

Ehlers-Danlos Syndrome (EDS)

Main Features

- Group of heritable connective tissue disorders
- tissue fragility
- hypermobility
- skin and joint hyperextensibility

Eye Findings

- Floppy eyelids
- Ptosis 32%
- Infraorbital creases 30%
- Myopia 25%
- Epicanthal folds 19%
- Strabismus 8%
 - convergence insufficiency
- lenticular changes
 - Ectopia lentis
- Blue sclerae
- conjunctivochalasis
- Dry eye
- Thin corneal pachymetry and steep keratometry
- possibly angoid streaks (debatable)
 - only documented present in first report of this condition

Other Findings

- Vascular type
 - cervical artery dissection
 - multiple bone fractures
 - prolonged recovery from concussion
 - inguinal hernia
 - poor exercise tolerance
 - pain intolerance

Etiology

- Gene defect involving collagen
 - usually collagen types I, III, and V
 - [Ehlers-Danlos Syndrome International Classification 2017](#)

- Disruption of the collagen fibrils
- Classical EDS
 - mutations in COL5A1 and COL5A2
- Vascular EDS
 - mutations in COL3A1

Ddx of joint hypermobility

- [Marfan Syndrome](#)
- Cutis Laxa
- Pseudoxanthoma elasticum
- Loeys-Deitz syndrome

Reference

EyeWiki

EL, Doyle JJ. Angioid Streaks Are Not a Common Feature of Ehlers-Danlos Syndrome. *JAMA Ophthalmol.* 2019;137(3):239.

2017 International Classification

[syndrome](#)

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