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Case Presentation

Basal cell carcinomas in a young woman with Steinert's disease.

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Abstract

Steinert's disease or Myotonic dystrophy type I (DM1) is an autosomal dominant disease characterized by myotonia, muscular dystrophy, cataracts, hypogonadism, frontal balding, and electrocardiographic alterations.Several tumors have been associated with DM1 such as pilomatricoma, thymomas and insulinomas. Herein, we describe the unusual onset of multiple basal cell carcinomas in a young woman with DM1.

Keywords: Steinert's disease, basal cell carcinomas, dermoscopy.

Introduction

Myotonic dystrophy type I (DM1; MIM 160900), also called Steinert's disease, is an autosomal dominant disease characterized by myotonia, muscular dystrophy, cataracts, hypogonadism, frontal balding, and electrocardiographic alterations. It is the most common form of adult onset muscular dystrophy, with an incidence of approximately 13.5 per 100 000 live births [1].

In DM1 there is a triplet repeat of cytosine–thymine–guanine (CTG) in the DMPK gene located on chromosome 19q13.3.

Several tumors have been associated with DM1. The best known is pilomatricoma, but other benign and malignant neoplasms, including thymomas, pleomorphic parotic adenomas, insulinomas, testicular cancers, laryngeal squamous cell carcinomas, endometrioid ovarian carcinomas, and gastric as well as sigmoid colon adenocarcinomas have also been reported [2].

We report herein the extraordinary occurrence of multiple basal cell carcinomas (BCCs) in a female adolescent with DM1.

Case synopsis

An 18-year-old woman presented with a chief complaint of a new, asymptomatic growth that had enlarged since its appearance a month earlier in a pre-exiting well circumscribed pigmented lesion on her right parasternal region. She had Fitzpatrick skin type IV with a tendency to tan easily and not to burn. The patient reported a diagnosis of DM1 confirmed by a genetic study, although she had relatively mild disability from the disease. Several members of her family also had myotonic dystrophy. One year ago, two skin lesions had been removed (left breast and abdominal region) that were diagnosed

histologically as basal cell carcinomas. There was no history of either sun bed usage or excessive sun exposure. In addition, we excluded a diagnosis of Gorlin-Goltz because there were no diagnostic manifestations of this syndrome. Routine laboratory tests, electrocardiograms and eye examinations were normal.

Clinical examination revealed a smooth, palpable glistening pearly translucent papule arising within a pre-existing atypical appearing nevus, measuring 0.4x0.5 cm in diameter. The new papule had irregular borders and variegated shades of pigment (Figure 1). The dermoscopic evaluation showed small branching vessels, lobulated leaf-like brown areas, and a reticulate and well-demarcated pigmented network adjacent to the raised portion of the lesion (Figure 2). These dermoscopic findings were suggestive for a basal cell carcinoma and an adjacent benign nevus. The papule was completely excised and submitted en toto for histopathology.

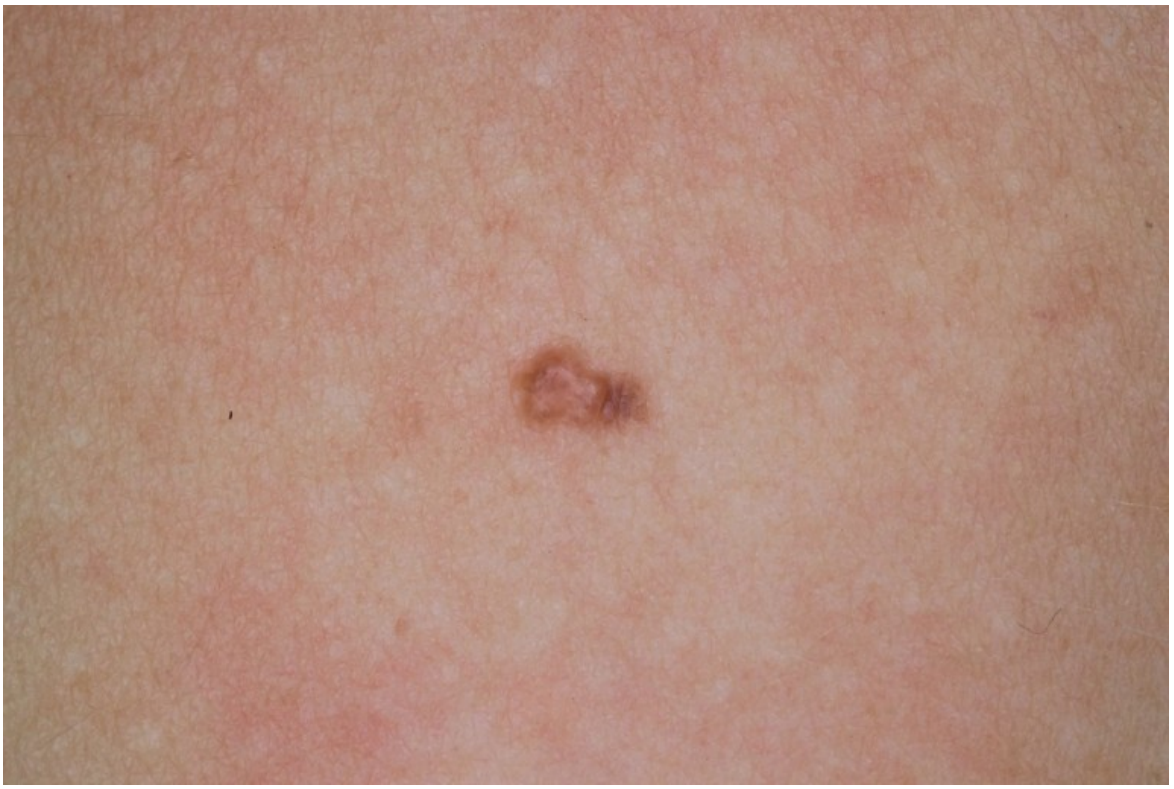

Figure 1. Clinical picture of basal cell carcinoma in a girl with Steinert's disease

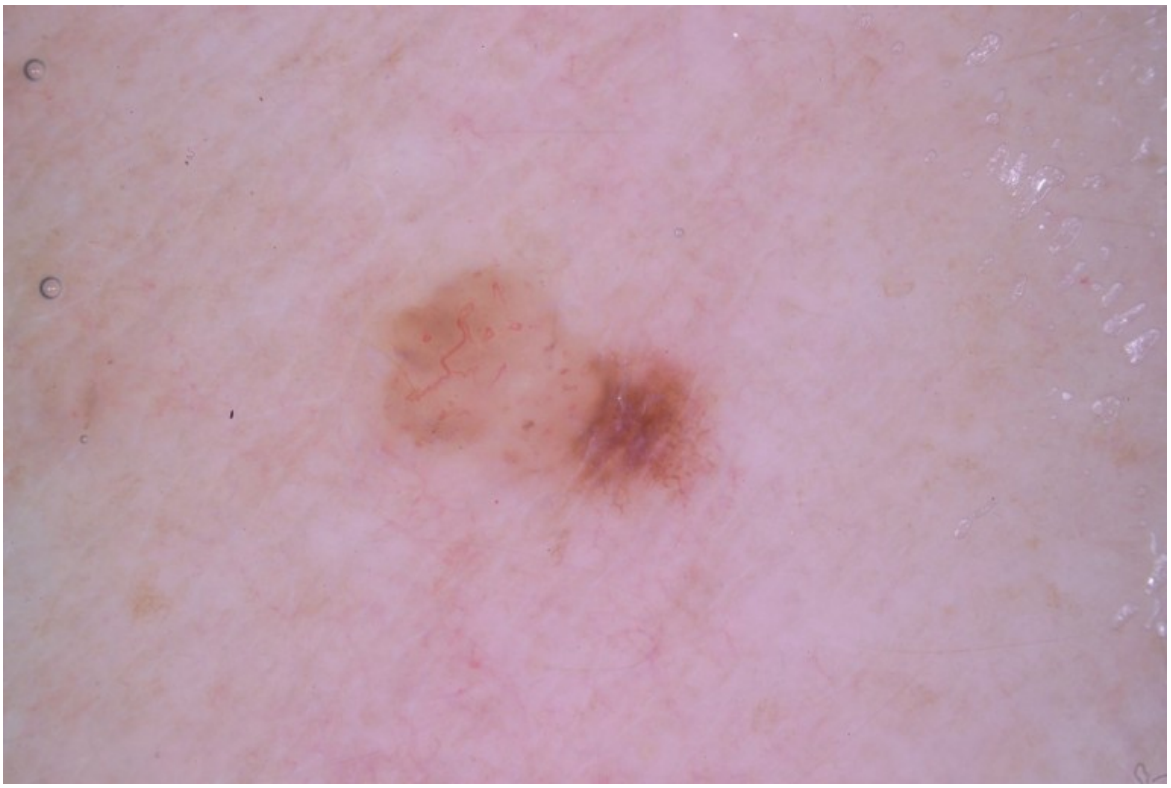

Figure 2. Dermatoscopic image: small branching vessels and lobulated leaf-like brown areas with a reticulate pigmented network adjacent to the popular portion of the lesion

Histopathologic findings revealed proliferating basaloid cells in strands and basaloid islands invading the dermis. The peripheral cells of the basaloid islands have a characteristic “palisading” arrangement with speckled pigmentation suggestive of a pigmented basal cell carcinoma.

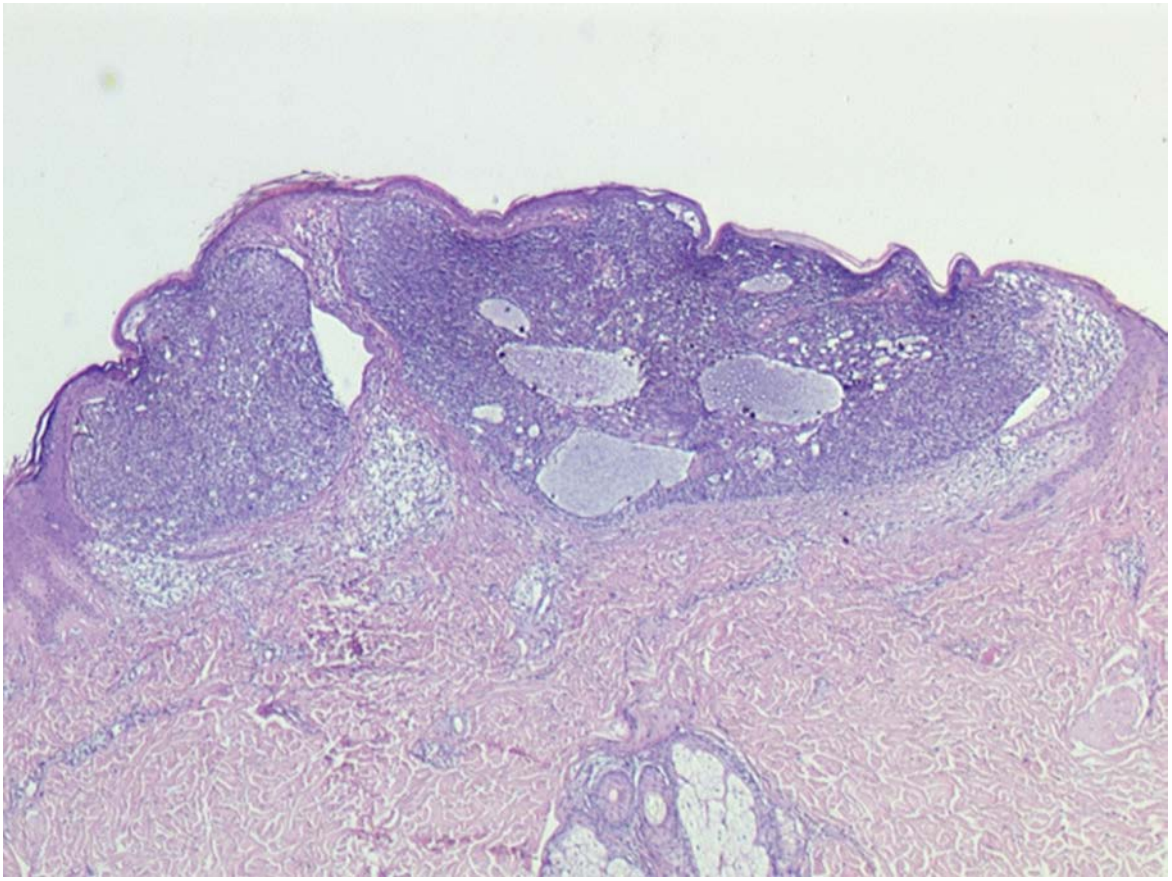

Figure 3. Proliferating basaloid cells in strands and basaloid islands invading the dermis.

In the medical literature, only other five cases of this rare association have been reported; in all cases, however they were adult patients (34-62 years) [1, 3-6]. Our case is the youngest reported to date. BCCs rarely develop in young persons because 95% of these neoplasms affect people over 35 years of age [7].

In the pediatric and adolescent age groups, BCCs usually occurs in the setting of a known genetic defect as Xeroderma Pigmentosum and Gorlin Goltz syndrome [8]. Moreover, in our patient, the BCCs were not present in sun-exposed areas and the surrounding area was not photodamaged.

Several publications have linked 19q defects to a higher risk of basal cell carcinoma. Defects in this gene could justify both the onset of multiple cancers at a young age and the DM1 [9, 10].

We cannot exclude with certainty that the above association is a coincidental finding. Further studies are needed to evaluate an associated risk; but we recommend skin examinations in patients with DM1, even at a young age.

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