Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation. ORPHA:178396

Hemorrhagic disease due to alpha-1-antitrypsin Pittsburg mutation is a rare, genetic, constitutional coagulation factor defect disorder characterized by a bleeding tendancy of variable severity due to methionine 358 to arginine replacement (Pittsburgh mutation) in the alpha-1-antitrypsin protein. Patients present with spontaneous hematomas, hematomas following minor trauma or surgery and, in female patients, ovarian hematomas after ovulation.