Dural ectasia in Loeys-Dietz syndrome: comprehensive study of 30 patients with a TGFBR1 or TGFBR2 mutation.

The purpose of this study was to assess the frequency, severity, and clinical associations of dural ectasia (DE) in Loeys-Dietz syndrome (LDS). Database analysis of three German metropolitan regions identified 30 patients with LDS and TGFBR1 mutation in 6 and a TGFBR2 mutation in 24 individuals (17 men; mean age: 31 ± 19 years), as well as 60 age and sex-matched control patients with Marfan syndrome carrying a FBN1 mutation. DE was present in 22 patients with LDS (73%), and it related to skeletal score points (p = 0.008), non-skeletal score points (p=7), systemic score points (p = 0.010). Similarly, the severity of DE was related to body height (p = 0.010) and non-skeletal score points (p = 0.004). Frequency (p = 0.131) and severity of DE (p = 0.567) was similar in LDS and Marfan syndrome. DE is a manifestation of LDS that occurs with similar frequency and severity as in Marfan syndrome. Severity of DE may serve as a marker of the overall connective tissue disease severity. LDS may be considered in patients with DE.