Opis choroby *

Definicja

A rare, autosomal recessive systemic dysfunction of the hemostatic pathway, that is due to a defect in the coagulation factor XII (FXII or Hageman factor), and is either asymptomatic or characterized by a prolonged activated partial thromboplastin time and an increased risk for thromboembolism. FXII deficiency is strongly associated with primary recurrent abortions.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital Hageman factor deficiency
Wrodzony Niedobór czynnika Hagemana

Kod ORPHA
330

Kod OMIM
234000

Kod ICD10
D68.2

Kod ICD11
3B15

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*Źródło

orphanet