SHORT COMMUNICATION

Keloids, Spontaneous or After Minor Skin Injury: Importance of Not Missing Bethlem Myopathy

Constanza Echeverría1, Alejandra Díaz2, Bernardita Suárez2,3, Jorge A. Bevilacqua1, Carsten Bonnemann1, Enrico Bertini6 and Claudia Castiglioni3*

1School of Medicine, Pontificia Universidad Católica de Chile, 2National Institute of Rehabilitation, Pedro Aguirre Cerda, 3Departamento de Neurología y Neurocirugía, Hospital Clínico Universidad de Chile y Programa de Anatomía y Biología del Desarrollo, Facultad de Medicina, Universidad de Chile, 4Department of Pediatrics, Neurology Unit, Clínica Las Condes, Lo Fontecilla 441, Santiago, Chile, 5Neuromuscular and Neurogenetic Disorders of Childhood Section, Neurogenetics Branch, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, USA, and 6Unit of Neuromuscular and Neurodegenerative Disorders, Department of Neurosciences, IRCCS Bambino Gesù Children’s Hospital, Rome, Italy. E-mail: ccastiglioni@clc.cl

Accepted Aug 22, 2016; Epub ahead of print Aug 26, 2016

Collagen VI-related muscular dystrophies (COL6-RD) comprise a continuous spectrum of clinical severity, ranging from the most severe phenotype Ullrich congenital muscular dystrophy (UCMD), through intermediate phenotypes, to a milder Bethlem myopathy (BM) (1). These inherited diseases are caused by autosomal dominant or recessive mutations in the genes COL6A1, COL6A2 and COL6A3, which encode the main 3 α-chains of collagen VI, a component of the extracellular matrix (ECM) that is present in the vast majority of connective tissues and is implicated in its organization (2). The prevalence of COL6-RD is estimated as 0.77:100,000 in BM and 0.13:100,000 in UCMD (3). Clinical features are secondary to the dysfunction of the collagen VI in the ECM of muscle and connective tissues of tendons, subcutaneous and dermal layers of the skin (1, 2). BM is characterized by slow progressive weakness of the proximal muscles and contractures that characteristically involve multiple joints. Although the course of the disease is slowly progressive, many patients exhibit a marked decrease in muscle strength between the 4th and 5th decades, with approximately half of them becoming wheelchair-dependent after the 5th decade (1, 2). Cutaneous manifestations include: presence of keratosis pilaris, mostly in the extensor surfaces of proximal legs and arms; formation of abnormal scars, especially keloids, either spontaneous or after minor trauma; rough or dry skin; striae rubrae; cigarette paper scars; and, in younger patients with UCMD, soft velvety skin on the palms and soles (4–7). Preclinical studies have observed decreased tensile strength of the skin and altered collagen fibril and basement membrane architecture in Col6a1 null mice (8).

We report here a case of spontaneous formation of keloids in a previously non-diagnosed BM patient who had 3 daughters also displaying features of BM.

CASE REPORT

The patient, a male aged 45 years, was born with irreducible congenital torticollis, needing surgery to release the contracture. He had normal development with no delay in motor milestones and did not report any difficulties until the third decade of life. After the age of 25 years, he perceived mild difficulty in climbing stairs and reported prominent acne and their skin was not darkly pigmented.

DISCUSSION

Keloids are a multifactorial disorder characterized by an abnormal response to tissue regeneration. Genetic susceptibility has been postulated by its prevalence

1http://www.medicaljournals.se/acta/content/?doi=10.2340/00015555-2523
whether the degree of propensity for keloid formation correlates with other aspects of the disease, such as contracture formation.

REFERENCES