Dural ectasia in individuals with Marfan-like features but exclusion of mutations in the genes FBN1, TGFBR1 and TGFBR2.

Mutations in the genes FBN1, TGFBR1, and TGFBR2 can result in heritable connective tissue disorders comprising the Marfan syndrome and the Loeys-Dietz syndrome. Dural ectasia is a characteristic manifestation of both syndromes. However, dural ectasia has not yet been investigated in connective tissue disorders that are unrelated to mutations in the FBN1, TGFBR1 or TGFBR2 genes. Here, we assessed dural ectasia in 33 individuals both with typical manifestations of heritable connective tissue disease and in whom mutations in all three genes had been excluded. We identified 19 individuals with dural ectasia (58%), who exhibited major skeletal manifestations of the Marfan syndrome more frequently than the remaining 14 persons without dural ectasia (p = 0.06). Moreover, only persons with dural ectasia fulfilled clinical criteria of the Marfan syndrome (p = 0.01). Conversely, aortic aneurysm (12 patients; p = 0.8), aortic dissection (five patients; p = 0.1), spontaneous dissection of the carotid arteries (five patients; p = 1), and mitral valve prolapse (13 patients; p = 0.4) were similarly frequent irrespective of dural ectasia. We conclude that dural ectasia is a marker for connective tissue disease which coincides with skeletal rather than with
cardiovascular manifestations, and which may involve currently uncharacterized pathogenetic mechanisms and syndromes.