## *Pubblicazioni del Registro con i contributi dei Centri partecipanti*

An *ex vivo* test to investigate genetic factors conferring susceptibility to atypical haemolytic uremic syndrome

Gastoldi S, S, Aiello S, Galbusera M, Breno M, Alberti M, Bresin E, Mele C, Piras R, Liguori L, Santarsiero D, Benigni A, Remuzzi G, Noris M.

Front Immunol. 2023 Feb 9;14:1112257.

Molecular Studies and an *ex vivo* Complement Assay on Endothelium Highlight the Genetic Complexity of Atypical Hemolytic Uremic Syndrome: The Case of a Pedigree

Piras R, Iatropoulos P, Bresin E, Todeschini M, Gastoldi S, Valoti E, Alberti M, Mele C, Galbusera M, Cuccarolo P, Benigni A, Remuzzi G, Noris M.

With a Null CD46 Variant. Front Med (Lausanne). 2020 Nov 3;7:579418.

An *ex vivo* test of Complement Activation on Endothelium for Individualized Eculizumab Therapy in Hemolytic Uremic Syndrome

Galbusera M, Noris M, Gastoldi S, Bresin E, Mele C, Breno M, Cuccarolo P, Alberti M, Valoti E, Piras R, Donadelli R, Vivarelli M, Murer L, Pecoraro C, Ferrari E, Perna A, Benigni A, Portalupi V, Remuzzi G.

Am J Kidney Dis. 2019 Jul;74(1):56-72.

ADAMTS13 Secretion and Residual Activity among Patients with Congenital Thrombotic Thrombocytopenic Purpura with and without Renal Impairment

Rurali E, Banterla F, Donadelli R, Bresin E, Galbusera M, Gastoldi S, Peyvandi F, Underwood M, Remuzzi G, Noris M.

Clin J Am Soc Nephrol. 2015 Nov 6;10(11):2002-12.

Dynamics of complement activation in aHUS and how to monitor eculizumab therapy.

Noris M, Galbusera M, Gastoldi S, Macor P, Banterla F, Bresin E, Tripodo C, Bettoni S, Donadelli R, Valoti E, Tedesco F, Amore A, Coppo R, Ruggenenti P, Gotti E, Remuzzi G.

Blood. 2014 Sep 11;124(11):1715-26. doi: 10.1182/blood-2014-02-558296. Epub 2014 Jul 18.

PMID: 25037630 [PubMed - indexed for MEDLINE]

Combined complement gene mutations in atypical hemolytic uremic syndrome influence clinical phenotype.

Bresin E, Rurali E, Caprioli J, Sanchez-Corral P, Fremeaux-Bacchi V, Rodriguez de Cordoba S, Pinto S, Goodship TH, Alberti M, Ribes D, Valoti E, Remuzzi G, Noris M; European Working Party on Complement Genetics in Renal Diseases.

J Am Soc Nephrol. 2013 Feb;24(3):475-86. doi: 10.1681/ASN.2012090884. Epub 2013 Feb 21.

PMID: 23431077 [PubMed - indexed for MEDLINE]

Relative role of genetic complement abnormalities in sporadic and familial aHUS and their impact on clinical phenotype.

Noris M, Caprioli J, Bresin E, Mossali C, Pianetti G, Gamba S, Daina E, Fenili C, Castelletti F, Sorosina A, Piras R, Donadelli R, Maranta R, van der Meer I, Conway EM, Zipfel PF, Goodship TH, Remuzzi G.

Clin J Am Soc Nephrol. 2010 Oct;5(10):1844-59. Epub 2010 Jul 1.

PMID: 20595690 [PubMed - indexed for MEDLINE]

Thrombomodulin mutations in atypical hemolytic-uremic syndrome.

Delvaeye M, Noris M, De Vriese A, Esmon CT, Esmon NL, Ferrell G, Del-Favero J, Plaisance S, Claes B, Lambrechts D, Zoja C, Remuzzi G, Conway EM.

N Engl J Med. 2009 Jul 23;361(4):345-57.

PMID: 19625716 [PubMed - indexed for MEDLINE]

Outcome of renal transplantation in patients with non-Shiga toxin-associated hemolytic uremic syndrome: prognostic significance of genetic background.

Bresin E, Daina E, Noris M, Castelletti F, Stefanov R, Hill P, Goodship TH, Remuzzi G; International Registry of Recurrent and Familial HUS/TTP.

Clin J Am Soc Nephrol. 2006 Jan;1(1):88-99. Epub 2005 Nov 2. Review.

PMID: 17699195 [PubMed - indexed for MEDLINE]

Genetics of HUS: the impact of MCP, CFH, and IF mutations on clinical presentation, response to treatment, and outcome.

Caprioli J, Noris M, Brioschi S, Pianetti G, Castelletti F, Bettinaglio P, Mele C, Bresin E, Cassis L, Gamba S, Porrati F, Bucchioni S, Monteferrante G, Fang CJ, Liszewski MK, Kavanagh D, Atkinson JP, Remuzzi G; International Registry of Recurrent and Familial HUS/TTP.

Blood. 2006 Aug 15;108(4):1267-79. Epub 2006 Apr 18.

PMID: 16621965 [PubMed - indexed for MEDLINE]

In-vitro and in-vivo consequences of mutations in the von Willebrand factor cleaving protease ADAMTS13 in thrombotic thrombocytopenic purpura.

Donadelli R, Banterla F, Galbusera M, Capoferri C, Bucchioni S, Gastoldi S, Nosari S, Monteferrante G, Ruggeri ZM, Bresin E, Scheiflinger F, Rossi E, Martinez C, Coppo R, Remuzzi G, Noris M; International Registry of Recurrent and Familial HUS/TTP.

Thromb Haemost. 2006 Oct;96(4):454-64.

PMID: 17003922 [PubMed - indexed for MEDLINE]

Complement factor H mutation in familial thrombotic thrombocytopenic purpura with ADAMTS13 deficiency and renal involvement.

Noris M, Bucchioni S, Galbusera M, Donadelli R, Bresin E, Castelletti F, Caprioli J, Brioschi S, Scheiflinger F, Remuzzi G; International Registry of Recurrent and Familial HUS/TTP.

J Am Soc Nephrol. 2005 May;16(5):1177-83. Epub 2005 Mar 30.

PMID: 15800115 [PubMed - indexed for MEDLINE]

Complement factor H mutations and gene polymorphisms in haemolytic uraemic syndrome: the C257T, the A2089G and the G2881T polymorphisms are strongly associated with the disease.

Caprioli J, Castelletti F, Bucchioni S, Bettinaglio P, Bresin E, Pianetti G, Gamba S, Brioschi S, Daina E, Remuzzi G, Noris M; International Registry of Recurrent and Familial HUS/TTP.

Hum Mol Genet. 2003 Dec 15;12(24):3385-95. Epub 2003 Oct 28.

PMID: 14583443 [PubMed - indexed for MEDLINE]

Familial haemolytic uraemic syndrome and an MCP mutation.

Noris M, Brioschi S, Caprioli J, Todeschini M, Bresin E, Porrati F, Gamba S, Remuzzi G; International Registry of Recurrent and Familial HUS/TTP.

Lancet. 2003 Nov 8;362(9395):1542-7.

PMID: 14615110 [PubMed - indexed for MEDLINE]

von Willebrand factor cleaving protease (ADAMTS13) is deficient in recurrent and familial thrombotic thrombocytopenic purpura and hemolytic uremic syndrome.

Remuzzi G, Galbusera M, Noris M, Canciani MT, Daina E, Bresin E, Contaretti S, Caprioli J, Gamba S, Ruggenenti P, Perico N, Mannucci PM; Italian Registry of Recurrent and Familial HUS/TTP.

Blood ,2002 Aug 1;100(3):778-85

PMID: 12130486 [PubMed - indexed for MEDLINE]