Mutations in TGFBR2 Gene Cause Spontaneous Cervical Artery Dissection

Supplemental Material

Supplemental Table 1

Prevalence of connective tissue abnormalities in the study group.

* Beighton Score [1] ≥5

Supplemental Table 2.

Polymorphisms found in *TGFBR1* and *TGFBR2* genes.

Nucleotide numbering reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence (*TGFBR1*: cDNA: NM_004612.2, protein: NP_004603.1; *TGFBR2*: cDNA: NM_003242.5, protein: NP_003233.4). The initiation codon is codon 1.

Clinically detectable connective tissue abnormalities	**n (%)**
Skeletal features	
Steinberg thumb sign	0 (0.0)
Walker-Murdoch wrist sign	0 (0.0)
Scoliosis	4 (7.1)
Arachnodactily	3 (5.3)
Pes planus	0 (0.0)
Joint hyperextensibility *	4 (7.1)
Joint laxity	17 (30.3)
Recurrent joint dislocation	3 (5.3)
Pectus deformity	3 (5.3)
Craniofacial features	
Micrognathia	3 (5.3)
Proptosis	1 (1.8)
Hypertelorism	2 (3.6)
Abnormal uvula	3 (5.3)
Malar hypoplasia	2 (3.6)
Cleft palate	1 (1.8)
Blue sclerae	1 (1.8)
Cutaneous features	
Hyperextensibility of the skin	0 (0.0)
Easy bruising	1 (1.8)
Striae distensae (without apparent cause)	2 (3.6)
Velvety texture	3 (5.3)
Widened and atrophic scars	1 (1.8)
Other features	
Hernia	3 (5.3)
Hiatal hernia	1 (1.8)
Umbelical hernia	2 (3.6)
Inguinal hernia	0 (0.0)
Cardiovascular features	
Cerebral aneurysm	3 (5.3)
Atrial septal defect	16 (28.6)
Mitral valve prolapse	6 (10.7)
Arterial tortuosity	3 (5.3)
Aortic dissection	1 (1.8)
Recurrent artery dissection	2 (3.6)
Dural arteriovenous fistula	1 (1.8)

Supplemental Table 1

	refSNP ID	Nucleotide Variation	Protein Variation	Exon/Intron
TGFBRI	*novel*	c.49 C>T	p.L17L	Exon 1
	rs11466445	c.69_77delGGCGGCGGC	p.A23_A25delinsA	Exon 1
	rs56014374	c.457 G>A	p.V153I	Exon 3
	rs7861780	c.1125 A>C	p.T375T	Exon 6
	rs334354	c.1255+24 G>A		Intron 7
	novel	c.1256-109 InsA		Intron 7
	rs67687202	c.1386+90_1386+94delTCTT		Intron 8
	rs868	c.*69A>G		3'UTR
TGFBR2				
	rs2306856	c.-128 C>G		5'UTR
	rs17025709	c.-64 G>C		5'UTR
	rs1155705	c.263+7 A>G		Intron 2
	rs11466512	c.455-4 T>A		Intron 3
	novel	c.984 C>T	p.H328H	Exon 4
	rs2229102	c.999 A>G	p.L333L	Exon 4
	rs2228048	c.1167 C>T	p.N389N	Exon 4
	rs2228047	c.1266 A>G	p.A422A	Exon 5
	rs2276767	c.1525-91 C>A		Intron 6

Supplemental Table 2

Supplemental references

1. Beighton P, Solomon L, Soskolne CL. Articular mobility in an African population. Ann Rheum Dis 1973;32:413–418.