

An International Network for Recurrent and Familial Forms of Hemolytic Uremic Syndrome and Thrombotic Thrombocytopenic Purpura

Research networks

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Hemolytic Uremic Syndrome (HUS) and Thrombotic Thrombocytopenic Purpura (TTP) are two closely related rare entities (HUS with an incidence of 2.5:10.000 cases/year and TTP with 3.7:1.000.000 cases/year) characterised by microangiopathic haemolytic anaemia and thrombocytopenia with renal and cerebral involvement. Even more rare subsets of these diseases, called atypical HUS/TTP, often occur in families. These forms are characterised by frequent relapses after recovery of the first episode leading to permanent renal and neurological sequelae. In 1996 an International Registry of Recurrent and Familial HUS/TTP was established under the coordination of the Clinical Research Center for Rare Diseases Aldo e Cele Daccò, (Bergamo, Italy). The Registry is a network comprised of 154 Haematology and Nephrology Units from 12 European countries (Italy, Belgium, Czech Republic, Denmark, Estonia, Germany, Greece, Portugal, Serbia, Spain, Switzerland, United Kingdom), with the contribution of few extra-European centres.

The aim of this project is to improve the knowledge on the pathogenesis of these disorders and in particular to identify genetic and biochemical factors predisposing to the development of recurrent, atypical forms of HUS /TTP and also to provide genetic counselling and help in the clinical management.

Clinical and laboratory data of all patients are collected by a uniform data extraction form. The family history and personal data of the unaffected relatives are also collected. Biological samples are collected from all patients and available relatives, for the biochemical and genetic analyses. The Registry database contains data of more than 400 cases of HUS/TTP, one of the largest collections of data in the literature.

Some genetically determined forms of HUS/TTP, associated to defects of complement regulatory factors, or congenital deficiency of ADAMTS13 enzyme, have been identified and have allowed establishing some genotype/phenotype correlations in the analysed HUS/TTP patients referred to the Registry.

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