# DISEASE:
# Periodontal Ehlers-Danlos syndrome

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| **NAME:** | Periodontal Ehlers-Danlos syndrome |
| **DESCRIPTION:** | A rare type of Ehlers-Danlos syndrome characterized by childhood or adolescence onset of severe, intractable periodontitis, lack of attached gingiva, and presence of pretibial plaques. Additional manifestations are easy bruising, hypermobility mainly of the distal joints, skin hyperextensibility and fragility, abnormal scarring, recurrent infections, hernias, marfanoid facial features, acrogeria, and prominent vasculature. |
| **ORPHACODE:** | 75392 |
| **SYNONYMS:** | EDS VIII<br>Ehlers-Danlos syndrome type 8<br>Ehlers-Danlos syndrome, periodontitis type<br>Periodontal EDS<br>pEDS |
| **XREF(S):** | Orphanet<br>OMIM<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | C1S<br>C1R |

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| **CREATED:** | 13 May 2019 - 01:02 |
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# RELATED CONTENT

## Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Gent

## Related Analytes

- complement C1r
- complement C1s

## Related Gene Panels

- Ehlers Danlos periodontal type (2 genes) - UGent
- Ehlers-Danlos syndrome -UGent