

## EDS - översikt

Clinical EDS subtype	Abbreviation	IP*	Genetic basis	Protein
Classical EDS	cEDS	AD	Major: COL5A1, COL5A1	Type V collagen
			Rare: COL1A1 c.934C>T, p.(Arg312Cys)	Type I collagen
Classical-like EDS	clEDS	AR	TNXB	Tenascin XB
Cardiac-valvular	cvEDS	AR	<i>COL1A2 (biallelic mutations that lead to COL1A2 NMD and absence of pro <math>\alpha 2(I)</math> collagen chains)</i>	Type I collagen
Vascular EDS	vEDS	AD	Major: COL3A1	Type III collagen
			Rare: COL1A1 c.934C>T, p. (Arg312Cys) c.1720C>T, p.(Arg574Cys) c.3227C>T, p.(Arg1093Cys)	Type I collagen
Hypermobile EDS	hEDS	AD	Unknown	Unknown
Arthrochalasia EDS	aEDS	AD	<i>COL1A1, COL1A2</i>	Type I collagen
Dermatosparaxis EDS	dEDS	AR	<i>ADAMTS2</i>	ADAMTS-2
Kyphoscoliotic EDS	kEDS	AR	<i>PLOD1</i>	LH1
			<i>FKBP14</i>	FKBP22
Brittle Cornea syndrome	BCS	AR	<i>ZNF469</i>	ZNF469
			<i>PRDM5</i>	PRDM5
Spondylodysplastic EDS	spEDS	AR	<i>B4GALT7</i>	$\beta 4$ GalT7
			<i>B3GALT6</i>	$\beta 3$ GalT6
			<i>SLC39A13</i>	ZIP13
Musculocontractural EDS	mcEDS	AR	<i>CHST14</i>	D4ST1
			<i>DSE</i>	DSE
Myopathic EDS	mEDS	AD or AR	<i>COL12A1</i>	Type XII collagen
Periodontal EDS	pEDS	AD	<i>C1R</i>	C1r
			<i>C1S</i>	C1s

\* Inheritance Pattern: AD = autosomal dominant; AR = autosomal recessive