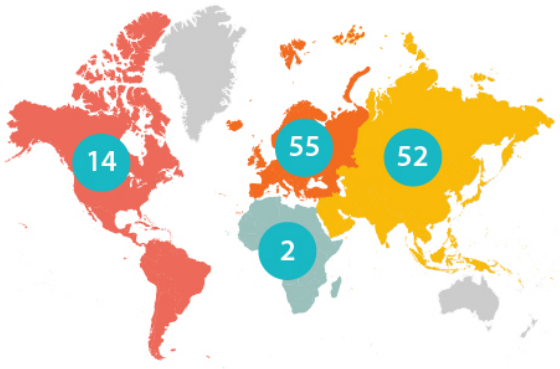
Presentation of an international registry for the ultra-rare disease congenital thrombotic thrombocytopenic purpura

Hereditary thrombotic thrombocytopenic purpura registry

Patient recruitment from 2006 to 2017

123 Patients with congenital thrombotic thrombocytopenic purpura

Median age	Years
• at enrollment	26.1 (range 0.1-75.0)
• at clinical diagnosis	16.7 (range 0-69.8)
• at reported overt disease onset	4.5 (range 0-69.8)



ADAMTS13 parameters

- ADAMTS13 activity values
- Functional inhibitors
- Anti-ADAMTS13 antibodies
- Molecular analysis of the ADAMTS13 gene

Results

- Identified in 121/123 patients, all values <10%
- Negative in all reported cases
- Positive in 12/103 patients analyzed
- 98 different ADAMTS13 mutations were identified

- The most frequent mutation observed was **ADAMTS13 c.4143_4144dupA** in exon 29, present on 60/246 alleles
- 19 of the 98 mutations have not been reported before

van Dorland *et al.*, Haematologica, 2019