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Ehlers Danlos Syndrome, molecular mechanism, clinical and structural correlations

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Ehlers Danlos Syndrome (EDS), is a clinically heterogeneous abnormality of extracellular matrix characterised by cutaneous fragility, ligamentous laxity and variable vascular fragility. Up to nine varieties were previously recognised, more recently subclassified into five main subgroups. Most of these are caused by mutations in various connective tissue matrix components, such as the COL1A1, COL1A2, COL3A1, COL5A1 and COL5A2 genes. Nearly all of them disrupt collagen matrix assembly and are detectable by structural alterations of collagen fibril organisation, as detected by cutaneous electron microscopy. This talk correlates clinical phenotype with structural and molecular changes in these disorders.

References

Pope FM, Burrows NP (1997) *Ehlers Danlos Syndrome has varied molecular mechanisms* Journal of Medical Genetics 34(5) 400-410

Beighton P, De Paepe A, Steinmann B et al (1998) *Ehlers Danlos Syndromes, revised nosology, Villefranche 1997, American Journal of Medical Genetics 77, 31-37.*