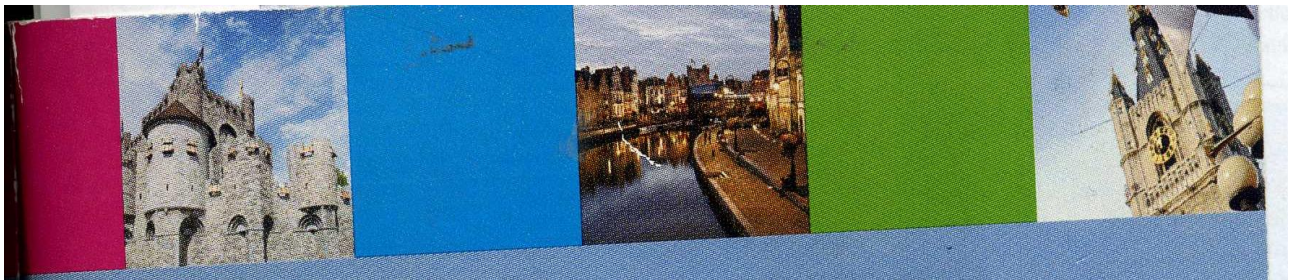

First International Symposium on the Ehlers-Danlos Syndrome

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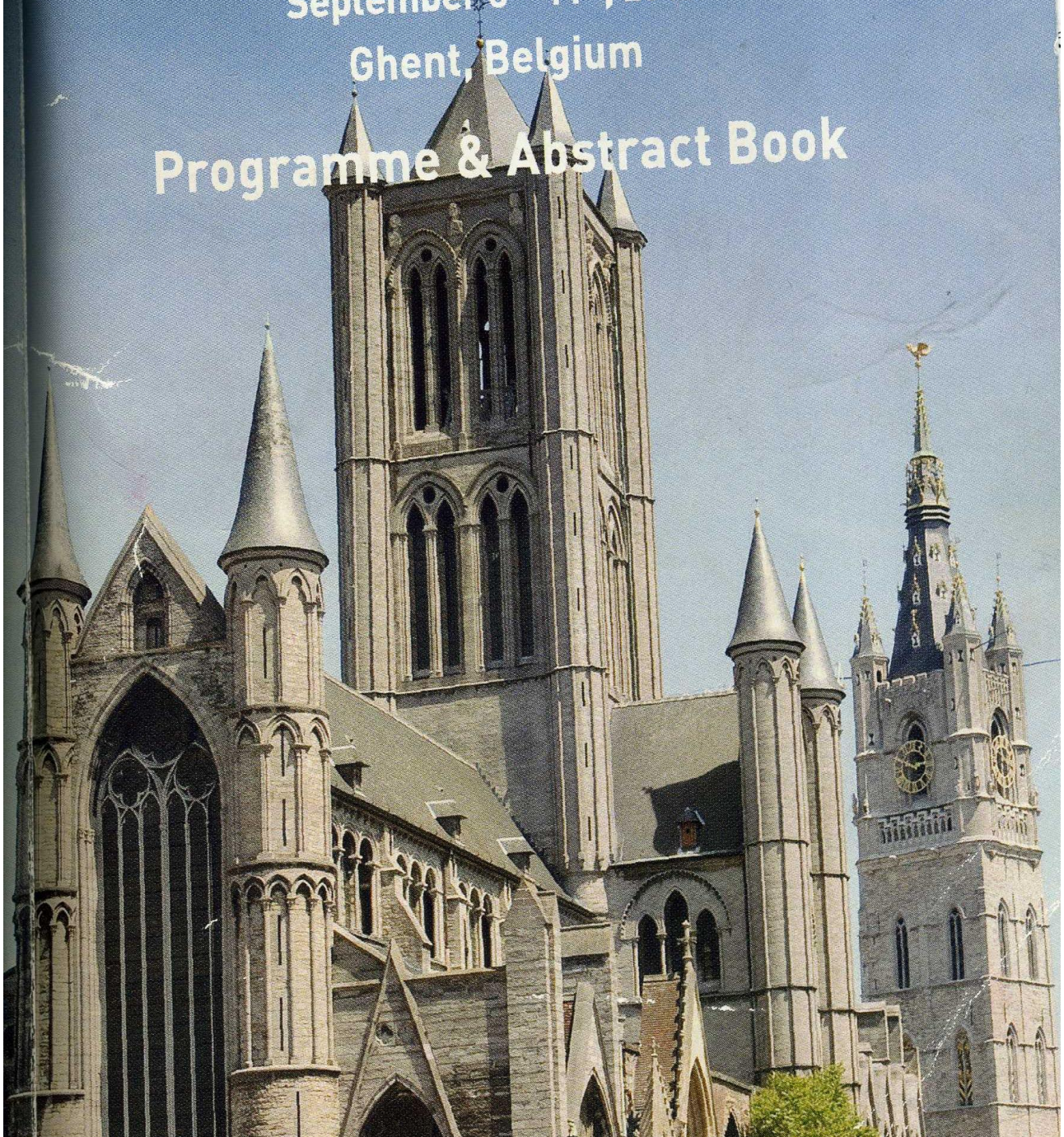
Ghent, Belgium

Programme & Abstract Book



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Diagnosis of vascular Ehlers-Danlos syndrome in Italy: clinical findings and novel COL3A1 mutations

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Vascular Ehlers-Danlos Syndrome (vEDS) (MIM #130050) is a dominantly inherited rare connective tissue disorder characterised by fragility of skin, arteries, and internal organs. vEDS is due to mutations in the COL3A1 gene encoding type III collagen (COLLIII).
Patients with a clinical vEDS diagnosis were tested by SDS/PAGE for COLLIII features and/or for mutations in COL3A1 by cDNA and/or gDNA sequencing.
We characterised 22 vEDS Italian patients. Thin and translucent skin was observed in 82% of the patients, easy bruising in 27% and acrogeria in 18%; typical facial appearance was present in 82%. A major complication occurred in 86% of the patients; the most frequent first complications were vascular dissections and/or rupture, or aneurysms/fistulae (54%), spontaneous splenic rupture (18%) and bowel perforation (14%). The mean age of the first complication among the males was 23,1 years and among the females 31,4. COLLIII showed reduced levels and/or abnormal migration. In all patients a COL3A1 causal mutation was disclosed, 7 known and 15 novel, i.e., 12 missense, affecting a glycine in the COLLIII collagenous domain and 3 splice-site mutations, leading to in-frame exon skipping. In a patient, in addition to the causal missense mutation, the novel p.P517L substitution was identified, likely modulating the phenotype.
This first report on the vEDS Italian patients adds insights to the knowledge of this disorder at clinical and molecular level.