

### DISCUSSION

Pilomatricoma can occur at any age, but most cases have been recorded in the first two decades and are rarely familial (1). The lesions usually present as a solitary, firm-to-hard, deep-seated, lobulated mass with overlying normal skin in the head and neck areas. There have been a few reports of multiple lesions associated with myotonic dystrophy (8, 9). Pilomatricoma with a bullous appearance was reported (4, 5).

Julian & Bowers (10) reviewed 209 cases of pilomatricoma. They observed that, in pilomatricomas that were covered by vascular atrophic skin, the overlying dermis was filled with dilated lymphatic vessels lined with attenuated endothelial cells in association with numerous blood vessels and a chronic inflammatory cell infiltrate. They described this as a lymph-angiectatic variant. These features were also seen in our cases and in the earlier reports of pilomatricomas with bullous and anetodermic changes. These variants, along with the lymph-angiectatic variant, could represent a continuum of changes, a common factor being the lymphatic and blood vessel abnormalities, and the difference in manifestation could be due to the magnitude of the vessel abnormalities. Our case 2, which, clinically, appeared to be an anetodermic variant, did not have any elastin abnormalities, though other changes such as dilated blood vessels could be seen.

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H. R. Y. Prasad<sup>1</sup>, K. K. Verma<sup>1\*</sup>, B. K. Khaitan<sup>1</sup> and M. K. Singh<sup>2</sup>  
 Departments of <sup>1</sup>Dermatology & Venereology and <sup>2</sup>Pathology, All India Institute of Medical Sciences, New Delhi -110 029, India.  
 \*E-mail: kkverma61@hotmail.com

## Focal Dermal Hypoplasia Syndrome in a Male

*Sir,*

Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genodermatosis affecting tissue derived from embryonic mesoderm and ectoderm. Goltz et al. (1) first reported 3 cases of FDH in 1962 and reviewed 20 probable cases from the literature. The abnormalities seen in this multisystem condition include linear and reticular pigmentation, telangiectasias and papillomas, as well as skeletal, dental and ocular defects (1, 2). More than 200 cases of FDH have been reported or have been otherwise known (3). Most cases are seen in females and X-linked dominant inheritance fatal to male infants has been proposed as the likely mode of inheritance (4). Rare cases reflecting half chromatin mutation have been reported in male subjects (4–6). An additional case of FDH in a male with cystic bone defects is described herein.

### CASE REPORT

An 18-year-old man was first seen in our department for evaluation of multiple congenital anomalies. Since birth he had disseminated areas of slightly erythematous and telangiectatic macules with atrophic areas in linear pattern on his extremities (Fig. 1). The lesions were asymptomatic but slowly progressed to involve his 4 extremities and trunk. Numerous papillomas were noted in the perianal area, on the penis, inguinal region, right nasal fold and upper lip (Fig. 2).

Skeletal abnormalities included absence of the third and fourth digits of the right hand. The left foot had a lobster claw deformity with syndactyly of the second and third toes. Microcephaly, triangular face, asymmetric limbs and dorsolumbar scoliosis were also present. All his nails were thinned and many of them showed linear streaking. His ears, although fully formed, were asymmetric. The rest of the physical examination was normal.

Results of the ophthalmologic examination revealed microphthalmia, strabismus, nystagmus and coloboma of the left choroid. Odontologic examination disclosed irregularly arranged, deformed teeth with enamel hypoplasia. The patient's intellectual and motor development was markedly retarded.

Radiologic survey revealed cystic defects involving the right tibia. No evidence of osteopathia striata was noted in the patient's long bones. There was no family history of any cutaneous or other associated findings recorded in FDH. Chromosomal analysis showed a 46, XY karyotype.

Two skin biopsies were performed. The first specimen from one of the papillomas revealed epidermal hyperplasia with pronounced papillomatosis, parakeratosis and spongiform pustules in the epidermal invagination and moderate inflammatory infiltrate in the papillary dermis. A biopsy specimen from the atrophic lesions showed irregular mild acanthosis, oedematous papillary dermis with scanty lymphocytic infiltrate and thinning of the dermis.

Cutaneous and extracutaneous features permitted us to establish the diagnosis of FDH.



*Fig. 1.* Multiple linear, atrophic, erythematous macules of the legs.

### DISCUSSION

Goltz et al. (1) and Gorlin et al. (2) coined the term FDH for this rare genodermatosis on the basis of the histologically apparent areas of connective tissue hypoplasia. They summarized cutaneous, skeletal, ocular, dental and soft tissue defects associated with this syndrome. The clinical features of FDH have marked heterogeneity, with widely varying presentations (7). Stalder et al. (6) suggested that clinical spectrum of the disease is similar in both sexes.

Cutaneous findings are the most common manifestations of FDH (6). The present case showed the most prominent dermatologic manifestations including atrophic skin lesions, linear and reticular areas of telangiectasias, hypopigmentation, hyperpigmentation and multiple papillomas. Linear