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Collagen VI-related myopathy with scoliosis alone: a case report

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The symptoms, severity, and age of onset of collagen type VI-related disorders vary greatly. In most cases, affected individuals have muscle weakness and degeneration and skeletal abnormalities such as curvature of the spine (scoliosis) and contractures. Bethlem Myopathy (Benign Congenital Myopathy with Contractures)

Collagen type VI-related myopathy | Practical Neurology

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Oct 01, 2017 - His perinatal history and prior motor milestones were normal, and there was no relevant family history. On examination, he had a scoliosis, multiple joint contractures with distal hypermobility, follicular hyperkeratosis and keloid scar formation (figure 1). Figure 1 Examination findings in collagen type VI-related myopathies.

Cited by: 1 Author: Paulo Victor Sgobbi de Souza, Thiago Bo... Publish Year: 2017

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Collagen VI

Collagen VI is a type of collagen primarily associated with the extracellular matrix of skeletal muscle. ColVI maintains regularity in muscle function and stabilizes the cell membrane. It is synthesized by a complex, multistep pathway that leads to the formation of a unique network of linked microfilaments located in the extracellular matrix. ColVI plays a vital role in numerous cell types, including chondrocytes, neurons, myocytes, fibroblasts, and cardiomyocytes. ColVI molecules are made up of three alpha chains: $\alpha 1(VI)$, $\alpha 2(VI)$, and $\alpha 3(VI)$. It is encoded by 6 genes: COL6A1, COL6A2, COL6A3, COL6A4, COL6A5, and COL6A6. The chain lengths of $\alpha 1(VI)$ and $\alpha 2(VI)$ are about 1,000 amino acids. The chain length of $\alpha 3(VI)$ is roughly a third larger than those of $\alpha 1(VI)$ and $\alpha 2(VI)$, and it consists of several spliced variants within the range of 2,500 to 3,100 amino acids.

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Author: Carsten G. Bönnemann

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Collagen VI-related myopathy with scoliosis alone: A case report

Li JY *et al.* Collagen VI-related myopathy with scoliosis alone

Jun-Yu Li, Shuo-Zi Liu, Dan-Feng Zheng, Ying-Shuang Zhang, Miao Yu

Abstract

BACKGROUND

Scoliosis is a complex three-dimensional deformity of spine and one of the common complications of collagen VI-related myopathy, caused by mutations in *COL6A1*, *COL6A2* and *COL6A3* genes. The typical clinical presentations of collagen VI-related myopathy include weakness, hypotonia, laxity of distal joints, contractures of proximal joints and skeletal deformities.

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Author: Carsten G. Bönnemann

Therapy of Collagen VI-Related Myopathies (Bethlem and ...

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The collagen VI-related myopathies comprise two major forms, Bethlem myopathy (BM) and Ullrich congenital muscular dystrophy (UCMD), which show a variable combination of muscle wasting and ...

[PDF] A novel de novo COL6A1 mutation emphasizes the role ...

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Collagen VI-related myopathy is a group of disorders affecting skeletal muscles and connective tissue. The most common symptoms are muscle weakness and joint deformities which limit the movement and progress over time. Several forms of collagen VI-related myopathies have been described: Bethlem myopathy, an intermediate form and Ullrich

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Author: Agnieszka A. Koppolu, Agnieszka Madej...

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Axial myopathy: an overlooked feature of muscle diseases ...

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Classically, myopathies are categorized according to limb or cranial nerve muscle affection, but with the growing use of MRI it has become evident that many well-known myopathies have significant involvement of the axial musculature (Kornblum et al. , 2006 ; Kesper et al. , 2009 ; Quijano-Roy et al. , 2012). New disease entities with selective axial muscle involvement have also been described (Loseth et al. , 2013), but overall t...

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Author: Nanna Witting, Linda K. Andersen, John ...

Publish Year: 2016

Published: Dec 14, 2015