Stefano Volpi, MD, PhD

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Genova 26/05/25

Track Record

Scientific output, retrieved on Scopus on 25/05/2025

Published articles: 136
Citations: 5853
h-index: 40

In my career I focused on the study of genetic causes of immune deficiency and on their disease mechanism through induced pluripotent stem cell in vitro models and animal models.
During my Residency and PhD (2007-2013) from the research I performed in Harvard, USA **I described as the first author a novel immunodeficiency** caused by mutation in *EXTL3* gene (JEM PMID 28148688). I contributed to the description of **3 other immunodeficiencies** (Cell PMID 29474921, Nature PMID 25307056 and Science PMID 25814066). I performed **basic science studies on primary immunodeficiencies using mouse models or iPSC** (Blood PMID 22302739; Nature Immunology Journal cover PMID 30127432; JACI as first author PMID 26409660; PNAS PMID 30154162; Frontiers in Immunology PMID 25101082).
I moved to Lausanne in 2014 where I set up from zero the new lab of my new PI and completed a study on an animal model as first author (Blood Journal cover PMID 26468226).
I returned in Italy in 2015, I focused on genetic diseases with immune dysregulation or autoinflammation and I **described as the first author and corresponding author a novel primary immunodeficiency** (JACI PMID 30771411). I co-shared the first authorship in two publication (JACI PMID 33556464 and 32437739) revealing the response to the anti-IL-1R drug anakinra in patients with COVID19 (authorized by EMA).
I was elected in 2020 as one of the eleven component (treasurer) of the executive board of the European Society of Immunodeficiency.
I obtained as a PI or collaborator over **700.000 euros** from competitive grants, allowing me to fund my own research since 2015. I supervised 5 post docs and 1 PhD student.
I was **invited for oral presentations** at the Mexican national conference of Immunology (2021), the Pediatric Rheumatology European Society biannual meeting (2021), the European Society of Immunodeficiency biannual meeting (2020), the National conference of the Italian Society of Human Genetics (2017) and the Annual symposium on Autoimmunity, Harvard Medical School, Boston (2012).

On type I interferonopathies, SAVI and disease modelling I described as the first author the response to JAK inhibitors in SAVI (JoCI PMID 31144250) and type I interferon activation in COPA (Clin Imm PMID 29030294). I contributed to the description of a novel disease (**Nature Communication** PMID 29259162). I collaborated with

two consortiums for disease characterization of SAVI and COPA (JACI PMID 33217613, JoCI PMID 29977900). As **last author I described activation of type I interferon in RALD** (JoCI PMID 34455097) and **collaborated to the characterization of a novel 3D lung model from iPSC with Dr Snoeck at Columbia University** (Nature Cell Biol PMID 28436965). I set up two international collaboration with Dr. Lo at Hong Kong and Dr Reizis at NYU, USA, to dissect disease mechanism in the type I interferonopathy DNase1L3, (JEM PMID 33783474, Am J Hum Genetics PMID 33022220) and with Dr Genovese at Harvard for gene editing in iPSC and HSC.
I recently won a grant as a collaborator for a project on gene editing in genetic diseases (PRIN 20175XHBPN) and a 1 million grant (FIS00002235).

Work experiences

Elected member of the management board of the European Society for Primary Immunodeficiencies (ESID) 2021-2024

Associate Professor **Mar 2024- to date**

Associate Professor at Genoa University, Genova

Pediatrician at the Pediatric Rheumatology Unit and the Center for Autoinflammatory Diseases, Gaslini Institute, Genova, Italy

Research Fellow (Ricercatore a tempo determinato RTDa) **Mar 2018- Feb 2024**

Research Fellow at Genoa University, Genova

Pediatrician at the Pediatric Rheumatology Unit and the Center for Autoinflammatory Diseases and Immunodeficiencies, Gaslini Institute, Genova, Italy

Research Fellow **Sept 2014-March 2018**

Research fellow at Gaslini Institute, Genova, Italy, in Dott. Gattorno's group, working on immune deficiencies and type 1 interferonopathies with both clinical and research responsibilities.

Collaborateur de Recherche **Sept 2014-Aug 2015**

Collaborateur de Recherche (post doctoral student) at Lausanne University Hospital, Lausanne, Switzerland, in Prof. Candotti's laboratory: complete setup of a new laboratory, supervision of a PhD student and development of a new project on the diagnosis of Type I interferonopathies. Study of immune disregulation in a mouse model of Wiskott-Aldrich syndrome.

Research Fellow **Oct 2011- July 2014**

Post doctoral research fellow at Children's Hospital Boston, Harvard Medical School, in Prof. Notarangelo's laboratory of Paediatrics immunology, focusing on the study of primary immunodeficiencies through a mouse model of Wiskott Aldrich Syndrome, a zebrafish model of a novel immunodeficiency due to *EXTL3* gene mutation in collaboration with Prof. Zon at Boston Children's Hospital and the generation of iPS cell lines from patients with genetic susceptibility to Herpes Simplex encephalitis, in collaboration with Prof. Casanova at Rockefeller University.

Research Fellow **Sept 2007- Apr 2008**

Research fellow at the Institute for Research in Biomedicine, Bellinzona, Switzerland, under supervision of Prof. Fabio Grassi studying a mouse model of systemic lupus erythematosus.

Research Intern **Sept 2006- Aug 2007**

Research intern at CEBR, Centre of Excellence for Biomedical Research, Genova, under supervision of Dott. Elisabetta Traggiai studying the effect of mesenchymal stem cells

Clerkship **Aug 2006**

Clerkship at Korle Bu University Hospital, Accra, Ghana, at the Paediatric Department, under supervision of Dott. Onike Rodriguez.

Intern **Apr 2004- Mar 2005**

Research intern at the Laboratory of Experimental Oncology, Utrecht University, The Netherlands and the Hubrecht laboratory for developmental biology under supervision of Prof. Rachel Giles and Prof. Emile Voest working on a zebrafish model of angiogenesis.

Education and clinical certificates

Knowledge of Good Clinical Practice (GCP) online course 13/03/2024

Experiences in clinical trials

IVIG in hypogammaglobulinemia (site PI) **Mar 2021**

Post Doctoral Student **Sept 2014 – Aug 2015**

Lausanne University, Lausanne University Hospital, Switzerland.

Post Doctoral Student **Jan 2011 – Jul 2013**

Harvard Medical School, Boston Children's Hospital, USA.

PhD **Jan 2013 – Dec 2015**

PhD student in Genetics, Genoa University, Italy. Thesis: Identification and characterization of novel primary immunodeficiencies: *EXTL3* gene mutation causes a novel form of Immuno-Osseus dysplasia and unveils a critical role of heparan sulfate in thymopoiesis.

Residency **2007 - 2012**

Specialization *cum laude* in Pediatrics, Gaslini Children's Hospital, Genoa University, Italy.

Undergraduate School **1999-2006**

MD certificate *cum laude*, with honor, Genoa University Medical School, Italy.

Prizes

Bright spark in immunology **September 2015**

European Federation of Immunological Societies, European conference of Immunology 2015.

Grants

Fondo Italiano per la Scienza **Dec 2023-2028**
Combining gene editing and iPS to study and treat monogenic autoinflammatory diseases FIS00002235 953378 euro.

5 per mille Istituto Giannina Gaslini **June 2022-2025**
GASLINI genome-editing lab: development of an advanced cellular platform for the study of genetic diseases, 230000 euro.

Curiosity Driven Research Grant, Genova University **February 2021 – January 2023**
Combining CRISPR/Cas9 gene editing and induced pluripotent stem cells (iPS) to study and treat genetic diseases of the immune system, 65000 euro

Italian Health Ministry "Ricerca Finalizzata" (n. GR-2019-12369050) **February 2021 – January 2024**
Innovative pre-transplant conditionings to preserve bone marrow niche and improve the immunological reconstitution in immune dysregulation disorders, 80000 euro.

PRIN (n.20175XHBPN) Italian Research and University Ministry **Agosto 2019 – Agosto 2022**
Advanced genetic engineering to study and treat monogenic diseases, 140000 euro.

Italian Foreign Affairs Ministry (n. GR-2019-12369050) **February 2017 – January 2020**
Genetic basis of early-onset systemic lupus erythematosus in India and Italy, 146000 euro.

Oral presentations and Conference organization

Oral presentation **April 2025**

International congress of Familial Mediterranean Fever and Systemic Autoinflammatory Disease, ISSAID, (Paris, France).
Systemic inflammation, lymphoproliferation and vasculopathy in a patient with ARHGAP10 mutation

Invited speaker **June 2023**

Inborn error working party simposyom of the European Society for Bone Marrow transplantation (Brescia, Italy).
HSCT in ARPC1B and actinopathies

Invited speaker **October 2023**

European Society for Immunodeficiencies, biannual conference (Goteborg, Sweden)
Extinguishing the Fire: Inflammation Gone Wrong

Invited speaker **October 2021**

European Society for Immunodeficiencies, biannual conference (online)
ARPC1B related diseases

Invited speaker **February 2020**

Preceptorship on Autoinflammatory syndromes (Genova, Italy)
Interferonopathies

Oral presentation **June 2019**

International congress of Familial Mediterranean Fever and Systemic Autoinflammatory Disease, ISSAID, (Genova, Italy)
A combined immunodeficiency with severe infections, inflammation and allergy caused by ARPC1B deficiency

Invited speaker **November 2019**

Maternal and fetal infections (Genova, Italy)
The immunology of pregnancy, of the fetus and of the newborn

Invited speaker **October 2018**

Lupus 2018 (Florence, Italy)
Forme monogeniche di SLE.

Oral presentation **June 2018**

Federation of Clinical Immunology Society (FOCIS) meeting (San Francisco, USA)
A combined immunodeficiency with severe infections, inflammation and allergy caused by ARPC1B deficiency

Invited speaker **November 2017**

National conference of the Italian Society of Human Genetics (SIGU) (Napoli, Italy)
Genetic mechanisms in inflammatory diseases

Invited speaker **October 2017**

La sindrome di Aicardi-Goutieres: un modello di interferonopatia (Brescia, Italy)
Interferonopatie di tipo 1 in reumatologia pediatrica: l'approccio diagnostico.

Oral presentation **September 2017**

Pediatric Rheumatology European Society (PRES) annual meeting (Genova, Italy)
A severe case of polyarticular arthritis caused by mutation of the COPA gene.

Invited speaker **May 2017**

Preceptorship on autoinflammatory syndromes (Genova, Italy)
Immunodeficiencies

Eposter presentation **March 2017**

Clinical immunology society meeting (CIS) (Seattle, USA)
Efficacy of the JAK inhibitor ruxolitinib in two patients with SAVI syndrome

Oral presentation **Sept 2016**

Pediatric Rheumatology European Society (PRES) meeting (Genova, Italy)

Type 1 interferonopathies: diagnostic role of peripheral blood interferon signature and preliminary results of treatment with a JAK 1/2 inhibitor.

Oral presentation **April 2016**

Clinical Immunology Society (CIS) (Boston USA)

1) Blood Interferon signature as a screening for Type I Interferonopathies in children with early-onset SLE and vasculopathy.

2) Exostosin-like glycosyl transferase 3 (EXTL3) gene mutation causes a novel form of immuno-osseous dysplasia and unveils a critical role of heparan sulfate in thymopoiesis

Oral presentation **October 2015**

International Congress of Familial Mediterranean Fever and Systemic Autoinflammatory Diseases, ISSAID (Dresden, Germany)

"Identification of type I interferonopathies using blood interferon signature: the experience of a pediatric rheumatology center".

Oral presentation **September 2015**

EUtrain-Translational Research in Rheumatology Conference (Genova, Italy)

"Identification of type I interferonopathies using blood interferon signature: the experience of a pediatric rheumatology center".

Oral presentation **Sept 2015**

Research day of Lausanne University Hospital (Lausanne, Switzerland)

"Selective deficiency of WASP in Treg cells is sufficient to cause autoimmunity in mice".

Oral presentations **Sept 2015**

European Conference of Immunology (Wien, Austria)

1) "Exostosin-like glycosyl transferase 3 (*EXTL3*) gene mutation causes a novel form of immuno-osseous dysplasia and unveils a critical role of heparan sulfate in thymopoiesis"

2) Selective deficiency of WASP in Treg cells is sufficient to cause autoimmunity in mice.

Oral presentation **June 2015**

Annual European Congress of Rheumatology (EULAR), (Rome, Italy)

"Blood Interferon signature as a screening for Type I Interferonopathies in children with early-onset SLE and vasculopathy".

Oral presentation **May 2014**

Annual meeting of the Italian Paediatric Haematology and Oncology association (Genova, Italy)

"Role of Wasp and N-Wasp in b cell maturation, homing and function"

Invited speaker **Oct 2012**

Annual symposium on Autoimmunity, Beth Israel Medical Centre, Harvard Medical School (Boston, USA) "*The autoimmunity of the Wiskott-Aldrich syndrome*"

Oral presentation **May 2012**

PRES course "Translational and Clinical issues in pediatric rheumatology" (Genova, Italy)

- B cell intrinsic deficiency of the Wiskott-Adrich syndrome protein (WASP) causes severe abnormalities of the peripheral B cell compartment in mice
- T cell activation dependent purinergic signalling in the pathogenesis of experimental lupus glomerulonephritis

Scientific meeting organization **Mar 2018**

Organizing committee for the 2019 meeting of the International Society of Systemic Autoinflammatory Diseases (ISSAID) (Genoa, Italy).

Scientific meeting organization **Dec 2010**

Scientific committee member of the Meeting on mithocondrial diseases (Gaslini Institute, Genoa, Italy).

Scientific association membership and reviewer activity

Member of the European Society for Immune Deficiencies (ESID)

Member of the Pediatric Rheumatology European Society (PRES)

Review Editor for the journal "Frontiers in Medicine" and "Frontiers in Immunology".

Reviewer for "Frontiers in Immunology", "Journal of Allergy and Clinical Immunology", "Clinical Immunology", "Journal of Clinical Immunology", "Clinical Cytometry", "Current Medicinal Chemistry", "Pediatric Rheumatology".

Accreditation for animal experimentation "Felasa EU functions ACD".

Publications(1-136)

1. Woodward BL, Lahiri S, Chauhan AS, Garcia MR, Goodley LE, Clarke TL, et al. Inherited deficiency of DIAPH1 identifies a DNA double strand break repair pathway regulated by γ-actin. Nature communications. 2025;16(1).
2. Tesser A, Bocca P, Ulivi M, Pin A, Pastorino C, Cangelosi D, et al. Type I interferon signature: a quantitative standardized method for clinical application. Clinical and Experimental Immunology. 2025;219(1).
3. Simchoni N, Koide S, Likhite M, Kuchitsu Y, Kadirvel S, Law CS, et al. The common HAQ STING allele prevents clinical penetrance of COPA syndrome. The Journal of experimental medicine. 2025;222(4).
4. Palmeri S, Ferro J, Natoli V, Matucci-Cerinic C, Papa R, Rosina S, et al. Efficacy of High-Dose Intravenous Anakinra in Pediatric TAFRO Syndrome: Report of Two Cases and Literature Review. Pediatric Blood and Cancer. 2025.
5. Natoli V, Palmeri S, Rebollo-Giménez AI, Matucci-Cerinic C, Bocca P, Caorsi R, et al. Successful treatment of an anti-MDA5 antibody-positive Juvenile Dermatomyositis patient with refractory interstitial lung disease using tofacitinib. Pediatric Rheumatology. 2025;23(1).
6. Hadjadj J, Wolfers A, Borisov O, Hazard D, Leahy R, Jeanpierre M, et al. Clinical manifestations, disease penetrance, and treatment in individuals with SOCS1 insufficiency: a registry-based and population-based study. The Lancet Rheumatology. 2025;7(6):e391-e402.
7. Drago E, Fioredda F, Penco F, Prigione I, Bertoni A, Del Zotto G, et al. Inborn Error of WAS Presenting with SARS-CoV-2-Related Multisystem Inflammatory Syndrome in Children. Journal of clinical immunology. 2025;45(1).
8. Drago E, Bertoni A, Grossi A, Damasio MB, Anfigeno L, Miano M, et al. Majeed syndrome: first description in a patient of central-European ancestry. Rheumatology. 2025;64(5):3069-73.
9. Chan RWY, Serpas L, Ni M, Volpi S, Hiraki LT, Tam LS, et al. Erratum: Plasma DNA Profile Associated with DNASE1L3 Gene Mutations: Clinical Observations, Relationships to Nuclease Substrate Preference, and In Vivo Correction (The American Journal of Human Genetics (2020) 107(5) (882–894), (S0002929720303268), (10.1016/j.ajhg.2020.09.006)). American Journal of Human Genetics. 2025;112(5):1247.
10. Caorsi R, Bertoni A, Matucci-Cerinic C, Natoli V, Palmeri S, Rosina S, et al. Long-term efficacy of MAS825, a bispecific anti-IL1β and IL-18 monoclonal antibody, in two patients with systemic JIA and recurrent episodes of macrophage activation syndrome. Rheumatology. 2025;64(3):1528-33.
11. Al-Mayouf SM, Hadef D, Aljaberi N, Movahedi N, AlEed A, Almutairi A, et al. A proposed clinical tool to identify high-risk patients for monogenic lupus: a pilot study. Clinical and Experimental Rheumatology. 2025;43(3):538-44.
12. Volpi S, Angelotti ML, Palazzini G, Antonelli G, Ravaglia F, Garibotto F, et al. Lupus Nephritis Patterns and Response to Type I Interferon in Patients With DNASE1L3 Variants: Report of Three Cases. American Journal of Kidney Diseases. 2024;84(6):791-7.
13. Papa R, Caorsi R, Volpi S, Gattorno M. Expert Perspective: Diagnostic Approach to the Autoinflammatory Diseases. Arthritis and Rheumatology. 2024;76(2):166-77.
14. Palmeri S, Penco F, Bertoni A, Bustaffa M, Matucci-Cerinic C, Papa R, et al. Pyrin Inflammasome Activation Defines Colchicine-Responsive SURF Patients from FMF and Other Recurrent Fevers. Journal of clinical immunology. 2024;44(2).
15. Orsi SM, Pepino C, Rossoni L, Serafino M, Caorsi R, Volpi S, et al. Corrigendum: Case report: Multisystem inflammatory syndrome in children with associated proximal tubular injury(Front. Nephrol., (2023), 3, (1194989), 10.3389/fneph.2023.1194989). Frontiers in Nephrology. 2024;4.
16. Naviglio S, Cicalese MP, Rivers E, Ferrua F, Bonfim C, Cenciarelli S, et al. Interleukin-1 blockade in patients with Wiskott-Aldrich syndrome: a retrospective multinational case series. Blood. 2024;144(16):1699-704.
17. McDonnell J, Cousins K, Younger MEM, Lane A, Abolhassani H, Abraham RS, et al. COVID-19 Vaccination in Patients with Inborn Errors of Immunity Reduces Hospitalization and Critical Care Needs Related to COVID-19: a USIDNET Report. Journal of clinical immunology. 2024;44(4).
18. Matucci-Cerinic C, Herzum A, Ciccarese G, Rosina S, Caorsi R, Gattorno M, et al. Therapeutic Role of HPV Vaccination on Benign HPV-induced Epithelial Proliferations in Immunocompetent and Immunocompromised Patients: Case Study and Review of the Literature. Open Forum Infectious Diseases. 2024;11(7).

19. Matucci-Cerinic C, Corona F, Varnier GC, Pastore S, Bocca P, Palmeri S, et al. Baricitinib treatment in children with COPA syndrome. Journal of Allergy and Clinical Immunology: In Practice. 2024;12(8):2201-4.
20. Hou C, Theodoropoulou K, Zaffalon L, Wang Z, Bertoni A, Volpi S, et al. HSP90β controls NLRP3 autoactivation. Science Advances. 2024;10(9).
21. Federici S, Cinicola BL, La Torre F, Castagnoli R, Lougaris V, Giardino G, et al. Vasculitis and vasculopathy associated with inborn errors of immunity: an overview. Frontiers in Pediatrics. 2024;11.
22. David C, Badonyi M, Kechiche R, Insalaco A, Zecca M, De Benedetti F, et al. Interface Gain-of-Function Mutations in TLR7 Cause Systemic and Neuro-inflammatory Disease. Journal of clinical immunology. 2024;44(2).
23. Coppola E, Sgrulletti M, Cortesi M, Romano R, Cirillo E, Giardino G, et al. The Inborn Errors of Immunity—Virtual Consultation System Platform in Service for the Italian Primary Immunodeficiency Network: Results from the Validation Phase. Journal of clinical immunology. 2024;44(2).
24. Chan YH, Lundberg V, Le Pen J, Yuan J, Lee D, Pinci F, et al. SARS-CoV-2 brainstem encephalitis in human inherited DBR1 deficiency. Journal of Experimental Medicine. 2024;221(9).
25. Cafaro A, Grossi A, Barco S, Pigliasco F, Biondi M, Schena F, et al. Diagnostic workflow for Adenosine Deaminase-2 Deficiency (DADA2): a proposal. Biochimica Clinica. 2024;48(1):62-6.
26. Cafaro A, Baiardi G, Pigliasco F, Barco S, Mattioli F, Volpi S, et al. A Novel LC-MS/MS Method for Therapeutic Drug Monitoring of Baricitinib in Plasma of Pediatric Patients. Therapeutic Drug Monitoring. 2024;46(1):67-72.
27. Caballero-Oteyza A, Crisponi L, Peng XP, Yauy K, Volpi S, Giardino S, et al. GenIA, the Genetic Immunology Advisor database for inborn errors of immunity. Journal of Allergy and Clinical Immunology. 2024;153(3):831-43.
28. Bruschi M, Angeletti A, Prunotto M, Meroni PL, Ghiggeri GM, Moroni G, et al. A critical view on autoantibodies in lupus nephritis: Concrete knowledge based on evidence. Autoimmunity Reviews. 2024;23(5).
29. Sutera D, Bustaffa M, Papa R, Matucci-Cerinic C, Matarese S, D'Orsi C, et al. Corrigendum to "Clinical characterization, long-term follow-up, and response to treatment of patients with syndrome of undifferentiated recurrent fever (SURF)" Seminars in Arthritis and Rheumatism 55 (2022) 152024 (Seminars in Arthritis and Rheumatism (2022) 55, (S0049017222000750), (10.1016/j.semarthrit.2022.152024)). Seminars in Arthritis and Rheumatism. 2023;60.
30. Rossano M, Conti EA, Bocca P, Volpi S, Mastrangelo A, Cavalli R, et al. Novel heterozygous TREX1 mutation in a juvenile systemic lupus erythematosus patient with severe cutaneous involvement treated successfully with Jak-inhibitors: a case report. Frontiers in Immunology. 2023;14.
31. Orsi SM, Pepino C, Rossoni L, Serafino M, Caorsi R, Volpi S, et al. Case Report: Multisystem inflammatory syndrome in children with associated proximal tubular injury. Frontiers in Nephrology. 2023;3.
32. Matucci-Cerinic C, Malattia C, Pistorio A, Rosina S, Consolaro A, Viola S, et al. Skin manifestations help identifying different phenotypes of paediatric SAPHO syndrome. Seminars in Arthritis and Rheumatism. 2023;63.
33. Lancieri M, Bustaffa M, Palmeri S, Prigione I, Penco F, Papa R, et al. An Update on Familial Mediterranean Fever. International Journal of Molecular Sciences. 2023;24(11).
34. Hirschenberger M, Lepelley A, Rupp U, Klute S, Hunszinger V, Koepke L, et al. ARF1 prevents aberrant type I interferon induction by regulating STING activation and recycling. Nature communications. 2023;14(1).
35. Giardino G, Romano R, Lougaris V, Castagnoli R, Cillo F, Leonardi L, et al. Immune tolerance breakdown in inborn errors of immunity: Paving the way to novel therapeutic approaches. Clinical Immunology. 2023;251.
36. Fava D, Morandi F, Prigione I, Angelelli A, Bocca P, Pistorio A, et al. Blood Lymphocyte Subsets and Proinflammatory Cytokine Profile in ROHHAD(NET) and non-ROHHAD(NET) Obese Individuals. Journal of the Endocrine Society. 2023;7(9).
37. Dell'Orso G, Bagnasco F, Giardino S, Pierri F, Ferrando G, Di Martino D, et al. Hematopoietic stem cell transplantation for inborn errors of immunity: 30-year single-center experience. Frontiers in Immunology. 2023;14.
38. Benamar M, Chen Q, Chou J, Julé AM, Boudra R, Contini P, et al. The Notch1/CD22 signaling axis disrupts Treg function in SARS-CoV-2–associated multisystem inflammatory syndrome in children. Journal of Clinical Investigation. 2023;133(1).
39. Aldera E, Dufour F, Mercuri C, Rosina S, Volpi S, Gattorno M, et al. DADA-ism (2). Medico e Bambino. 2023;42(9):600.
40. Zhou Q, Kang G, Jiang P, Qiao R, Lam WKJ, Yu SCY, et al. Epigenetic analysis of cell-free DNA by fragmentomic profiling. Proceedings of the National Academy of Sciences of the United States of America. 2022;119(44).
41. Yang L, Booth C, Speckmann C, Seidel MG, Worth AJJ, Kindle G, et al. Phenotype, genotype, treatment, and survival outcomes in patients with X-linked inhibitor of apoptosis deficiency. Journal of Allergy and Clinical Immunology. 2022;150(2):456-66.

42. Wisniewski M, Chun A, Volpi S, Muscal E, Sexson Tejtel SK, Munoz F, et al. Outcomes after SARS-CoV-2 Vaccination among Children with a History of Multisystem Inflammatory Syndrome. JAMA Network Open. 2022.
43. Trivioli G, Gelain E, Angelotti ML, Ravaglia F, Allinovi M, Lodi L, et al. A Report of 2 Cases of Kidney Involvement in ADA2 Deficiency: Different Disease Phenotypes and the Tissue Response to Type I Interferon. American Journal of Kidney Diseases. 2022;80(5):677-82.
44. Sutera D, Bustaffa M, Papa R, Matucci-Cerinic C, Matarese S, D'Orsi C, et al. Clinical characterization, long-term follow-up, and response to treatment of patients with syndrome of undifferentiated recurrent fever (SURF). Seminars in Arthritis and Rheumatism. 2022;55.
45. Steiner A, Hrovat-Schaale K, Prigione I, Yu CH, Laohamonthonkul P, Harapas CR, et al. Deficiency in coatomer complex I causes aberrant activation of STING signalling. Nature communications. 2022;13(1).
46. Sin STK, Deng J, Ji L, Yukawa M, Chan RWY, Volpi S, et al. Effects of nucleases on cell-free extrachromosomal circular DNA. JCI Insight. 2022;7(8).
47. Signa S, Bertoni A, Penco F, Caorsi R, Cafaro A, Cangemi G, et al. Adenosine Deaminase 2 Deficiency (DADA2): A Crosstalk Between Innate and Adaptive Immunity. Frontiers in Immunology. 2022;13.
48. Penco F, Petretto A, Lavarello C, Papa R, Bertoni A, Omenetti A, et al. Proteomic Signatures of Monocytes in Hereditary Recurrent Fevers. Frontiers in Immunology. 2022;13.
49. Papa R, Rusmini M, Volpi S, Dell'Orso G, Giarratana MC, Caorsi R, et al. Progression of non-hematologic manifestations in SAMD9L-associated autoinflammatory disease (SAAD) after hematopoietic stem cell transplantation. Pediatric Allergy and Immunology. 2022;33(1).
50. Papa R, Caorsi R, Volpi S, Gattorno M. New monogenic autoinflammatory diseases: 2021 year in review. Immunology Letters. 2022;248:96-8.
51. Oliveira Mendoncą L, Matucci-Cerinic C, Terranova P, Casabona F, Bovis F, Caorsi R, et al. The challenge of early diagnosis of autoimmune lymphoproliferative syndrome in children with suspected autoinflammatory/autoimmune disorders. Rheumatology (United Kingdom). 2022;61(2):696-704.
52. Matucci-Cerinic C, Viglizzo G, Pastorino C, Corcione A, Prigione I, Bocca P, et al. Remission of eczema and recovery of Th1 polarization following treatment with Dupilumab in STAT3 hyper IgE syndrome. Pediatric Allergy and Immunology. 2022;33(4).
53. Lougaris V, Cancrini C, Rivalta B, Castagnoli R, Giardino G, Volpi S, et al. Activated phosphoinositide 3-dinase delta syndrome (APDS): An update. Pediatric Allergy and Immunology. 2022;33(S27):69-72.
54. Leonardi L, La Torre F, Soresina A, Federici S, Cancrini C, Castagnoli R, et al. Inherited defects in the complement system. Pediatric Allergy and Immunology. 2022;33(S27):73-6.
55. Giardino S, Volpi S, Lucioni F, Caorsi R, Schneiderman J, Lang A, et al. Hematopoietic Stem Cell Transplantation in ARPC1B Deficiency. Journal of clinical immunology. 2022;42(7):1535-44.
56. Drago E, Garbarino F, Signa S, Grossi A, Schena F, Penco F, et al. Case Report: Susceptibility to viral infections and secondary hemophagocytic lymphohistiocytosis responsive to intravenous immunoglobulin as primary manifestations of adenosine deaminase 2 deficiency. Frontiers in Immunology. 2022;13.
57. Ding SC, Chan RWY, Peng W, Huang L, Zhou Z, Hu X, et al. Jagged Ends on Multinucleosomal Cell-Free DNA Serve as a Biomarker for Nuclease Activity and Systemic Lupus Erythematosus. Clinical Chemistry. 2022;68(7):917-26.
58. Cinicola BL, Corrente S, Castagnoli R, Lougaris V, Giardino G, Leonardi L, et al. Primary atopic disorders and chronic skin disease. Pediatric Allergy and Immunology. 2022;33(S27):65-8.
59. Chiriaco M, Ursu GM, Amodio D, Cotugno N, Volpi S, Berardinelli F, et al. Radiosensitivity in patients affected by ARPC1B deficiency: a new disease trait? Frontiers in Immunology. 2022;13.
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