

Eleonora GAMBINERI

PUBBLICAZIONI

1. Azzari C, Resti M, Moriondo M, **Gambineri E**, Rossi ME, Novembre E, Vierucci A. Lack of transmission of TTV through immunoglobulins. *Transfusion* 2001 Dec;41(12):1505-8.
2. **Gambineri E**, Torgerson RT and Ochs HD, Immune dysregulation, polyendocrinopathy, enteropathy, and X-linked inheritance (IPEX), a syndrome of systemic autoimmunity caused by mutations of FOXP3, a critical regulator of T-cell homeostasis. *Curr Opin Rheumatol*. 2003 Jul;15(4):430-5. Review.
3. Lee WI, Zhu Q, **Gambineri E**, Jin Y, Welcher AA and Ochs HD. Inducible CO-Stimulator molecule (ICOS), a candidate gene for defective isotope switching, is normal in patients with hyper IgM syndrome of unknown molecular diagnosis. *J Allergy Clin Immunol*. 2003 Nov;112(5):958-64.
4. Azzari C, **Gambineri E**, Resti M, Moriondo M, Betti L, Saldias LR, G Gelli AM, Vierucci A. Safety and immunogenicity of measles-mumps-rubella vaccine in children with congenital immunodeficiency (DiGeorge syndrome). *Vaccine*. 2005 Feb 25;23(14):1668-71.
5. Mazzolari E, Forino C, Fontana M, D'Ippolito C, Lanfranchi A, **Gambineri E**, Ochs H, Badolato R, Notarangelo LD. A new case of IPEX receiving bone marrow transplantation. *Bone Marrow Transplant*, 2005; 35(10):1033-4.
6. Notarangelo LD, **Gambineri E**, Badolato R. Immunodeficiencies with autoimmune consequences. *Adv Immunol*. 2006;89:321-70. Review.
7. Bacchetta R, Passerini L, **Gambineri E**, Dai M, Allan SE, Sartirana C, Azzari C, Ziegler S, Levings MK, and Roncarolo MG. Defective regulatory and effector t cell functions in patients with foxp3 mutations. *JCI* 2006 Jun;116(6):1713-22.
8. Torgerson TR, Linane A, Moes N, Anover S, Mateo V, Rieux-Laucat F, Hermine O, Vijay S, **Gambineri E**, Cerf-Bensussan N, Fischer A, Ochs HD, Goulet O, Ruemmele FM. Severe food allergy as a variant of IPEX syndrome caused by a deletion in a noncoding region of the FOXP3 gene. *Gastroenterology*. 2007 May;132(5):1705-17.

9. Bacchetta R, **Gambineri E**, Roncarolo MG. Role of regulatory T cells and FOXP3 in human diseases. *J Allergy Clin Immunol*. 2007 Aug;120(2):227-35; quiz 236-7. Review.
10. Ochs HD, **Gambineri E**, Torgerson TR. IPEX, FOXP3 and regulatory T-cells: a model for autoimmunity. *Immunol Res*. 2007 Jul;38(1-3):112-121.
11. Azzari C, Moriondo M, Indolfi G, Betti L, **Gambineri E**, de Martino M, Resti M. Higher risk of hepatitis C virus perinatal transmission from drug user mothers is mediated by peripheral blood mononuclear cell infection. *J Med Virol*. 2008 Jan;80(1):65-71.
12. de Beaucoudrey L, Puel A, Filipe-Santos O, Cobat A, Ghandil P, Chrabieh M, Feinberg J, von Bernuth H, Samarina A, Janni  re L, Fieschi C, St  phan JL, Boileau C, Lyonnet S, Jondeau G, Cormier-Daire V, Le Merrer M, Hoarau C, Lebranchu Y, Lortholary O, Chandesris MO, Tron F, **Gambineri E**, Bianchi L, Rodriguez-Gallego C, Zitnik SE, Vasconcelos J, Guedes M, Vitor AB, Marodi L, Chapel H, Reid B, Roifman C, Nadal D, Reichenbach J, Caragol I, Garty BZ, Dogu F, Camcioglu Y, G  lle S, Sanal O, Fischer A, Abel L, Stockinger B, Picard C, Casanova JL. Mutations in STAT3 and IL12RB1 impair the development of human IL-17-producing T cells. *J Exp Med*. 2008 Jun 30.
13. **Gambineri E**, Perroni L, Passerini L, Bianchi L, Doglioni C, Meschi F, Bonfanti R, Sznajer Y, Tommasini A, Lawitschka A, Junker A, Dunstheimer D, Heidemann PH, Cazzola G, Cipolli M, Friedrich W, Janic D, Azzi N, Richmond E, Vignola S, Barabino A, Chiumello G, Azzari C, Roncarolo MG, Bacchetta R. Clinical and molecular profile of a new series of IPEX patients: inconsistent correlation between FOXP3 protein expression and disease severity *J Allergy Clin Immunol* 2008 Dec;122(6):1105-1112.
14. Olivito B, Simonini G, Ciullini S, Moriondo M, Betti L, **Gambineri E**, Cantarini L, DE Martino M, Azzari C, Cimaz R. Th17 Transcription Factor RORC2 Is Inversely Correlated with FOXP3 Expression in the Joints of Children with Juvenile Idiopathic Arthritis. *J Rheumatol*. 2009 Sep;36(9):2017-24.
15. Violi F, Sanguigni V, Carnevale R, Plebani A, Rossi P, Finocchi A, Pignata C, De Mattia D, Martire B, Pietrogrande MC, Martino S, **Gambineri E**, Soresina AR, Pignatelli P, Martino F, Basili S, Loffredo L. Hereditary deficiency of gp91(phox) is associated with enhanced arterial dilatation: results of a multicenter study. *Circulation*. 2009 Oct 20;120(16):1616-22.
16. Colarusso, G., Gambineri, E., Lapi, E., Casini, T., Tucci, F., Lippi, F., Azzari, C. Evans syndrome and antibody deficiency: An atypical presentation of chromosome 22q11.2 deletion syndrome (2010) *Pediatric Reports*, 2 (2), pp. 42-43

17. Stagi S, Lapi E, **Gambineri E**, Salti R, Genuardi M, Colarusso G, Conti C, Jenuso R, Chiarelli F, Azzari C, de Martino M. Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11)). Clin Endocrinol (Oxf). 2010 Jun;72(6):839-44.
18. Stagi S, Lapi E, **Gambineri E**, Manoni C, Genuardi M, Colarusso G, Conti C, Chiarelli F, de Martino M, Azzari C. Bone density and metabolism in subjects with microdeletion of chromosome 22q11 (del22q11). Eur J Endocrinol. 2010 Aug;163(2):329-37. Epub 2010 Jun 1.
19. Olivito B, Taddio A, Simonini G, Massai C, Ciullini S, **Gambineri E**, de Martino M, Azzari C, Cimaz R. Defective FOXP3 expression in patients with acute Kawasaki disease and restoration by intravenous immunoglobulin therapy. Clin Exp Rheumatol. 2010 Jan-Feb;28(1 Suppl 57):93-7.
20. Tsuda M, Torgerson TR, Selmi C, **Gambineri E**, Carneiro-Sampaio M, Mannurita SC, Leung PS, Norman GL, Gershwin ME. The spectrum of autoantibodies in IPEX syndrome is broad and includes anti-mitochondrial autoantibodies. J Autoimmun. 2010 Nov;35(3):265-8.
21. McMurchy AN, Gillies J, Allan SE, Passerini L, **Gambineri E**, Roncarolo MG, Bacchetta R, Levings MK. Point mutants of forkhead box P3 that cause immune dysregulation, polyendocrinopathy, enteropathy, X-linked have diverse abilities to reprogram T cells into regulatory T cells. J Allergy Clin Immunol. 2010 Dec;126(6):1242-51.
22. Passerini L, Di Nunzio S, Gregori S, **Gambineri E**, Cecconi M, Seidel MG, Cazzola G, Perroni L, Tommasini A, Vignola S, Guidi L, Roncarolo MG, Bacchetta R. Functional type 1 regulatory T cells develop regardless of FOXP3 mutations in patients with IPEX syndrome. Eur J Immunol. 2011 Apr;41(4):1120-31.
23. Fioredda F, Calvillo M, Bonanomi S, Coliva T, Tucci F, Farruggia P, Pillon M, Martire B, Ghilardi R, Ramenghi U, Renga D, Menna G, Pusiol A, Barone A, **Gambineri E**, Palazzi G, Casazza G, Lanciotti M, Dufour C. Congenital and acquired neutropenias consensus guidelines on therapy and follow-up in childhood from the Neutropenia Committee of the Marrow Failure Syndrome Group of the AIEOP (Associazione Italiana Emato-Oncologia Pediatrica). Am J Hematol. 2011 Oct 31.

24. **Gambineri E**, Torgerson TR. Genetic disorders with immune dysregulation. *Cell Mol Life Sci*. 2012 Jan;69(1):49-58.
25. **Gambineri E**. New frontiers in primary immunodeficiency disorders: immunology and beyond. *Cell Mol Life Sci*. 2012 Jan;69(1):1-5.
26. Zennaro D, Scala E, Pomponi D, Caprini E, Arcelli D, **Gambineri E**, Russo G, Mari A. Proteomics plus genomics approaches in primary immunodeficiency: the case of immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome. *Clin Exp Immunol*. 2012 Jan;167(1):120-8.
27. Barzaghi F, Passerini L, **Gambineri E**, Ciullini Mannurita S, Cornu T, Kang ES, Choe YH, Cancrini C, Corrente S, Ciccocioppo R, Cecconi M, Zuin G, Discepolo V, Sartirana C, Schmidtke J, Ikinogullari A, Ambrosi A, Roncarolo MG, Olek S, Bacchetta R. Demethylation analysis of the FOXP3 locus shows quantitative defects of regulatory T cells in IPEX-like syndrome. *J Autoimmun*. 2012 Feb;38(1):49-58.
28. de Vries E; European Society for Immunodeficiencies (ESID) members. Patient-centred screening for primary immunodeficiency, a multi-stage diagnostic protocol designed for non-immunologists: 2011 update. *Clin Exp Immunol*. 2012 Jan;167(1):108-19.
29. Magini P, Monica MD, Uzielli ML, Mongelli P, Scarselli G, **Gambineri E**, Scarano G, Seri M. Two novel patients with Bohring-Opitz syndrome caused by de novo ASXL1 mutations. *Am J Med Genet A*. 2012 Apr;158A(4):917-21.
30. Fernandes JF, Rocha V, Labopin M, Neven B, Moshous D, Gennery AR, Friedrich W, Porta F, Diaz de Heredia C, Wall D, Bertrand Y, Veys P, Slatter M, Schulz A, Chan KW, Grimley M, Ayas M, Gungor T, Ebell W, Bonfim C, Kalwak K, Taupin P, Blanche S, Gaspar HB, Landais P, Fischer A, Gluckman E, Cavazzana-Calvo M; **Eurocord and Inborn Errors Working Party of European Group for Blood and Marrow Transplantation**. Transplantation in patients with SCID: mismatched related stem cells or unrelated cord blood? *Blood*. 2012 Mar 22;119(12):2949-55.
31. Uzel G, Sampaio EP, Lawrence MG, Hsu AP, Hackett M, Dorsey MJ, Noel RJ, Verbsky JW, Freeman AF, Janssen E, Bonilla FA, Pechacek J, Chandrasekaran P, Browne SK, Agharahami A, Gharib AM, Mannurita SC, Yim JJ, **Gambineri E**, Torgerson T, Tran DQ, Milner JD, Holland SM. Dominant gain-of-function STAT1 mutations in FOXP3 wild-type immune dysregulation-polyendocrinopathy-enteropathy-X-linked-like syndrome. *J Allergy Clin Immunol* 2013, 131 (6): 1611-1623

32. Goudy K, Aydin D, Barzaghi F, Gambineri E, Vignoli M, Ciullini Mannurita S, Doglioni C, Ponzoni M, Cicalese MP, Assanelli A, Tommasini A, Brigida I, Dellepiane RM, Martino S, Olek S, Aiuti A, Ciceri F, Roncarolo MG, Bacchetta R. Human IL2RA null mutation mediates immunodeficiency with lymphoproliferation and autoimmunity. *Clin Immunol*. 2013 Mar;146(3):248-61. Epub 2013 Jan 24.
33. Loffredo L, Carnevale R, Sanguigni V, Plebani A, Rossi P, Pignata C, De Mattia D, Finocchi A, Martire B, Pietrogrande MC, Martino S, Gambineri E, Giardino G, Soresina AR, Martino F, Pignatelli P, Violi F. Does NADPH oxidase deficiency cause artery dilatation in humans? *Antioxid Redox Signal*. 2013 Apr 20;18(12):1491-6.
34. Ciullini Mannurita, S., Vignoli, M., Bianchi, L., Kondi, A., Gerloni, V., Breda, L., ten Cate, R., Alessio, M., Ravelli, A., Falcini, F., Gambineri, E. CACP syndrome: identification of five novel mutations and of the first case of UPD in the largest European cohort (2013) *European Journal of Human Genetics*, Article in Press.
35. Nademi Z, Slatter M, Gambineri E, Mannurita SC, Barge D, Hodges S, Bunn S, Thomas J, Haugk B, Hambleton S, Flood T, Cant A, Abinun M, Gennery A. Single centre experience of haematopoietic SCT for patients with immunodysregulation, polyendocrinopathy, enteropathy, X-linked syndrome. *Bone Marrow Transplant*. 2013 Nov 25.
36. Dickinson RE, Milne P, Jardine L, Zandi S, Swierczek SI, McGovern N, Cookson S, Ferozepurwalla Z, Langridge A, Pagan S, Gennery A, Heiskanen-Kosma T, Hämäläinen S, Seppänen M, Helbert M, Tholouli E, Gambineri E, Reykdal S, Gottfreðsson M, Thaventhiran JE, Morris E, Hirschfield G, Richter AG, Jolles S, Bacon CM, Hambleton S, Haniffa M, Bryceson Y, Allen C, Prchal JT, Dick JE, Bigley V, Collin M. The evolution of cellular deficiency in GATA2 mutation. *Blood*. 2014 Feb 6;123(6):863-74.
37. Moschese V, Martire B, Soresina A, Chini L, Graziani S, Monteferrario E, Bacchetta R, Cancrini C, Fiorilli M, Gambineri E, Pession A, Pignata C, Quinti I, Rondelli R, Rossi P, Ugazio AG, Plebani A, Pietrogrande MC. Anti-infective prophylaxis for primary immunodeficiencies: what is done in Italian Primary Immunodeficiency Network centers (IPINet) and review of the literature. *J Biol Regul Homeost Agents*. 2013 Oct-Dec;27(4):935-46. Review.
38. Routes J, Abinun M, Al-Herz W, Bustamante J, Condino-Neto A, De La Morena MT, Etzioni A, Gambineri E, Haddad E, Kobrynski L, Le Deist F, Nonoyama S, Oliveira JB, Perez E, Picard C, Rezaei N, Sleasman J, Sullivan KE, Torgerson T. ICON: the early

diagnosis of congenital immunodeficiencies. *J Clin Immunol*. 2014 May;34(4):398-424. Review.

39. Sayar E, Uygun DF, Islek A, Hazar-Sayar E, Akkaya B, Vignoli M, Gambineri E, Yesilipek MA, Artan R. Langerhans cell histiocytosis in IPEX syndrome: possible role for natural regulatory T cells? *Pediatr Allergy Immunol*. 2014 Oct;25(6):601-3.

40. Cancrini C, Puliafito P, Digilio MC, Soresina A, Martino S, Rondelli R, Consolini R, Ruga EM, Cardinale F, Finocchi A, Romiti ML, Martire B, Bacchetta R, Albano V, Carotti A, Specchia F, Montin D, Cirillo E, Cocchi G, Trizzino A, Bossi G, Milanese O, Azzari C, Corsello G, Pignata C, Aiuti A, Pietrogrande MC, Marino B, Ugazio AG, Plebani A, Rossi P; Italian Network for Primary Immunodeficiencies. Clinical features and follow-up in patients with 22q11.2 deletion syndrome. *J Pediatr*. 2014 Jun;164(6):1475-80.

41. Zama D, Cocchi I, Masetti R, Specchia F, Alvisi P, Gambineri E, Lima M, Pession A. Late-onset of immunodysregulation, polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) with intractable diarrhea. *Ital J Pediatr*. 2014 Oct 18;40(1):68.

42. Gambineri E, Ciullini Mannurita S, Robertson H, Vignoli M, Haugk B, Lionetti P, Hambleton S, Barge D, Gennery AR, Slatter M, Nademi Z, Flood TJ, Jackson A, Abinun M, Cant AJ. Gut immune reconstitution in immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome after hematopoietic stem cell transplantation. *J Allergy Clin Immunol*. 2015 Jan;135(1):260-262.