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Hum. Mol. Genet. 3: 1583-1587.

Abstract 1 of 1

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ARTICLES

Assignment of a locus for dominantly inherited venous malformations to chromosome 9p

LM Boon, JB Mulliken, M Vikkula, H Watkins, J Seidman, BR Olsen and ML Warman

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Venous malformation is the most common type of vascular anomaly. Depending upon size and location, these slow-flow anomalies may cause pain, anatomic distortion, or threaten life. Most venous malformations occur sporadically and present as solitary lesions. They also occur in several syndromes, some of which demonstrate Mendelian inheritance. We have mapped the locus for an autosomal dominant disorder in a three generation family that manifests as multiple cutaneous and mucosal venous malformations. This locus lies within a 24 cM interval on chromosome 9p, defined by the markers D9S157 and D9S163. The alpha and beta interferon gene cluster and the putative tumor suppressor genes MTS1 and MTS2 are also in this region. Characterization of the gene responsible for this disorder should yield insights into the precise pathogenic mechanisms for venous malformations.

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