

# Genotype and Phenotype Correlation in Hereditary Thrombotic Thrombocytopenic Purpura (Upshaw-Schulman Syndrome)

NCT01257269

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**Status** RECRUITING  
**Sponsor** Insel Gruppe AG, University Hospital Bern  
**Enrollment** 450 participants

## Key Eligibility Criteria

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### Inclusion (3)

- Severe ADAMTS13 deficiency ( < 10% activity) and no ADAMTS 13 inhibitor on two or more occasions at least one month apart
- Being a family member of a confirmed or suspected patient
- Molecular analysis of ADAMTS13 gene with one or more mutations and/or positive infusion trial (full recovered ADAMTS13 activity after infused fresh frozen plasma (FFP) with a plasma half-life of 2-4 days)

## Locations (7 total)

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University of Oklahoma Health Sciences Center, Department of Medicine, PO Box 26901, Oklahoma City, Oklahoma, United States

Medical University of Vienna, Department of Medicine 1, Div. Hematology and Hemostasis Waehringer Guertel 18-20, Vienna, Austria

Institute of Hematology and Blood Transfusion, Coagulation Laboratory, U nemocnice 1, Prague, Czechia

... and 4 more locations