ANNA VILLA
CURRICULUM VITAE

Anna Villa	Director of the Institute of Genetics and Biomedicine (IRGB) – National Research Council Head of Unit of Telethon Institute for Gene Therapy (San Raffaele-Milano)
Current address:	Home : Via Camillo Hajech 22 Milano 20129-Italy Cell Phone: +0039 3488400856 Lab: San Raffaele Hospital, Telethon Institute for Gene Therapy Via Olgettina 60 Milano 20132 Tel. +0039 02 26435273 e.mail address: villa.anna@hsr.it

Educational/Training

INSTITUTION AND LOCATION	DEGREE	YEAR(s)	FIELD OF STUDY
University of Milan, Milan, Italy	M.D	1986	Medicine Genetics
University of Milan, Milan, Italy	PhD	1990	Oncology
National Research Council Institute of Biotechnology (ITB) Segrate (Milan)- Italy	Post doc.	1991	Molecular Biology CNR, Milan, Italy
National Research Council Institute of Biotechnology (ITB) Segrate (Milan)- Italy	Researcher	1998	Molecular Biology

A. POSITION AND HONORS

Positions and Employment

1993-1998	Senior Investigator, Institute of Biomedical Technology, Segrate (Milan) Italy
1999-2001	Researcher at Consiglio Nazionale delle Ricerche, Institute of Biomedical Technology, Segrate (Milan) Italy
1998-2002	Professor of Biotechnology, University of Milan, Pharmacology Faculty.
2002—2006	Associate Professor, CNR, Institute of Biomedical Technology, Segrate (Milan) Italy
2006- 2011	Director of Research at CNR Institute of Biomedical Technology, Segrate (Milan) Italy
2011-present	Director of Research at CNR, IRGB, Milan Unit
2006-present	Head of Unit of "Omenn and Wiskott Aldrich group" at Telethon Institute for Gene Therapy, San Raffaele, Milan, Italy
2008-present	Head of Human Genome Unit, Istituto Clinico Humanitas, Milan Italy
July 2019- Sept2020	Director of the Institute of Genetics and Biomedicine (IRGB) -CNR

Other experiences and Professional Memberships

Academic and International Activities

1999-present	Reviewer for Primary Immunodeficiency Association (PiA) and Association Francaise contre les Myopathies (AFM)
2000-present	Responsible for data bank of Rag Defects and Osteopetrosis : http://www.uta.fi/bioinfo
2000-2006	Head of the Genetic Working Party- European Society for Immunodeficiencies
2008-present	Coordinator of the Retrospective Study on Osteopetrosis – Inborn Error Group (EBMT)
2010-2014	Secretary of the European Society of Immunodeficiency (ESID)

International Journal Reviewer:

Journal of Allergy and Clinical Immunology
Journal of Clinical Immunology
Journal of Leukocyte Biology
Blood
Bone and Osteoporosis Inter.
Journal of Bone and Mineral Research
Molecular Therapy
Expert Opinion on Orphan Drug
Frontiers in Immunology

International Journal Board Member

Journal of Allergy and Clinical Immunology, Journal of Clinical Immunology, Frontiers in Immunology

Grant Reviewer:

Action Medical Research for Children (UK)

Agence Nationale delle Recherche (ANR_AFM) (France)
Fondation pour le Recherche Medicale Equipe FRM (France)
Centre Hospitalier Regionael Universitarie Montpellier (France)
Primary Immunodeficiency Association (PiA) and Association Francaise contre les Myopathies (AFM)
External Panel Reviewer, ERC Starting Grants
Member of the Panel Board of Advanced ERC Grants

Patent:

Genetic markers for bone mass. The University Court of the University Aberdeen (Scotland)
Patent N. 03737357.8-2402-GB0300470
Date of filing: 04.02.03

Honors

2005 Winner, 2005 "Descartes Prize for Research"
2008 Member of Kunkel Society of Immunology

Meeting Organizer Committee
2012 2nd International Workshop of Translational Medicine, Istituto Clinico Humanitas, Rozzano (Milan)
2012 ESID Biennial Meeting – Florence
2017 4th Meeting of Translational Immunology of pathogenesis ad therapy of immunemediated diseases, Palermo
2019 5th Meeting of Translational Immunology of pathogenesis ad therapy of immunemediated diseases, Milano

Invited Presentations to peer-reviewed internationally established conferences

26 May 2002	Invited speaker at the 29th European Symposium on Calcified Tissue (ECTS), Zagreb
10 May 2004	Invited speaker at ESID Spring Meting, Prague
May 2004	Invited speaker at XI Biennial Meeting of the European Society of Immunodeficiency, Versailles
6 October 2006	Invited speaker at IPOPI, XII Biennial Meeting of the European Society of Immunodeficiency, Budapest
June 2007	Invited speaker at International Union of Immunological Societies (IUIS), Jackson Hole Wyoming, USA
May 2008	Invited speaker at Second Course of Dysphormology, Bertinoro
June 2008	Invited speaker at Kunkel Society Meeting- Santa Margherita
25 June 2009	Invited speaker at 5th International Conference on Children's Bone Health
11 February 2010	Invited speaker at USGEB, Palazzo dei Congressi, Lugano, Switzerland
5 October 2011	Invited speaker Ospedale Bambin Gesu', Rome (Host: Prof. Paolo Rossi)
23 November 2011	Invited speaker at 6th European Workshop on Immune Mediated Inflammatory Diseases, Nice
23 January 2012	Invited speaker at University of NewCastle, Department of Immunology NewCastle (UK). Seminar (Host: Prof. A. Cant)
8-10 March 2012	Invited speaker 2nd African Society of Immunodeficiency (ASID), Hammamet,

	Tunisia
3-6 October 2012	Invited speaker at 15th ESID Biennial Meeting, Florence
5 September 2012	Invited Chairperson at European Congress of Immunology (ECI), Glasgow
8 April 2013	Invited speaker at 39th Annual Meeting of European Group for Blood and Bone Marrow Transplantation(EBMT), London
June 2013	Invited speaker at AIEOP, Rome
6 February 2014	Invited speaker at The 100 J Project Meeting, Antalya
Ocotber 2014	Invited speaker at XVI Biennial Meeting of the European Society of Immunodeficiency, Prague.
12-14 June 2014	Invited speaker "Wiskott-Aldrich Syndrome from the B cell point of view". Frontiers in Immunology, From Molecules to Diseases. Stockholm
June 2015	Inivited speaker at International Conference on Children and Bone's Health, ICCBH, Salzburg
9 October 2015	Invited speaker at "Intestinal Microbiota in Omenn Syndrome". Ospedale Bambin Gesu'
10 October 2015	Invited speaker Conference on Autoimmunity in Severe Combined Immunodeficiency" Ospedale Pediatrico Bambin Gesu Rome
13 November 2015	Invited speaker at XV Congresso S.I.O.M.M.S. Bologna
6 November 2016	Invited speaker at FPID, NewPort Beach, DC
15 June 2016	Invited speaker at Forum in Bone and Minerla Research, Torino
1 December 2017	Invited speaker at BSRT Meeting, Berlin
18 October 2017	Invited speaker at the Annual Meeting of the Scandinavian Society for Immunology
28 March 2017	Invited speaker at the 4th Conference of Translational Medicine on pathogenesis and therapy of immune mediated diseases
9 June 2017	Invited speaker at II Update in Immunologia
26 Maggio 2017	Invited speaker at Ricerca traslazionale come ausilio per il management del paziente con Immunodeficienze Primitive
1 May 2018	Invited speaker at Brown Bag Seminar, NIH USA
25 May 2018	Invited speaker ECTS, Valencia
18 January 2019	Invited speaker 2 Journee Recherche FAI ^{2}R/CRI IMIDIATE, Paris
22 March 2019	Invited speaker at Kunkel Society, New York

Grants:

1995-1998	Project E.0945 Telethon Grant entitled 'Identification of the gene responsible for T-B- severe combined immunodeficiency'. Principle Investigator- 1995-1998
1999-2001	Telethon Grant "SCID due to defects in VDJ recombination : human studies and animal models E0917. 2002-2005 Scientific Responsible for AFM (Association Francaise contre le Myopathie) e Comitato Telethon "Terapia Genica delle Immunodeficienze combinate gravi
2002-2006	Ministry of Education and Research-FIRB Italian Grant: "Deciphering cell identify and cell differentiation mechanisms in the post-genome era by using three paragdimatic systems: breast, bone and lymphocytes.
2006-2010	Scientific Responsible for Telethon Project A04 " Basic Biology of Omenn syndrome"

2006-2010 Scientific Responsible for Telethon Project A02 " Gene Therapy of Wiskott Aldrich syndrome"

2007-2010 Scientific responsible for Nobel CARIPLO "Genetics and functional genomics of myelomonocytic cells"

2009-2012 Scientific responsible for Fondazione CARIPLO Project " Cellular and molecular mechanisms of autoimmune manifestations in primary immunodeficiency due to partial defects in V(D)J recombination"

2010-2015 EC FP7 2010 call- CELL-PID: Translational research on cell based immunotherapy.

2010-2012 Responsible Research Project "Novel Approaches exploiting gene targeting and cell reprogramming in stem cells based regenerative medicine". PRIN 2009 (2008SR75557)

2011-2016 Scientific responsible of Telethon Core grant "Autoimmunity in Primary Immunodeficiencies".
TIGETA3 Telethon A3: Dissection of cellular and molecular mechanisms underlying autoimmunity in Omenn syndrome. Euro 512.600
TIGET A2 Telethon A2: Cellular and molecular basis of autoimmunity of Wiskott Aldrich syndrome. Euro 533.700

2011-2013 Induced pluripotent stem cells (iPSC) in the treatment of TCIRG1-dependent osteopetrosis: a step by step reproduction of the entire therapeutic process in the mouse. Minister of Health RF-2009-1499542

2012-2015 Molecular and cellular bases of intestinal mucosal pathology in Omenn Syndrome. Fondazione CARIPLO 2012-0519. Euro 240.000

2013-2017 EC FP72012 call-SYBIL: “System biology for the functional validation for genetic determinanats of skeletal diseases” Grant . 602300. Euro 496.800

2016-2020 Horizon 2020GRANT AGREEMENT NO: 666908: “Developing genetic Medicines for Severe Combined Immunodeficiency (SCIDNET)” Euro 473.000

2017-2020 PRIN 2015_ Ministry of Education and Research “New experimental therapies for genetic skeletal diseases”. Euro 83.506

2018-2022 Recomb “Stem-cell based gene therapy for recombination deficient SCID (RECOMB)”. Euro 532.500/ Participant

2019-2021 PRIN 2017 Ministero della Educazione e Ricerca “Advanced genetic engineering to study and treat monogenic”

SCIENTIFIC ACTIVITIES

Dr Villa has published more than 190 papers in international scientific journals for a total citation of 8558 and Scopus "*h*" index of 51. ORCID: http://orcid.org/0000-0003-4428-9013

The main focus of her research has been the molecular and cellular dissection of severe combined immunodeficiencies. In her early career, Anna Villa identified the genes responsible for different forms of severe combined immunodeficiency (SCID). In particular, she identified Jak3 as the gene responsible for T^+B^- SCID published on Nature in 1995 and later she discovered mutations in WAS gene in patients suffering from X-linked thromobocytopenia (*Nature Genetics*,1995). During the following years, she addressed her studies to a peculiar form of SCID, named Omenn syndrome, characterized by activated T cells and absence of B cells in the presence of high level of IgE (*Cell*, 1998). She was able to characterize the molecular defects underlying this enigmatic immunodeficiency. She showed that hypomorphic mutations in Rag1 and Rag2 genes impairing but not abolishing the protein activities, are responsible for this peculiar SCID form. She focused her analysis on the molecular and biochemical effects of these mutations trying to correlate the clinical signs of Omenn syndrome with the molecular defects. To further address the pathophysiology of this disease, she generated a murine model carrying a hypomorphic mutation in Rag2 gene found in an Omenn patient (*Journal of Clinical Investigation*, 2007). Thanks to the availability of this mouse model, which well recapitulates the human phenotype, she was able to demonstrate defects in the mechanisms of central and peripheral tolerance. She also performed studies on regulatory T cells in patients demonstrating a defect in suppression activity function. Defects in B cells in OS mouse model and patients have been also analysed (*JEM*,2010). In parallel with the identification of genes involved in SCID, Anna Villa has addressed her studies to the efficacy and safety of gene therapy of Wiskott Aldrich syndrome, a complex and severe X-linked disorder characterized by micro-thrombocytopenia, eczema, immunodeficiency, and increased risk to develop autoimmunity. Using third generation of lentiviral vector carrying human WAS gene driven by its own promoter, she demonstrated that gene therapy can restore functional defects in T cells and more recently in B cells. Thanks to the preclinical studies, a lentiviral vector based clinical trial for the human WAS disease is now undergoing at the San Raffaele Institute (*Science, 2013, JCI 2015, JACI 2015*). In parallel with gene therapy studies, she has also analyzed the pathophisiology of Wiskott Aldrich syndrome demonstrating that the lack of WASp causes a defect in maturation and function of iNKT cells (*JEM,* 2009), dendritic cells and B cells (*JEM 2013, JACI 2014, JCI 2015*). She is currently evaluating the follow up of gene therapy patients focusing on B cell and platelet defects.

Her group is now involved in the correction of RAG defects and in close collaboration with Notarangelo group (NIH) she has optimized a gene editing platform to correct RAG1 defect in CD34+ cells. In parallel, she has tested a novel conditioning approach based on the use of immunotoxin conjugated to anti-CD45 (*JACI, 2020*)

Anna Villa has also strongly contributed to the molecular dissection of another important genetic disease. In the last years, she has directed her interests to a heterogeneous group of bone diseases, named Osteopetrosis. These studies represent the beginning of the molecular dissection of this complex and heterogenous inherited bone defects.

Her group has indeed identified TCIRG1 as the gene responsible for autosomal recessive form of osteopetrosis (ARO) (*Nature Genetics, 2000*) and later she contributed to the characterization of two other forms of ARO due to defect in Grey Lethal and Pleckstrin genes, respectively (Nature Medicine, 2003; Journal of Clinical Investigation, 2007). More recently her group has described RANKL and RANK as genes responsible for the osteoclast poor Osteopetrosis (*Nature Genetics, 2007; Am J Human Genetics, 2008*). The molecular dissection of ARO has important implication not only for the molecular diagnosis, but also for the treatment of the disease. RANKL dependent ARO forms cannot be cured by bone marrow transplantation and the recognition of this molecular

defect could candidate these patients to alternative therapeutic approaches. To this end, *Rankl*$^{-/-}$ mice have been treated with the soluble form of RANKL demonstrating the feasibility and efficacy of this substitutive treatment as preclinical model of ARO caused by defect in this molecule (*J. Bone Miner Res 2012*). In parallel with these studies, her team has contributed to further understand the spectrum of clinical manifestations in autosomal recessive and dominant forms caused by defects in ClCN7 gene. Finally, analysis of exome sequencing has allowed identifying novel cases of CTSK gene mutations in patients originally diagnosed as interemediate osteopetrosis.
Currently, Villa's group is involved in the identification of candidate genes responsible for novel forms of ARO and ADO by genome wide sequencing and in parallel testing novel gene correction approaches. To this end, her lab has recently established a cellular data bank of induced Pluripotent stem cells (iPSC) obtained from patients carrying defects in the main genes involved in ARO forms. The generation of this cellular lines source represents a disease modelling relevant for the comprehension of the molecular and cellular bases of the disease and for testing novel cellular therapeutic approaches (*Stem Cell Reports,* 2015). To this regard, in the past Villa's group has performed pioneer studies aimed at understaining the feasibility and efficacy of in utero stem cell transplantation in the murine model of osteopetrosis (*Proc Natl Acad Sci, 2005*). More recently, her group has tested and optimized a novel strategy to correct TCIRG1 gene defect by gene targeting in murine iPSC obtained from the murine model of osteoepetrosis (*oc/oc* mouse). Finally, Villa's group has generated a third generation lentiviral vector carrying human RANKL gene to cure osteoclast-poor form of ARO casued by defects in rankl molecule. Experiments to test the efficacy of this gene therapy approach to cure RANKL defective osteopetrosis are currently ongoing.

PUBLICATIONS

1 Villa A, Cairo G, Pozzi MR, Schiaffonati L, Bardella L, Delia D, Biunno I, and Vezzoni P. "Lack of TdT and immunoglobin and T cell receptor gene rearrangements in Hodgkin's disease". Int. J. Biol. Markers 2: 65-70, 1987

2 Villa A, Biunno I, Sacco MG, Cairo G, Besana C, Rilke F, and Vezzoni P. Specificity of the rearrangements of the T cell gamma gene in human lymphomas. Tumori 74: 257-260, 1988

3 Villa A, Sacco MG, Cairo G, Biunno I, Mathieu-Mahul D, Larsen J, and Vezzoni P. An analysis in human lymphomas of a J region involved in a C-Myc-J alpha-translocation, relationship with TCR alpha. Biochem. Biophys. Research Comm. 154: 550-558, 1988

4 Vezzoni P, Villa A, and Pozzi MR. The rise of a microparadigm in oncology. Biology Philosophy 4: 57-67, 1989

5 Cattoretti G, Villa A, Giardini R, Vezzoni P, and Rilke F. Malignant hystiocitosis: phenotypic and genotypic heterogeneity. Am J Pathol 136: 1009-1919, 1990

6 Zocchi MR, Poggi A, Villa A, Inverardi L, Sabbadini MG, and Ferrarini M. Signal requirements for activation of leukemic T cells from a chronic lymphocytic leukemia TLL. Clin Exp Immunol 82:108-113, 1990

7 Patrosso MC, Frattini A, Susani L, Vezzoni P, and Villa A. Fidelity of a Yac clone in the region of human MCF-2 gene. Biochem Biophys Research Comm 181:877-883, 1991

8 Tribioli C, Tamiani F, Patrosso MC, Milanesi L, Villa A, Pergolizzi R, Rivella S, Bione S, Mancini M, Vezzoni P, and Toniolo D. "Methylation and sequence analysis around EagI sites: identification of 28 new CpG islands in Xq24-Xq28 ". Nucleic Acid Res. 4:727-733, 1992

9 Lucchini F, Sacco MG, Hu N, Villa A, Brown J, Cesana L, Mangiarini L, Rindi G, Kindl S, Sessa F, Vezzoni P, and Clerici L. Early and multifocal tumors in breast, salivary, Harderian and epididimal tissues developed in MMTV neu transgenic mice. Cancer Letters 64:203-209, 1992

10 Villa A, Patrosso MC, Biunno I, Frattini A, Repetto M, Mostardini M, Evans G, Susani L, Strina D, Redolfi E, Lazzari B, Pellegrini M, and P Vezzoni . Isolation of a zinc finger motif mapping on chromosome Xq25-26. Genomics 13:1231-1236, 1992

11 Frattini A, Zucchi I, Villa A, Patrosso MC, Repetto M, Susani L, Redolfi E, Vezzoni P, Romano G, Palmieri G, Esposito T, d'Urso M. Type 2 vasopressin receptor gene, the gene responsible for nephrogenic diabetes insipidus, maps to Xq28 close to L1CAM gene. Biochem Biophys Res Comm 3:864-871, 1993

12 Maestrini E, Patrosso MC, Mancini M, Rivella S, Rocchi M, Repetto M, Villa A, Frattini A, Zoppè M, Vezzoni P, and Toniolo D. Mapping of two genes encoding isoforms of ABP-280, a dystrophin like protein, to Xq28 and to chromosome 7: candidate genes for the Emery Dreyfuss Muscular Dystrophy. Hum Mol Gen 2:761-766, 1993

13 Villa A, Zucchi I, Pilia G, Strina D, Susani L, Morali F, Patrosso MC, Frattini A, Lucchini F, Repetto M, Sacco MG, Zoppé M, Vezzoni P.ZNF75: isolation of a cDNA clone of the KRAB zinc finger gene subfamily mapped in YACs 1 Mb telomeric of HPRT. Genomics, 18:223-229, 1993.

14 Mulder L, Sacco MG, Mangiarini L, Brown J, Collotta A, Villa A, De Giovanni A, Vezzoni P, Clerici P. Preimplantation embryo sexing by polymerase chain reaction amplification of the sry gene on single mouse blastomeres Genet Anal Applic Techn,10:147-149, 1993

15 Villa A, Strina D, Macchi P, Patrosso MC, Vezzoni P, Tovo PA, Giliani S, Ugazio AG, Notarangelo LD. C to T mutation causing premature termination of CD40 ligand at aminoacid 221 in a patient affected by HyperIgM syndrome Human Mut, 3:73-76, 1994

16 Patrosso MC, Repetto M, Villa A, Milanesi L, Frattini A, Faranda S, Mancini M, Maestrini E, Toniolo D, Vezzoni P. The exon-intron organization of the human X-linked gene enconding ABP 280. Genomics, 21: 71-76, 1994.

17 Villa A, Notarangelo LD, Di Santo J, Macchi P, Strina D, Frattini A, Lucchini F, Patrosso MC, Giliani S, Mantuano E, Agosti S, Nocera G, Kroczek RA, Ugazio A, de Saint Basil G, Vezzoni P. Organization of the human CD40L gene: implications for molecular defects in X-linked hyper-IgM syndrome and prenatal diagnosis. Proc Natl Acad Sci USA, 91:2110-2114,1994.

18 Ferlini A, Patrosso MC, Repetto M, Frattini A, Villa A, Vezzoni P, Fini S, Salvi F, and Forabosco A. A new mutation (GLY47ALA) in the transthyretin gene associated with hereditary amylodoisis: detection by sequence analysis and by primer-created restriction polymorphism. Human Mutation 138: 39-59,1994

19 Kroczek R, Graf D, Brugnoni D, Giliani S, Korthauer U, Ugazio A, Senger G, Mages H, Villa A, Notarangelo L. Defective expression of CD40 ligand on T cells causes X linked immunodeficiency

with hyper-Ig M (HIGM1). Immunological Review,138:39-59, 1994

20 Frattini A, Faranda S, Redolfi E, Zucchi I, Villa A, Patrosso MC, Strina D, Susani L, Vezzoni P. " Genomic organization of the human VP16 accessory protein (HCF), a housekeeping gene mapping to Xq28". Genomics, 23:30-35, 1994

21 Pilia G, Porta G, Padayacie M, Malcolm S, Zucchi I, Villa A, Macchi P, Vezzoni P, Schlessinger D. Human CD40L gene, gp39, maps to 2 megabases telomeric to HPRT. Genomics, 22:249-251, 1994

22 Di Bacco A, Susani L, Villa A, Strina D, Frattini A, Vezzoni P, Zucchi I. Rapid isolation of cDNA clones by aliquot testing via PCR amplification. PCR Methods Applic 4:126-128, 1994

23 Macchi P, Notarangelo L, Giliani S, Strina D, Repetto M, Sacco MG, Vezzoni P, Villa A. The genomic organization of the human transcription factor 3 (TFE3) gene. Genomics 28:491-494, 1995

24 Villa A, Notarangelo L, Macchi P, Mantuano E, Cavagni G, Brugnoni D, Strina D, Patrosso MC, Ramenghi U, Sacco MG, Ugazio A, Vezzoni P. X-linked thrombocytopenia and Wiskott-Aldrich syndrome are allelic diseases with mutations in the Wasp gene. Nature Genet 9:414.417, 1995

25 Macchi P, Villa A, Strina D, Sacco MG, Morali F, Brugnoni D, Giliani S, Mantuano E, Fasth A, Andersson B, Zegers BJM, Cavagni G, Reznick I, Levy J, Zan-Bar I, Porat Y, Airò P, Plebani A, Vezzoni P, Notarangelo L. Characterization of nine novel mutations in the CD40 ligand gene in patients with X-linked Hyper IgM syndrome of various ancestry. Am J Hum Genet, 56:898-906, 1995

26 Sacco MG, Mangiarini L, Villa A, Macchi P, Barbieri O, Sacchi MC, Monteggia E, Fasolo V, Vezzoni P, Clerici L. Local regression of breast tumors following intrammary ganciclovir administration in double transgenic mice expressing neu oncogene and herpes simplex virus thymidine kinase. Gene Therapy 2:493-497, 1995

27 Macchi P, Villa A*, Giliani S, Sacco MG, Frattini A, Porta F, Ugazio A, Johnston J, Candotti F, O' Shea J, Vezzoni P, Notarangelo G. Mutations of JAK3 gene in patients with autosomal severe combined immunodeficiency (SCID). Nature 377:65-68, 1995 ***Corresponding author**

28 Villa A, Sironi M, Macchi P, Matteucci C, Notarangelo L, Vezzoni P, Mantovani A. Monocyte function in SCID patient with a donor splice site mutation in the JAK3 gene. Blood 88:817-823, 1996

29 Sacco MG, Benedetti S, Duflot-Dancer A, Mesnil M, Bagnasco L, Strina D, Fasolo V, Villa A, Macchi P, Faranda S, Vezzoni P, Finocchiaro G. Partial regression, yet incomplete eradication of mammary tumors in transgenic mice by retroviral-mediated HSV-TK transfer in vivo. Gene Therapy 3:1151-1156, 1996

30 Frattini A, Chatterjee A, Faranda S, Sacco MG, Villa A, Herman GE, Vezzoni P. The Chromosome localization and the HCF repeats of the Human Host Cell Factor Gene are conserved in the Mouse Homologue. Genomics, 32:277-280, 1996

31 Villa A, Strina D, Frattini A, Faranda S, Macchi P, Bozzi F, Susani L, Arcidiacono N, Rocchi M, Vezzoni P. The ZNF75 zinc finger gene subfamily: isolation and mapping of the four members in

humans and great apes. Genomics, 35:312-320, 1996

32 Bozzi F, Bertuzzi S, Strina D, Giannetto C, Vezzoni P, Villa A. The exon-intron structure of human LHX1 gene. Biochem Biophys Res Comm. 229:494-497, 1996

33 Notarangelo LD, Villa A, Candotti F, Giliani S, Mella P, Brugnoni D, Macchi P, Badolato R, Schumacher RF, Mazzolari E, Pennacchio M, Porta F, Ugazio AG, O'Shea JJ, Vezzoni P. Severe combined immune deficiency due to defects of the JAK3 tyrosine kinase. Progr Immunodef 6:61-68, 1996

34 Candotti F, Oakes SA, Johnston JA, Giliani S, Schumacher RF, Mella P, Fiorini M, Ugazio AG, Badolato R, Notarangelo LD, Bozzi F, Macchi P, Strina D, Vezzoni P, Blaese MR, O'Shea JJ, Villa A. Structural and functional basis for JAK3-deficient severe combined immunodeficiency. Blood 90:3996-4003, 1997

35 Sacco MG, Gribaldo L, Barbieri O, Turchi G, Zucchi I, Collotta A, Bagnasco L, Barone D, Montagna C, Villa A, Marafante E and Vezzoni P. Establishment and characterization of a new mammary adenocarcinoma cell line derived from MMTV neu transgenic mice. Breast Cancer Res Treat, 47: 171-180, 1998

36 Brugnoni D, Notarangelo LD, Sottini A, Airò P, Pennacchio M, Mazzolari E, Signorini S, Candotto F, Villa A, Mella P, Vezzoni P, Cattaneo R, Ugazio AG, Imberti L. Development of autologous, oligoclonal, poorly functioning T lymphocytes in a patient with autosomal recessive severe combined immunodeficiency due to defects of the Jak3 tyrosine kinase. Blood 91:949-955, 1998

37 Villa A, Santagata S, Bozzi F, Giliani S, Frattini A, Imberti L, Benerini Gatta L, Ochs HD, Schwarz K, Notarangelo L, Vezzoni P and Spanopoulou E. Partial V(D)J recombination activity leads to Omenn syndrome. Cell 93: 885-896, 1998

38 Colombo I, Monteggia E, Moretti S, Mangiarini L, Sacco MG, Villa A, Rapelli S, Clerici L, Berra B. Oncogene transgenic mice: a useful model to sudy in vivo the relationships between gangliosides and oncogenes. Cancer Biochem Biophys, 16:229-242, 1998

39 Candotti F, O'Shea JJ, Villa A. Severe combined immune deficiencies due to defects of the common gamma-chain-JAK3 signaling pathway. Springer Seminars Immunopathology 19: 401-405, 1998

40 Bozzi P, Lefranc G, Villa A, Badolato R, Schumacher RF, Khalil G, Loiselet J, Bresciani S, O'Shea JJ, Vezzoni P, Notarangelo LD, Candotti F. Molecular and biochemical characterization of JAK3 deficiency in a patient with severe combined immunodeficiency over 20 years after bone marrow transplantation: implications for treatment. Brit. J. Haemat :102:1363-1366, 1998

41 Candotti F, Villa A, Notarangelo LD. Severe combined immunodeficiency due to defects of Jak3 tyrosine kinase. In Primary Immunodeficiencies Diseases, Ochs H, Smith ECI, Puck J (eds) pp. 111-120, Oxford University Press, New York-Oxford, 1999

42 Schwarz K, Notarangelo LD, Spanopoulou E, Vezzoni P, Villa A. Recombination defects. In Primary Immunodeficiencies Diseases, Ochs H, Smith ECI, Puck J (eds) pp 155-166, Oxford University Press, New York-Oxford, 1999

43 Villa A, Santagata S, Imberti L, Bozzi F, Notarangelo LD. Omenn syndrome: a disorder of Rag1 and Rag2 genes. J Clin Immunol 19:87-97, 1999

44 Signorini S, Imberti L, Pirovano S, Villa A, Facchetti F, Ungari M, Bozzi F, Albertini A, Ugazio AG, Vezzoni P, Notarangelo LD. Intrathymic restriction and peripheral expansion of the T-cell repertoire in omenn syndrome. Blood 94:3468-78, 1999.

45 Schumacher RF, Mella P, Lalatta F, Fiorini M, Giliani S, Villa A, Candotti F, Notarangelo L. Prenatal diagnosis of JAK3 deficient SCID. Pren Diagn 19:653-656, 1999

46 Notarangelo LD, Villa A, Schwarz K. Rag and Rag defects. Curr Opin Immunol 11:435-442, 1999

47 Santagata S, Besmer E, Villa A, Bozzi F, Allingham JS, Sobacchi C, Hainford DB, Vezzoni P, Nussenzweig MC, Pan ZQ, Cortes P. The RAG1/RAG2 Complex Constitutes a 3'Flap Endonuclease: Implications for junctional diversity in V(D)J and transpositional recombination. Mol Cell. 4:1-20, 1999

48 Villa A, Bozzi F, Sobacchi C, Strina D, Fasth A, Pasic S, Notarangelo LD, Vezzoni P. Prenatal diagnosis of RAG-deficient Omenn syndrome. Prenatal Diagnosis, 20:56-59, 2000.

49 Schwarz K, Villa A. Rag mutations in severe combined and Omenn's syndrome. Immunol Allergy Clin North Amer 20:129-142, 2000.

50 Frattini F, Orchard PJ, Sobacchi C, Giliani S, Abinun M,. Mattsson JP, Keeling DJ, Andersson AK, Wallbrandt P, Zecca L, Notarangelo LD, Vezzoni P and Villa A. Defects in the TCIRG1-encoded 116kD subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genet, 25:343-346, 2000.

51 Gomez CA, Ptaszek LM, Villa A, Bozzi A, Sobacchi C, Brooks EG, Notarangelo LD, Spanopoulou E, Pan ZQ, Vezzoni P, Cortes P and Santagata S. Mutations in conserved regions of the predicted RAG2 kelch repeats block initiation of V(D)J recombination and result in primary immunodeficiencies. Mol Cell Biol, 20:5653-5664, 2000.

52 Vihinen M, Villa A, Mella P, Schumacher RF, Savoldi G, O'Shea JJ, Candotti F, Notarangelo LD. Molecular Modeling of the Jak3 Kinase Domains and Structural Basis for Severe Combined Immunodeficiency. Clin Immunol. 96:108-118, 2000.

53 Sacco MG, Ungari M, Mira Catò E, Villa A, Strina D, Notarangelo LD, Jonkers J, Zecca L, Facchetti F, Vezzoni P. Lymphoid abnormalities in CD40L transgenic mice suggest the need for tight regulation in gene therapy approaches to Hyper IgM syndrome. Cancer Gene Therapy, 7:1299-1206, 2000.

54 Santagata S, Villa A, Sobacchi C, Cortes P, Vezzoni P. The genetic and biochemical basis of Omenn syndrome. Immunol Rev, 178:64-74, 2000

55 Santagata S, Gomez CA, Sobacchi C, Bozzi F, Abinun M, Pasic S, Cortes P, Vezzoni P, Villa A. N-terminal RAG1 frameshift mutations in Omenn syndrome: internal methionine usage leads to partial V(D)J recombination activity and reveals a fundamental role in vivo for the N-terminal domains. Proc Natl Acad Sci USA, 97:14572-14577, 2000.

56 Vihinen M, Arredondo-Vega FX, Casanova JL, Etzioni A, Giliani S, Hammarstrom L, Hershfield MS, Heyworth PG, Hsu AP, Lahdesmaki A, Lappalainen I, Notarangelo LD, Puck JM, Reith W, Roos D, Schumacher RF, Schwarz K, Vezzoni P, Villa A, Valiaho J, Smith CI. Primary immunodeficiency mutation databases. Adv Genet. 43:103-188, 2001.

57 Villa A, Sobacchi C, Notarangelo LD, Bozzi F, Abinun M, Abrahamsen TG, Arkwright PD, Baniyash M, Brooks EG, Conley ME, Cortes P, Duse M, Fasth A, Filipovich AM, Infante AJ, Jones A, Mazzolari E, Muller SM, Pasic S, Rechavi G, Sacco MG, Santagata S, Schroeder ML, Seger R, 58 Strina D, Ugazio U, Väliaho J, Vihinen M, Vogler LB, Ochs H, Vezzoni P, Friedrich W, Schwarz K. V(D)J recombination defects in lymphocytes due to RAG mutations: a severe immunodeficiency with a spectrum of clinical presentations. Blood, 97: 81-88, 2001.

59 Sobacchi C, Frattini A, Orchard P, Porras O, Tezcan I, Andolina M, Babul-Hirji R, Baric I, Canham N, Chitayat D, Dupuis-Girod S, Ellis I, Etzioni A, Fasth A, Fisher A, Gerritsen B, Gulino V, Horwitz E, Klamroth V, Lanino E, Mirolo M, Musio A, Matthijs G, Nonomaya S, Notarangelo LD, Ochs HD, Superti Furga A, Valiaho J, van Hove JLK, Vihinen M, Vujic D, Vezzoni P, Villa A. The mutational spectrum of human malignant autosomal recessive osteopetrosis. Hum Mol Genet 10: 1767-1773, 2001.

60 Kumaki S, Villa A, Asada H, Kawai S, Ohashi Y, Takahashi M, Hakozaki I, Nitanai E, Minegishi M, Tsuchiya S. Identification of anti-herpes simplex virus antibody-producing B cells in a patient with an atypical RAG1 immunodeficiency. Blood. 98:1464-8, 2001.

61 Notarangelo LD, Santagata S, Villa A. Recombinase activating gene enzymes of lymphocytes. Curr Opin Hematol. 8:41-46, 2001.

62 Villa A, Sobacchi C, Vezzoni P. Recombination activating gene and its defects. Curr Op Allergy Clin Immunol, 1:491-495, 2001.

63 Villa A, Sobacchi C, Vezzoni P. Omenn syndrome in the context of other B cell-negative severe combined immunodeficiencies. Isr Med Assoc J. 4:218-221, 2002.

64 Taranta A, Migliaccio S, Recchia I, Caniglia M, Luciani M, De Rossi G, Dionisi-Vici C, Pinto RM, Francalanci P, Boldrini R, Lanino E, Dini G, Morreale G, Ralston SH, Villa A, Vezzoni P, Del Principe D, Cassiani F, Palumbo G, Teti A. Genotype-Phenotype Relationship in Human ATP6i-Dependent Autosomal Recessive Osteopetrosis. Am J Pathol. 162:57-68, 2003.

65 Chalhoub N, Benachenhou N, Rajapurohitam V, Pata M, Ferron M, Frattini A, Villa A, Vacher J. Mutation in grey lethal induce severe malignant autosomal recessive osteopetrosis in mouse and human. Nat. Med 9: 399-406, 2003.

66 Frattini A, Pangrazio A, Susani L, Sobacchi C, Mirolo M, Abinun M, Andolina M, Flanagan A, Horwitz EM, Mihci E, Notarangelo LD, Ramenghi U, Teti A, Van Hove J, Vujic D, Young T, Albertini A, Orchard PJ, Vezzoni P, Villa A. Chloride channel ClCN7 mutations are responsible for severe recessive, dominant, and intermediate osteopetrosis. J.Bone Miner Res. 18: 1740-1747,2003.

67 Sobacchi C, Vezzoni P, Reid DM, McGuigan FE, Frattini A, Mirolo M, Albhaga OM, Musio A, Villa A, Ralston SH. Association between a polymorphism affecting an AP1 binding site in the promoter of the TCIRG1 gene and bone mass in women. Calcif Tissue Int. 35-41, 2004 .

68 Blair HC, Borysenko CW, Villa A, Schlesinger PH, Kalla SE, Yaroslavskiy BB, Garcia-Palacios V, Oakley JI, Orchard PJ. In Vitro differentiation of CD14 cells from osteopetrotic subjects: contrasting phenotypes wuth TCIRG1, CLCN7 and attachments defects. J Bone Miner Re:1329-1338,2004.

69 Susani L, Pangrazio A, Sobacchi C, Taranta A, Mortier G, Savarirayan R, Villa A, Orchard P, Vezzoni P, Albertini A, Frattini A, Pagani F. TCIRG1-dependent recessive osteopetrosis: Mutation analysis, functional identification of the splicing defects, and in vitro rescue by U1 snRNA. Hum Mutat. 24:225-235, 2004.

70 Chen L, Morio T, Minegishi Y, Nakada S, Nagasawa M, Komatsu K, Chessa L, Villa A, Lecis D, Delia D, Mizutani S. Ataxia-telangiectasia-mutated dependent phosphorylation of Artemis in response to DNA damage. Cancer Sci. 96:134-141, 2005.

71 Cavadini P, Vermi W, Facchetti F, Fontana S, Nagafuchi S, Mazzolari E, Sediva A, Marrella V, Villa A, Fischer A, Notarangelo LD, Badolato R. AIRE deficiency in thymus of 2 patients with Omenn syndrome. J Clin Invest.115(3):728-32, 2005.

72 Musio A, Marrella V, Sobacchi C, Rucci F, Fariselli L, Giliani S, Lanzi G, Notarangelo LD, Delia D, Colombo R, Vezzoni P, Villa A. Damaging-agent sensitivity of Artemis-deficient cell lines. Eur J Immunol. 35:1250-6, 2005.

73 Smith AN, Jouret F, Bord S, Borthwick KJ, Al-Lamki RS, Wagner CA, Ireland DC, Cormier-Daire V, Frattini A, Villa A, Kornak U, Devuyst O, Karet FEVacuolar H+-ATPase d2 Subunit: Molecular Characterization, Developmental Regulation, and Localization to Specialized Proton Pumps in Kidney and Bone. J Am Soc Nephrol. 16:1245-1256,2005.

74 Frattini A, Blair HC, Sacco MG, Cerisoli F, Faggioli F, Cato EM, Pangrazio A, Musio A, Rucci F, Sobacchi C, Sharrow AC, Kalla SE, Bruzzone MG, Colombo R, Magli MC, Vezzoni P, Villa A. Rescue of ATPa3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation in utero. Proc Natl Acad Sci U S A. 102:14629-34, 2005.

75 Pettersson U, Albagha OM, Mirolo M, Taranta A, Frattini A, McGuigan FE, Vezzoni P, Teti A, Van Hul W, Reid DM, Villa A, Ralston SH.J. Polymorphisms of the CLCN7 Gene are associated with BMD in women. J Bone Miner Res. 20:1960-19677, 2005.

76 Kilic SS,Cilic E,Meral A, Villa A. Cardiac Thrombus in Omenn Syndrome. Pediatr Cardiol. 2005

77 Sobacchi C, Marrella V, Rucci F, Vezzoni P, Villa A. RAG-dependent primary immunodeficiencies. Hum Mutat. 27:1174-1184, 2006.

78 Rucci F, Cattaneo L, Marrella V, Sacco MG, Sobacchi C, Lucchini F, Nicola S, Della Bella S, Villa ML, Imberti L, Gentili F, Montagna C, Tiveron C, Tatangelo L, Facchetti F, Vezzoni P, Villa A. Tissue-specific sensitivity to AID expression in transgenic mouse models. Gene. 377:150-158, 2006.

79 Villa A, Vezzoni P, Frattini A. Osteopetroses and immunodeficiencies in humans.Curr Opin Allergy Clin Immunol. 6:421-427, 2006.

80 Pangrazio A, Poliani PL, Megarbane A, Lefranc G, Lanino E, Di Rocco M, Rucci F, Lucchini F,

Ravanini M, Facchetti F, Abinun M, Vezzoni P, Villa A, Frattini A. Mutations in OSTM1 (grey lethal) define a particularly severe form of autosomal recessive osteopetrosis with neural involvement. J Bone Miner Res. 21:1098-1105, 2006.

81 Sobacchi C, Frattini A, Guerrini MM, Abinun M, Pangrazio A, Susani L, Bredius R, Mancini G, Cant A, Bishop N, Grabowski P, Del Fattore A, Messina C, Errigo G, Coxon FP, Scott DI, Teti A, Rogers MJ, Vezzoni P, Villa A, Helfrich MH. Osteoclast-poor human osteopetrosis due to mutations in the gene encoding RANKL. Nat Genet. 39:960-962, 2007.

82 Souraty N, Noun P, Djambas-Khayat C, Chouery E, Pangrazio A, Villa A, Lefranc G, Frattini A, Mégarbané A.Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis.Eur J Med Genet. 50:188-199, 2007.

83 Van Wesenbeeck L, Odgren PR, Coxon FP, Frattini A, Moens P, Perdu B, MacKay CA, Van Hul E, Timmermans JP, Vanhoenacker F, Jacobs R, Peruzzi B, Teti A, Helfrich MH, Rogers MJ, Villa A, Van Hul W. Involvement of PLEKHM1 in osteoclastic vesicular transport and osteopetrosis in incisors absent rats and humans. J Clin Invest. 117:919-30, 2007.

84 Marrella V, Poliani PL, Casati A, Rucci F, Frascoli L, Gougeon ML, Lemercier B, Bosticardo M, Ravanini M, Battaglia M. Villa A. A hypomorphic R229Q Rag2 mouse mutant recapitulates human Omenn syndrome. J Clin Invest. 117:1260-1269, 2007.

85 Frattini A, Vezzoni P, Villa A, Sobacchi C. The Dissection of Human Autosomal Recessive Osteopetrosis Identifies an Osteoclast-Poor Form due to RANKL Deficiency. Cell Cycle. 6:3027-3033, 2007.

86 Marrella V, Poliani L, Sobacchi C, Rucci F, Grassi F, Villa A. Of Omenn and mice. Trends in Immunol 29:133-140, 2008

87 Villa A, Marrella V, Rucci F, Notarangelo LD. Genetically determined lymphopenia and autoimmune manifestations. Curr Opin Immunol. 20:318-323, 2008.

88 Guerrini MM, Sobacchi C, Cassani B, Abinun M, Kilic SS, Pangrazio A, Moratto D, Mazzolari E, Clayton-Smith J, Orchard P, Coxon FP, Helfrich MH, Crockett JC, Mellis D, Vellodi A, Tezcan I, Notarangelo LD, Rogers MJ, Vezzoni P, Villa A, Frattini A. Human osteoclast-poor osteopetrosis with hypogammaglobulinemia due to TNFRSF11A (RANK) mutations. Am J Hum Genet.: 64-76, 2008.

89 Villa A, Notarangelo LD, Roifman CM. Omenn syndrome: Inflammation in leaky severe combined immunodeficiency. J Allergy Clin Immunol. 122:1082-6, 2008.

90 Matangkasombut P, Pichavant M, Saez DE, Giliani S, Mazzolari E, Finocchi A, Villa A, Sobacchi C, Cortes P, Umetsu DT, Notarangelo LD. Lack of iNKT cells in patients with combined immune deficiency due to hypomorphic RAG mutations. Blood.111:271-4, 2008 .

91 Marrella V, Poliani PL, Sobacchi C, Grassi F, Villa A. Of Omenn and mice. Trends Immunol. 29:133-140, 2008.

92 Locci M, Draghici E, Marangoni F, Bosticardo M, Catucci M, Aiuti A, Cancrini C, Marodi L, Espanol T, Bredius RG, Thrasher AJ, Schulz A, Litzman J, Roncarolo MG, Casorati G, Dellabona P, Villa A. The Wiskott-Aldrich syndrome protein is required for iNKT cell maturation and function. J

Exp Med. 206: 735-742, 2009.

93 Marangoni F, Bosticardo M, Charrier S, Draghici E, Locci M, Scaramuzza S, Panaroni C, Ponzoni M, Sanvito F, Doglioni C, Liabeuf M, Gjata B, Montus M, Siminovitch K, Aiuti A, Naldini L, Dupré L, Roncarolo MG, Galy A, Villa A Evidence for Long-term Efficacy and Safety of Gene Therapy for Wiskott-Aldrich Syndrome in Preclinical Models. Mol Ther. 6: 1073-1082, 2009.

94 Villa A, Guerrini MM, Cassani B, Pangrazio A, Sobacchi C. Infantile malignant, autosomal recessive osteopetrosis: the rich and the poor. Calcif Tissue Int. 84:1-12, 2009.

95 Pangrazio A, Caldana ME, Sobacchi C, Panaroni C, Susani L, Mihci E, Cavaliere ML, Giliani S, Villa A, Frattini A. Characterization of a novel Alu-Alu recombination-mediated genomic deletion in the TCIRG1 gene in five osteopetrotic patients. J Bone Miner Res. 24:162-1677, 2009.

96 Poliani PL, Facchetti F, Ravanini M, Gennery AR, Villa A, Roifman CM, Notarangelo LD. Early defects in human T-cell development severely affect distribution and maturation of thymic stromal cells: possible implications for the pathophysiology of Omenn syndrome.Blood.114: 105-108, 2009.

97 Bosticardo M, Marangoni F, Aiuti A, Villa A, Roncarolo MG. Recent advances in understanding the pathophysiology of Wiskott-Aldrich syndrome. Blood. 113:6288-6295, 2009.

98 Schinke T, Schilling AF, Baranowsky A, Seitz S, Marshall RP, Linn T, Blaeker M, Huebner AK, Schulz A, Simon R, Gebauer M, Priemel M, Kornak U, Perkovic S, Barvencik F, Beil FT, Fattore AD, Frattini A, Streichert T, Pueschel K, Villa A, Debatin KM, Rueger JM, Teti A, Zustin J, Sauter G, Amling M. Impaired gastric acidification negatively affects calcium homeostasis and bone mass. Nat Med.15: 674-681, 2009.

99 Mazzolari E, Forino C, Razza A, Porta F, Villa A, Notarangelo LD. A single-center experience in 20 patients with infantile malignant osteopetrosis.Am J Hematol. 84:473-479, 2009.

100 Sauer AV, Mrak E, Hernandez RJ, Zacchi E, Cavani F, Casiraghi M, Grunebaum E, Roifman CM, Cervi MC, Ambrosi A, Carlucci F, Roncarolo MG, Villa A, Rubinacci A, Aiuti A. ADA-deficient SCID is associated with a specific microenvironment and bone phenotype characterized by RANKL/OPG imbalance and osteoblast insufficiency.Blood.114: 3216-3226, 2009.

101 Poliani PL, Kisand K, Marrella V, Ravanini M, Notarangelo LD, Villa A, Peterson P, Facchetti F. Human Peripheral Lymphoid Tissues Contain Autoimmune Regulator-Expressing Dendritic Cells. Am J Pathol. 176:1104-1112, 2010.

102 Pangrazio A, Pusch M, Caldana E, Frattini A, Lanino E, Tamhankar PM, Phadke S, Lopez AG, Orchard P, Mihci E, Abinun M, Wright M, Vettenranta K, Bariae I, Melis D, Tezcan I, Baumann C, Locatelli F, Zecca M, Horwitz E, Mansour LS, Van Roij M, Vezzoni P, Villa A, Sobacchi C. Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Hum Mutat. 31: 1071-1080, 2010.

103 Chouery E, Pangrazio A, Frattini A, Villa A, Van Wesenbeeck L, Piters E, Van Hul W, Coxon FP, Schouten T, Helfrich M, Lefranc G, Mégarbané A. A new familial sclerosing bone dysplasia. J Bone Miner Res.25:676-680, 2010

104 Couëdel C, Roman C, Jones A, Vezzoni P, Villa A, Cortes P. Analysis of mutations from SCID

and Omenn syndrome patients reveals the central role of the Rag2 PHD domain in regulating V(D)J recombination. J Clin Invest. 120:1337-1344, 2010.

105 Trifari S, Scaramuzza S, Catucci M, Ponzoni M, Mollica L, Chiesa R, Cattaneo F, Lafouresse F, Calvez R, Vermi W, Medicina D, Castiello MC, Marangoni F, Bosticardo M, Doglioni C, Caniglia M, Aiuti A, Villa A, Roncarolo MG, Dupré L. Revertant T lymphocytes in a patient with Wiskott-Aldrich syndrome: Analysis of function and distribution in lymphoid organs. J Allergy Clin Immunol.125: 439-448, 2010.

106 Cassani B, Poliani PL, Moratto D, Sobacchi C, Marrella V, Imperatori L, Vairo D, Plebani A, Giliani S, Vezzoni P, Facchetti F, Porta F, Notarangelo LD, Villa A*, Badolato R*. Defect of regulatory T cells in patients with Omenn syndrome. J Allergy Clin Immunol.125:209-216, 2010
* ***equal contribution***

107 Cassani B, Poliani PL, Marrella V, Schena F, Sauer AV, Ravanini M, Strina D, Busse CE, Regenass S, Wardemann H, Martini A, Facchetti F, van der Burg M, Rolink AG, Vezzoni P, Grassi F, Traggiai E, Villa A. Homeostatic expansion of autoreactive immunoglobulin-secreting cells in the Rag2 mouse model of Omenn syndrome. J Exp Med. 207:1525-1540, 2010.

108 Bosticardo M, Draghici E, Schena F, Sauer AV, Fontana E, Castiello MC, Catucci M, Locci M, Naldini L, Aiuti A, Roncarolo MG, Poliani PL, Traggiai E, Villa A. Lentiviral-mediated gene therapy leads to improvement of B-cell functionality in a murine model of Wiskott-Aldrich syndrome. J Allergy Clin Immunol. 127:1376-1384, 2011

109 Marrella V, Maina V, Villa A. Omenn syndrome does not live by V(D)J recombination alone. Curr Opin Allergy Clin Immunol. 11: 525-31, 2011

110 Pangrazio A, Boudin E, Piters E, Damante G, Iacono NL, D'Elia AV, Vezzoni P, Van Hul W, Villa A, Sobacchi C. Identification of the first deletion in the LRP5 gene in a patient with Autosomal Dominant Osteopetrosis type I. Bone. 49: 568-571, 2011

111 Catucci M, Prete F, Bosticardo M, Castiello MC, Draghici E, Locci M, Roncarolo MG, Aiuti A, Benvenuti F, Villa A. Dendritic cell functional improvement in a preclinical model of lentiviral-mediated gene therapy for Wiskott-Aldrich syndrome. Gene Ther. 2011 Dec 22. doi: 10.1038/gt.2011.202.

112 Sauer AV, Brigida I, Carriglio N, Jofra Hernandez R, Scaramuzza S, Clavenna D, Sanvito F, Poliani PL, Gagliani N, Carlucci F, Tabucchi A, Roncarolo MG, Traggiai E, Villa A, Aiuti A. Alterations in the adenosine metabolism and CD39/CD73 adenosinergic machinery cause loss of Treg cell function and autoimmunity in ADA-deficient SCID. Blood. 2011 Dec 19.

113. Michos A, Tzanoudaki M, Villa A, Giliani S, Chrousos G, Kanariou M. Severe combined immunodeficiency in Greek children over a 20-year period: rarity of γc-chain deficiency (X-linked) type. J Clin Immunol. 2011 Oct;31(5):778-83. Epub 2011 Jul 6.

114 Pangrazio A, Caldana ME, Iacono NL, Mantero S, Vezzoni P, Villa A, Sobacchi C. Autosomal recessive osteopetrosis: report of 41 novel mutations in the TCIRG1 gene and diagnostic implications. Osteoporos Int 23:2713-2718, 2012

115 Pangrazio A, Cassani B, Guerrini MM, Crockett JC, Marrella V, Zammataro L, Strina D, Schulz A, Schlack C, Kornak U, Mellis DJ, Duthie A, Helfrich MH, Durandy A, Moshous D, Vellodi A,

Chiesa R, Veys P, Iacono NL, Vezzoni P, Fischer A, Villa A, Sobacchi C. RANK-dependent autosomal recessive osteopetrosis: characterisation of 5 new cases with novel mutations. J Bone Miner Res.27: 342-351, 2012

116 Gioia R, Panaroni C, Besio R, Palladini G, Merlini G, Giansanti V, Scovassi IA, Villani S, Villa I, Villa A, Vezzoni P, Tenni R, Rossi A, Marini JC, Forlino A. Impaired Osteoblastogenesis in a Murine Model of Dominant Osteogenesis Imperfecta (OI), A New Target for OI Pharmacological Therapy. Stem Cells. 30: 1465-1476, 2012

117. Scaramuzza S, Biasco L, Ripamonti A, Castiello MC, Loperfido M, Draghici E, Hernandez RJ, Benedicenti F, Radrizzani M, Salomoni M, Ranzani M, Bartholomae CC, Vicenzi E, Finocchi A, Bredius R, Bosticardo M, Schmidt M, von Kalle C, Montini E, Biffi A, Roncarolo MG, Naldini L, Villa A, Aiuti A. Preclinical Safety and Efficacy of Human CD34(+) Cells Transduced With Lentiviral Vector for the Treatment of Wiskott-Aldrich Syndrome. Mol Ther. 21:175-184, 2012 .

118. Malu S, De Ioannes P, Kozlov M, Greene M, Francis D, Hanna M, Pena J, Escalante CR, Kurosawa A, Erdjument-Bromage H, Tempst P, Adachi N, Vezzoni P, Villa A, Aggarwal AK, Cortes P. Artemis C-terminal region facilitates V(D)J recombination through its interactions with DNA Ligase IV and DNA-PKcs. J Exp Med. 7;209(5):955-63,2012

119. van Til NP, de Boer H, Mashamba N, Wabik A, Huston M, Visser TP, Fontana E, Poliani PL, Cassani B, Zhang F, Thrasher AJ, Villa A, Wagemaker G. Correction of murine Rag2 severe combined immunodeficiency by lentiviral gene therapy using a codon-optimized RAG2 therapeutic transgene. Mol Ther. 20:1968-1980, 2012.

120. Marrella V, Poliani PL, Fontana E, Casati A, Maina V, Cassani B, Ficara F, Cominelli M, Schena F, Paulis M, Traggiai E, Vezzoni P, Grassi F, Villa A. Anti-CD3ε mAb improves thymic architecture and prevents autoimmune manifestations in a mouse model of Omenn syndrome: therapeutic implications. Blood. 120:1005-1014, 2012

121. Catucci M, Castiello MC, Pala F, Bosticardo M, Villa A. Autoimmunity in wiskott-Aldrich syndrome: an unsolved enigma. Front Immunol.3: 209; 2012.

122. Lo Iacono N, Blair HC, Poliani PL, Marrella V, Ficara F, Cassani B, Facchetti F, Fontana E, Guerrini MM, Traggiai E, Schena F, Paulis M, Mantero S, Inforzato A, Valaperta S, Pangrazio A, Crisafulli L, Maina V, Kostenuik P, Vezzoni P, Villa A, Sobacchi C. Osteopetrosis rescue upon RANKL administration to Rankl$^{-/-}$ mice: a new therapy for human RANKL-dependent ARO. J Bone Miner Res. 27: 2501-10, 2012

123. Pangrazio A, Frattini A, Valli R, Maserati E, Susani L, Vezzoni P, Villa A, Al-Herz W, Sobacchi C. A homozygous contiguous gene deletion in chromosome 16p13.3 leads to autosomal recessive osteopetrosis in a Jordanian patient. Calcif Tissue Int. 91: 250-254, 2012.

124. Aiuti A, Bacchetta R, Seger R, Villa A, Cavazzana-Calvo M. Gene therapy for primary immunodeficiencies: part 2. Curr Opin Immunol. 2012 Oct;24(5):585-91. doi: 10.1016/j.coi.2012.07.012. Epub 2012 Aug 18.

125. Mégarbané A, Pangrazio A, Villa A, Chouery E, Maarawi J, Sabbagh S, Lefranc G, Sobacchi C. Homozygous stop mutation in the SNX10 gene in a consanguineous Iraqi boy with osteopetrosis and corpus callosum hypoplasia. Eur J Med Genet. 56:32-35, 2013.

126. Pangrazio A, Fasth A, Sbardellati A, Orchard PJ, Kasow KA, Raza J, Albayrak C, Albayrak D, Vanakker OM, De Moerloose B, Vellodi A, Notarangelo LD, Schlack C, Strauss G, Kühl JS, Caldana E, Iacono NL, Susani L, Kornak U, Schulz A, Vezzoni P, Villa A, Sobacchi C. SNX10 mutations define a subgroup of human Autosomal Recessive Osteopetrosis with variable clinical severity. J Bone Miner Res. 28: 1041-1049, 2013

127. Prete F, Catucci M, Labrada M, Gobessi S, Castiello MC, Bonomi E, Aiuti A, Vermi W, Cancrini C, Metin A, Hambleton S, Bredius R, Notarangelo LD, van der Burg M, Kalinke U, Villa A, Benvenuti F. Wiskott-Aldrich syndrome protein-mediated actin dynamics control type-I interferon production in plasmacytoid dendritic cells. J Exp Med. 210(2):355-74, 2013.

128. Sobacchi C, Schulz A, Coxon FP, Villa A, Helfrich MH. Osteopetrosis: genetics, treatment and new insights into osteoclast function. Nat Rev Endocrinol. 9: 522-536, 2013.

129. Aiuti A, Biasco L, Scaramuzza S, Ferrua F, Cicalese MP, Baricordi C, Dionisio F, Calabria A, Giannelli S, Castiello MC, Bosticardo M, Evangelio C, Assanelli A, Casiraghi M, Di Nunzio S, Callegaro L, Benati C, Rizzardi P, Pellin D, Di Serio C, Schmidt M, Von Kalle C, Gardner J, Mehta N, Neduva V, Dow DJ, Galy A, Miniero R, Finocchi A, Metin A, Banerjee P, Orange J, Galimberti S, Valsecchi MG, Biffi A, Montini E, Villa A, Ciceri F, Roncarolo MG, Naldini L
Lentiviral Hematopoietic Stem Cell Gene Therapy in Patients with Wiskott-Aldrich Syndrome. Science. 341: 233151.2013 Jul 11.

130. Lo Iacono N, Pangrazio A, Abinun M, Bredius R, Zecca M, Blair HC, Vezzoni P, Villa A, Sobacchi C. RANKL Cytokine: From Pioneer of the Osteoimmunology Era to Cure for a Rare Disease. Clin Dev Immunol.:412768, 2013 doi: 10.1155/2013/412768

131. Moscatelli I, Thudium CS, Flores C, Schulz A, Askmyr M, Gudmann NS, Andersen NM, Porras O, Karsdal MA, Villa A, Fasth A, Henriksen K, Richter J. Lentiviral gene transfer of TCIRG1 into peripheral blood CD34(+) cells restores osteoclast function in infantile malignant osteopetrosis. Bone. 57(1):1-9,2013

132. Catucci M, Zanoni I, Draghici E, Bosticardo M, Castiello MC, Venturini M, Cesana D, Montini E, Ponzoni M, Granucci F, Villa A. Wiskott-Aldrich syndrome protein deficiency in natural killer and dendritic cells affects antitumor immunity. Eur J Immunol.44: 1039-45, 2014

133. Pangrazio A, Puddu A, Oppo M, Valentini M, Zammataro L, Vellodi A, Gener B, Llano-Rivas I, Raza J, Atta I, Vezzoni P, Superti-Furga A, Villa A, Sobacchi C.
Exome sequencing identifies CTSK mutations in patients originally diagnosed as intermediate osteopetrosis. Bone. 2014 59:122-6, 2014.

134. Castiello MC, Bosticardo M, Pala F, Catucci M, Chamberlain N, van Zelm MC, Driessen GJ, Pac M, Bernatowska E, Scaramuzza S, Aiuti A, Sauer AV, Traggiai E, Meffre E, **V**illa A*, van der Burg M. Wiskott-Aldrich Syndrome protein deficiency perturbs the homeostasis of B-cell compartment in humans. J Autoimmun. 50:42-50, 2014
*corresponding author

135. Bosticardo M, Musio S, Fontana E, Angiari S, Draghici E, Constantin G, Poliani PL, Pedotti R, Villa A. Development of central nervous system autoimmunity is impaired in the absence of Wiskott-Aldrich syndrome protein. PLoS One. 9: e86942, 2014

136. Sobacchi C, Pangrazio A, López AG, Gómez DP, Caldana ME, Susani L, Vezzoni P, Villa A. As Little as Needed: The Extraordinary Case of a Mild Recessive Osteopetrosis Due to a Novel Splicing Hypomorphic Mutation in the TCIRG1 Gene. J Bone Miner Res. 29: 1646-1650, 2014 .

137. Pasic S, Vujic D, Veljković D, Slavkovic B, Mostarica-Stojkovic M, Minic P, Minic A, Ristic G, Giliani S, Villa A, Sobacchi C, Lilić D, Abinun M. Severe combined immunodeficiency in Serbia and Montenegro between years 1986 and 2010: a single -center experience. J Clin Immunol. 34: 304-348, 2014.

138. Sic H, Kraus H, Madl J, Flittner KA, von Münchow AL, Pieper K, Rizzi M, Kienzler AK, Ayata K, Rauer S, Kleuser B, Salzer U, Burger M, Zirlik K, Lougaris V, Plebani A, Römer W, Loeffler C, Scaramuzza S, Villa A, Noguchi E, Grimbacher B, Eibel H. Sphingosine-1-phosphate receptors control B-cell migration through signaling components associated with primary immunodeficiencies, chronic lymphocytic leukemia, and multiple sclerosis. J Allergy Clin Immunol. 134: 420-428, 2014

139. Marrella V, Poliani PL, Notarangelo LD, Villa A. Rag defects and thymic stroma: lessons from animal models. Front Immunol. 5:259, 2014

140. van Til NP, Sarwari R, Visser TP, Hauer J, Lagresle-Peyrou C, van der Velden G, Malshetty V, Cortes P, Jollet A, Danos O, Cassani B, Zhang F, Thrasher AJ, Fontana E, Poliani PL, Cavazzana M, Verstegen MM, Villa A, Wagemaker G. Recombination-activating gene 1 (Rag1)-deficient mice with severe combined immunodeficiency treated with lentiviral gene therapy demonstrate autoimmune Omenn-like syndrome. J Allergy Clin Immunol. 133(4):1116-23, 2014.

141. Hoyoux C, Dresse MF, Forget P, Piette C, Rausin L, Villa A, Gothot A, Florkin B. Osteopetrosis mimicking juvenile myelomonocytic leukemia. Pediatr Int. 56: 779-82, 2014

142. Castiello MC, Scaramuzza S, Pala F, Ferrua F, Uva P, Brigida I, Sereni L, van der Burg M, Ottaviano G, Albert MH, Roncarolo MG, Naldini L, Aiuti A, Villa A, Bosticardo M. B-cell reconstitution after lentiviral vector-mediated gene therapy in patients with Wiskott-Aldrich syndrome. J Allergy Clin Immunol. 2015 Sep;136(3):692-702.e2

143. Marrella V, Lo Iacono N, Fontana E, Sobacchi C, Sic H, Schena F, Sereni L, Castiello MC, Poliani PL, Vezzoni P, Cassani B, Traggiai E, Villa A. IL-10 Critically Modulates B Cell Responsiveness in Rankl$^{-/-}$ Mice. J Immunol. 194: 4144-4153, 2015

144. Palagano E, Blair HC, Pangrazio A, Tourkova I, Strina D, Angius A, Cuccuru G, Oppo M, Uva P, Van Hul W, Boudin E, Superti-Furga A, Faletra F, Nocerino A, Ferrari MC, Grappiolo G, Monari M, Montanelli A, Vezzoni P, Villa A, Sobacchi C. Buried in the Middle, But Guilty: Intronic Mutations in the TCIRG1 Gene Cause Human Autosomal Recessive Osteopetrosis. J Bone Miner Res. 30(10):1814-1821, 2015.

145. Paulis M, Castelli A, Lizier M, Susani L, Lucchini F, Villa A, Vezzoni P.A pre-screening FISH-based method to detect CRISPR/Cas9 off-targets in mouse embryonic stem cells. Sci Rep. 2015 :12327. doi: 10.1038/srep12327

146. Neri T, Muggeo S, Paulis M, Caldana ME, Crisafulli L, Strina D, Focarelli ML, Faggioli F, Recordati C, Scaramuzza S, Scanziani E, Mantero S, Buracchi C, Sobacchi C, Lombardo A, Naldini L, Vezzoni P, Villa A, Ficara F. Targeted Gene Correction in Osteopetrotic-Induced Pluripotent Stem Cells for the Generation of Functional Osteoclasts. Stem Cell Reports. 5: 558-568, 2015.

147. Pala F, Morbach H, Castiello MC, Schickel JN, Scaramuzza S, Chamberlain N, Cassani B, Glauzy S, Romberg N, Candotti F, Aiuti A, Bosticardo M, Villa A*, Meffre E*. Lentiviral-mediated gene therapy restores B cell tolerance in Wiskott-Aldrich syndrome patients.
J Clin Invest. 125(10):3941-51,2015.
*** co-senior author**

148. Paulis M, Castelli A, Susani L, Lizier M, Lagutina I, Focarelli ML, Recordati C, Uva P, Faggioli F, Neri T, Scanziani E, Galli C, Lucchini F, Villa A, Vezzoni P. Chromosome transplantation as a novel approach for correcting complex genomic disorders.
Oncotarget. 2015 Oct 17. doi: 10.18632/

149. Rigoni R, Fontana E, Guglielmetti S, Fosso B, D'Erchia A.M, Maina V, Taverniti V, Castiello M.C, Mantero S, Pacchiana G, Musio S, Pedotti R, Mora J.R , Pesole G, Vezzoni P, Poliani LP, F. Grassi F, Villa A*, Cassani B. Intestinal microbiota sustains inflammation and autoimmunity induced by hypomorphic RAG defects. J Exp Med. 2016 Mar 7;213(3):355-75. doi: 10.1084/jem.20151116. Epub 2016 Feb 29. *corresponding author

150. Palagano E, Susani L, Menale C, Ramenghi U, Berger M, Uva P, Oppo M, Vezzoni P, Villa A, Sobacchi C. Synonymous Mutations Add a Layer of Complexity in the Diagnosis of Human Osteopetrosis. J Bone Miner Res. 2016 Jul 28. doi: 10.1002/jbmr.2929.

151. Farini A, Sitzia C, Cassani B, Cassinelli L, Rigoni R, Colleoni F, Fusco N, Gatti S, Bella P, Villa C, Napolitano F, Maiavacca R, Bosari S, Villa A, Torrente Y. Therapeutic Potential of Immunoproteasome Inhibition in Duchenne Muscular Dystrophy. Mol Ther. 2016 Sep 27. doi: 10.1038/mt.2016.162. [Epub ahead of print]

152. Rigoni R, Grassi F, Villa A, Cassani B. RAGs and BUGS: An alliance for autoimmunity. Gut Microbes. 2016 Aug 30:1-9. [Epub ahead of print]

153. Lleo A, Bian Z, Zhang H, Miao Q, Yang F, Peng Y, Chen X, Tang R, Wang Q, Qiu D, Fang J, Sobacchi C, Villa A, Di Tommaso L, Roncalli M, Gershwin ME, Ma X, Invernizzi P. Quantitation of the Rank-Rankl Axis in Primary Biliary Cholangitis. PLoS One. 2016 Sep 15;11(9):e0159612. doi: 10.1371/journal.pone.0159612. eCollection 2016.

154. Meneghini V, Frati G, Sala D, De Cicco S, Luciani M, Cavazzin C, Paulis M, Mentzen W, Morena F, Giannelli S, Sanvito F, Villa A, Bulfone A, Broccoli V, Martino S, Gritti A. Generation of Human Induced Pluripotent Stem Cell-Derived Bona Fide Neural Stem Cells for Ex Vivo Gene Therapy of Metachromatic Leukodystrophy. Stem Cells Transl Med. 2016 Sep 16. pii: sctm.2015-0414. [Epub ahead of print]

155. Sobacchi C, Villa A, Schulz A, Kornak U., Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. CLCN7-Related Osteopetrosis.GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2016.

156. Weisz Hubshman M, Basel-Vanagaite L, Krauss A, Konen O, Levy Y, Garty BZ, Smirin-Yosef P, Maya I, Lagovsky I, Taub E, Marom D, Gaash D, Shichrur K, Avigad S, Hayman Manzur L, Villa A, Sobacchi C, Shohat M, Yaniv I, Stein J. Homozygous Deletion of RAG1, RAG2 and 5' region TRAF6 Causes Severe Immune Suppression and Atypical Osteopetrosis.
Clin Genet. 2016 Nov 3. doi: 10.1111/cge.12916.

157. Schena F, Menale C, Caci E, Diomede L, Palagano E, Recordati C, Sandri M, Tampieri A, Bortolomai I, Capo V, Pastorino C, Bertoni A, Gattorno M, Martini A, Villa A, Traggiai E, Sobacchi C. Murine Rankl-/- Mesenchymal Stromal Cells Display an Osteogenic Differentiation Defect Improved by a RANKL-Expressing Lentiviral Vector. Stem Cells. 2017 May;35(5):1365-1377. doi: 10.1002/stem.2574

158. Sobacchi C, Palagano E, Villa A, Menale C. Soluble Factors on Stage to Direct Mesenchymal Stem Cells Fate. Front Bioeng Biotechnol. 2017 May 17;5:32.

159. Castiello MC, Pala F, Sereni L, Draghici E, Inverso D, Sauer AV, Schena F, Fontana E, Radaelli E, Uva P, Cervantes-Luevano KE, Benvenuti F, Poliani PL, Iannacone M, Traggiai E, Villa A, Bosticardo M. In Vivo Chronic Stimulation Unveils Autoreactive Potential of Wiskott-Aldrich Syndrome Protein-Deficient B Cells. Front Immunol. 2017 May 2; 8:490. doi: 10.3389/fimmu.2017.00490. eCollection 2017

160. Sereni L, Castiello MC, Villa A. Platelets in Wiskott-Aldrich syndrome: victims or executioners? J Leukoc Biol. 2017 Aug 29. pii: jlb.5MR0617-257R. doi: 10.1189/jlb.5MR0617-257R.

161. Faggioli F, Palagano E, Di Tommaso L, Donadon M, Marrella V, Recordati C, Mantero S, Villa A, Vezzoni P, Cassani B. B lymphocytes limit senescence-driven fibrosis resolution and favor hepatocarcinogenesis in mouse liver injury. Hepatology. 2017 Nov 3. doi: 10.1002/hep.29636. [Epub ahead of print]

162. Schiroli G, Ferrari S, Conway A, Jacob A, Capo V, Albano L, Plati T, Castiello MC, Sanvito F, Gennery AR, Bovolenta C, Palchaudhuri R, Scadden DT, Holmes MC, Villa A, Sitia G, Lombardo A, Genovese P, Naldini L. Preclinical modeling highlights the therapeutic potential of hematopoietic stem cell gene editing for correction of SCID-X1 Sci Transl Med. 2017 Oct 11;9(411). pii: eaan0820. doi:10.1126/scitranslmed.aan0820.

163. Cervantes-Luevano, K.E., Caronni, N., Castiello, M.C., Fontana E., Piperno, G., Naseem, A., Uva,P , Bosticardo, M., Marcovecchio, G.E., Notarangelo, L.D., Cicalese, M.P., Aiuti, A., Villa, A., Benvenuti, F. Neutrophils drive type-I interferon production and autoantibodies in Wiskott-Aldrich syndrome. J Allergy Clin Immunol J Allergy Clin Immunol. 2018 Feb 13. pii: S0091-6749(18)30207-0. doi: 10.1016/j.jaci.2017.11.063. [Epub ahead of print]

164. Valentina Capo, Maria Carmina Castiello, Elena Fontana, Sara Penna, Marita Bosticardo, Elena Draghici,; Luigi P. Poliani, Lucia Sergi Sergi, Rosita Rigoni, Barbara Cassani; Paola Carrera, Paolo Uva,; Kerry Dobs, Luigi D. Notarangelo, Niek P. van Til Gerard Wagemaker, Anna Villa Efficacy of lentiviral mediated gene therapy in Omenn syndrome is not hindered by inflammation and immune dysregulation. J Allergy Clin Immunol Allergy Clin Immunol. 2017 Dec 11. pii: S0091-6749(17)31886-9. doi: 10.1016/j.jaci.2017.11.015. [Epub ahead of print]
PMID: 29241731

165. Sereni L, Castiello MC, Marangoni F, Anselmo A, di Silvestre D, Motta S, Draghici E, Mantero S, Thrasher AJ, Giliani S, Aiuti A, Mauri P, Notarangelo LD, Bosticardo M, Villa A. Autonomous role of Wiskott-Aldrich syndrome platelet deficiency in inducing autoimmunity and inflammation. J Allergy Clin Immunol. 2018 Feb 6. pii: S0091-6749(18)30201-X. doi: 10.1016/j.jaci.2017.12.1000.

166. Palagano E, Menale C, Sobacchi C, Villa A. Genetics of Osteopetrosis.
Curr Osteoporos Rep. 2018 Feb;16(1):13-25. doi: 10.1007/s11914-018-0415-2. Review.

167. Palagano E, Zuccarini G, Prontera P, Borgatti R, Stangoni G, Elisei S, Mantero S, Menale C, Forlino A, Uva P, Oppo M, Vezzoni P, Villa A, Merlo GR, Sobacchi C. Mutations in the Neuroblastoma Amplified Sequence gene in a family affected by Acrofrontofacionasal Dysostosis type 1. Bone 2018 Sep;114: 125-136. doi: 10.1016/j.bone.2018.06.013.

168. Menale C, Campodoni E, Palagano E, Mantero S, Erreni M, Inforzato A, Fontana E, Schena F, Van't Hof R, Sandri M, Tampieri A, Villa A, Sobacchi C. Mesenchymal Stromal Cell-Seeded Biomimetic Scaffolds as a Factory of Soluble RANKL in Rankl-Deficient Osteopetrosis.
Stem Cells Transl Med. 2019 Jan;8(1):22-34. doi: 10.1002/sctm.18-0085.

169. Brigida I, Zoccolillo M, Cicalese MP, Pfajfer L, Barzaghi F, Scala S, Oleaga-Quintas C, Álvarez-Álvarez JA, Sereni L, Giannelli S, Sartirana C, Dionisio F, Pavesi L, Benavides-Nieto M, Basso-Ricci L, Capasso P, Mazzi B, Rosain J, Marcus N, Lee YN, Somech R, Degano M, Raiola G, Caorsi R, Picco P, Moncada Velez M, Khourieh J, Arias AA, Bousfiha A, Issekutz T, Issekutz A, Boisson B, Dobbs K, Villa A, Lombardo A, Neven B, Moshous D, Casanova JL, Franco JL, Notarangelo LD, Scielzo C, Volpi S, Dupré L, Bustamante J, Gattorno M, Aiuti A. T-cell defects in patients with ARPC1B germline mutations account for combined immunodeficiency.
Blood. 2018 Nov 29;132(22):2362-2374. doi: 10.1182/blood-2018-07-863431. Epub 2018 Sep 25.

170. Sobacchi C, Erreni M, Strina D, Palagano E, Villa A, Menale C. 3D Bone Biomimetic Scaffolds for Basic and Translational Studies with Mesenchymal Stem Cells.
Int J Mol Sci. 2018 Oct 13;19(10). pii: E3150. doi: 10.3390/ijms19103150. Review.

171.Villa A, Notarangelo LD. RAG gene defects at the verge of immunodeficiency and immune dysregulation Immunol Rev. 2019 Jan;287(1):73-90. doi: 10.1111/imr.12713. Review.

172. Pellicciotta M, Rigoni R, Falcone EL, Holland SM, Villa A, Cassani B. The microbiome and immunodeficiencies: Lessons from rare diseases. J Autoimmun. 2019 Mar; 98:132-148. doi: 10.1016/j.jaut.2019.01.008. Epub 2019 Jan 28. Review.

173. Crisafulli L, Muggeo S, Uva P, Wang Y, Iwasaki M, Locatelli S, Anselmo A, Colombo FS, Carlo-Stella C, Cleary ML, Villa A, Gentner B, Ficara F. MicroRNA-127-3p controls murine hematopoietic stem cell maintenance by limiting differentiation.
Haematologica. 2019 Feb 21. pii: haematol.2018.198499. doi: 10.3324/haematol.2018.198499.

174. Penna S, Capo V, Palagano E, Sobacchi C, Villa A. One Disease, Many Genes: Implications for the Treatment of Osteopetroses. Front Endocrinol (Lausanne). 2019 Feb 19; 10:85. doi: 10.3389/fendo.2019.00085. eCollection 2019. Invited Review.

175. Castelli A, Susani L, Menale C, Muggeo S, Caldana E, Strina D, Cassani B, Recordati C, Scanziani E, Ficara F, Villa A, Vezzoni P, Paulis M. Chromosome Transplantation: Correction of the Chronic Granulomatous Disease Defect in Mouse iPSCs. Stem Cells. 2019 Mar 20. doi: 10.1002/stem.3006. [Epub ahead of print]

176. Sereni L, Castiello MC, Di Silvestre D, Della Valle P, Brombin C, Ferrua F, Cicalese MP, Pozzi L, Migliavacca M, Bernardo ME, Pignata C, Farah R, Notarangelo LD, Marcus N, Cattaneo L, Spinelli M, Giannelli S, Bosticardo M, van Rossem K, D'Angelo A, Aiuti A, Mauri P, Villa A. Lentiviral gene therapy corrects platelet phenotype and function in Wiskott-Aldrich patients.

J Allergy Clin Immunol. 2019 Mar 26. pii: S0091-6749(19)30410-5. doi: 10.1016/j.jaci.2019.03.012. [Epub ahead of print]

177. Marcovecchio GE, Bortolomai I, Ferrua F, Fontana E, Imberti L, Conforti E, Amodio D, Bergante S, Macchiarulo G, D'Oria V, Conti F, Di Cesare S, Fousteri G, Carotti A, Giamberti A, Poliani PL, Notarangelo LD, Cancrini C, Villa A, Bosticardo M. Thymic Epithelium Abnormalities in DiGeorge and Down Syndrome Patients Contribute to Dysregulation in T Cell Development. Front Immunol. 2019 Mar 15; 10:447. doi: 10.3389/fimmu.2019.00447.

178. Ferrua F, Cicalese MP, Galimberti S, Giannelli S, Dionisio F, Barzaghi F, Migliavacca M, Bernardo ME, Calbi V, Assanelli AA, Facchini M, Fossati C, Albertazzi E, Scaramuzza S, Brigida I, Scala S, Basso-Ricci L, Pajno R, Casiraghi M, Canarutto D, Salerio FA, Albert MH, Bartoli A, Wolf HM, Fiori R, Silvani P, Gattillo S, Villa A, Biasco L, Dott C, Culme-Seymour EJ, van Rossem K, Atkinson G, Valsecchi MG, Roncarolo MG, Ciceri F, Naldini L, Aiuti A. Lentiviral haemopoietic stem/progenitor cell gene therapy for treatment of Wiskott-Aldrich syndrome: interim results of a non-randomised, open-label, phase 1/2 clinical study. Lancet Haematol. 2019 Apr 10. pii: S2352-3026(19)30021-3. doi: 10.1016/S2352-3026(19)30021-3.

179. Sobacchi C, Menale C, Villa A. The RANKL-RANK Axis: A Bone to Thymus Round Trip. Front Immunol. 2019 Mar 29; 10:629. doi: 10.3389/fimmu.2019.00629. eCollection 2019. Invited Review.

180. Cifaldi C, Brigida I, Barzaghi F, Zoccolillo M, Ferradini V, Petricone D, Cicalese MP, Lazarevic D, Cittaro D, Omrani M, Attardi E, Conti F, Scarselli A, Chiriaco M, Di Cesare S, Licciardi F, Davide M, Ferrua F, Canessa C, Pignata C, Giliani S, Ferrari S, Fousteri G, Barera G, Merli P, Palma P, Cesaro S, Gattorno M, Trizzino A, Moschese V, Chini L, Villa A, Azzari C, Finocchi A, Locatelli F, Rossi P, Sangiuolo F, Aiuti A, Cancrini C, Di Matteo G. Targeted NGS Platforms for Genetic Screening and Gene Discovery in Primary Immunodeficiencies. Front Immunol. 2019 Apr 11;10: 316. doi: 10.3389/fimmu.2019.00316. eCollection 2019.

181. Daley SR, Koay HF, Dobbs K, Bosticardo M, Wirasinha RC, Pala F, Castagnoli R, Rowe JH, Ott de Bruin LM, Keles S, Lee YN, Somech R, Holland SM, Delmonte OM, Draper D, Maxwell S, Niemela J, Stoddard J, Rosenzweig SD, Poliani PL, Capo V, Villa A, Godfrey DI, Notarangelo LD. Cysteine and hydrophobic residues in CDR3 serve as distinct T-cell self-reactivity indices. J Allergy Clin Immunol. 2019 Apr 30. pii: S0091-6749(19)30475-0. doi: 10.1016/j.jaci.2019.03.022. [Epub ahead of print] No abstract available.

182. Capo V, Penna S, Merelli I, Barcella M, Scala S, Basso-Ricci L, Draghici E, Palagano E, Zonari E, Desantis G, Uva P, Cusano R, Sergi Sergi L, Crisafulli L, Moshous D, Stepensky P, Drabko K, Kaya Z, Unal E, Gezdirici A, Menna G, Serafini M, Aiuti A, Locatelli SL, Carlo-Stella C, Schulz AS, Ficara F, Sobacchi C, Gentner B, Villa A. Expanded circulating hematopoietic stem/progenitor cells as novel cell source for the treatment of TCIRG1 osteopetrosis. Haematologica. 2020 Jan 16. pii: haematol.2019.238261. doi: 10.3324/haematol.2019.238261.

183. Paulis M, Susani L, Castelli A, Suzuki T, Hara T, Straniero L, Duga S, Strina D, Mantero S, Caldana E, Sergi LS, Villa A, Vezzoni P. Chromosome Transplantation: A Possible Approach to Treat Human X-linked Disorders. Mol Ther Methods Clin Dev. 2020 Jan 21; 17:369-377. doi: 10.1016/j.omtm.2020.01.003. eCollection 2020 Jun 12.

184. Mikulak J, Oriolo F, Bruni E, Roberto A, Colombo FS, Villa A, Bosticardo M, Bortolomai I, Lo Presti E, Meraviglia S, Dieli F, Vetrano S, Danese S, Della Bella S, Carvello MM, Sacchi M,

Cugini G, Colombo G, Klinger M, Spaggiari P, Roncalli M, Prinz I, Ravens S, di Lorenzo B, Marcenaro E, Silva-Santos B, Spinelli A, Mavilio D. NKp46-expressing human gut-resident intraepithelial Vδ1 T cell subpopulation exhibits high antitumor activity against colorectal cancer. JCI Insight. 2019 Dec 19;4(24): e125884. doi: 10.1172/jci.insight.125884.

185. Lanzi G, Ferraro RM, Masneri S, Piovani G, Barisani C, Sobacchi C, Villa A, Vezzoni P, Giliani S. Generation of 3 clones of induced pluripotent stem cells (iPSCs) from a patient affected by Autosomal Recessive Osteopetrosis due to mutations in TCIRG1 gene. Stem Cell Res. 2020 Jan;42: 101660. doi: 10.1016/j.scr.2019.101660. Epub 2019 Nov 20.

186. Palagano E, Muggeo S, Crisafulli L, Tourkova IL, Strina D, Mantero S, Fontana E, Locatelli SL, Monari M, Morenghi E, Carlo-Stella C, Barnett JB, Blair HC, Vezzoni P, Villa A, Sobacchi C, Ficara F. Generation of an immunodeficient mouse model of tcirg1-deficient autosomal recessive osteopetrosis. Bone Rep. 2020 Jan 7;12: 100242. doi: 10.1016/j.bonr.2020.100242. eCollection 2020 Jun.

187. Delmonte OM, Villa A, Notarangelo LD. Immune dysregulation in patients with RAG deficiency and other forms of combined immune deficiency. Blood. 2020 Feb 27;135(9):610-619. doi: 10.1182/blood.2019000923.

188. Rigoni R, Fontana E, Dobbs K, Marrella V, Taverniti V, Maina V, Facoetti A, D'Amico G, Al-Herz W, Cruz-Munoz ME, Schuetz C, Gennery AR, Garabedian EK, Giliani S, Draper D, Dbaibo G, Geha RS, Meyts I, Tousseyn T, Neven B, Moshous D, Fischer A, Schulz A, Finocchi A, Kuhns DB, Fink DL, Lionakis MS, Swamydas M, Guglielmetti S, Alejo J, Myles IA, Pittaluga S, Notarangelo LD, Villa A*, Cassani B*. Cutaneous Barrier Leakage and Gut Inflammation Drive Skin Disease in Omenn Syndrome. J Allergy Clin Immunol. 2020 Apr 17; S0091-6749(20)30492-9. doi: 10.1016/j.jaci.2020.04.005.

189. Garcia-Perez L, van Eggermond M, van Roon L, Vloemans SA, Cordes M, Schambach A, Rothe M, Berghuis D, Lagresle-Peyrou C, Cavazzana M, Zhang F, Thrasher AJ, Salvatori D, Meij P, Villa A, Van Dongen JJM, Zwaginga JJ, van der Burg M, Gaspar HB, Lankester A, Staal FJT, Pike-Overzet K. Successful Preclinical Development of Gene Therapy for Recombinase-Activating Gene-1-Deficient SCID. Mol Ther Methods Clin Dev. 2020 Mar 31;17: 666-682. doi: 10.1016/j.omtm.2020.03.016. eCollection 2020 Jun 12.

190. Castiello MC, Bosticardo M, Sacchetti N, Calzoni E, Fontana E, Yamazaki Y, Draghici E, Corsino C, Bortolomai I, Sereni L, Yu HH, Uva P, Palchaudhuri R, Scadden DT, Villa A, Notarangelo LD. Efficacy and safety of anti-CD45-Saporin as conditioning agent for RAG deficiency. J Allergy Clin Immunol. 2020 May 5:S0091-6749(20)30629-1. doi: 10.1016/j.jaci.2020.04.033. Online ahead of print. PMID: 32387109

191. Marita Bosticardo, Francesca Pala, Enrica Calzoni, Ottavia M. Delmonte, Kerry Dobbs, Cameron Gardner, Nicolo' Sacchetti, Tomoki Kawai, Elizabeth Garabedian, Debbie Draper, Jenna Bergerson, Suk See DeRavin, Alexandra F. Freeman, Tayfun Guengoer, Nicholas Hartog, Steven M. Holland, Donald B. Kohn, Harry L. Malech, Mary Louise Markert, Katja G. Weinacht, Anna Villa, Christopher S. Seet, Amelie Montel-Hagen, Gay M. Crooks, Luigi D. Notarangelo. Artificial thymic organoids represent a reliable tool to study T cell differentiation in patients with severe T cell lymphopenia. Blood Advances in press

192.Ritelli M, Palagano E, Cinquina V, Beccagutti F, Chiarelli N, Strina D, Hall IF, Villa A, Sobacchi C, Colombi M. Genome-first approach for the characterization of a complex phenotype

with combined NBAS and CUL4B deficiency. Bone. 2020 Nov;140:115571. doi: 10.1016/j.bone.2020.115571. Epub 2020 Aug 6.

193. Di Zanni E, Palagano E, Lagostena L, Strina D, Rehman A, Abinun M, De Somer L, Martire B, Brown J, Kariminejad A, Balasubramaniam S, Baynam G, Gurrieri F, Pisanti MA, De Maggio I, Abboud MR, Chiesa R, Burren CP, Villa A, Sobacchi C, Picollo A. Pathobiologic Mechanisms of Neurodegeneration in Osteopetrosis Derived From Structural and Functional Analysis of 14 ClC-7 Mutants. J Bone Miner Res. 2020 Oct 30. doi: 10.1002/jbmr.4200. Online ahead of print.

194. Villa A, Capo V, Castiello MC. Innovative Cell-Based Therapies and Conditioning to Cure RAG Deficiency. Front Immunol. 2020 Nov 19;11:607926. doi: 10.3389/fimmu.2020.607926. eCollection 2020.

195. Vavassori V, Mercuri E, Marcovecchio GE, Castiello MC, Schiroli G, Albano L, Margulies C, Buquicchio F, Fontana E, Beretta S, Merelli I, Cappelleri A, Rancoita PM, Lougaris V, Plebani A, Kanariou M, Lankester A, Ferrua F, Scanziani E, Cotta-Ramusino C, Villa A, Naldini L, Genovese P.Modeling, optimization, and comparable efficacy of T cell and hematopoietic stem cell gene editing for treating hyper-IgM syndrome. EMBO Mol Med. 2021 Jan 21:e13545. doi: 10.15252/emmm.202013545. Online ahead of print. PMID: 33475257

Book Chapters

Encyclopedia of Immunobiology. Edited by Elsevier. Autoimmune Polyendocrinopathy-Candidias-Ectordermal Dystrophy (APECED). In press

Stiehm's Immune Deficiencies. Edited by: E.R Stiehm& K.E Sullivan. Chapter 4: T Cell Defects page 88-126. Elsevier, 2014

Primary Immunodeficiency Diseases. A molecular and genetic approach. Third Edition. Edited by H.D Ochs, E. Smith, JM Puck . Chapter 13: V(D)J Recombination Defects. Page 168-187. Oxford, 2014

Primary Immunodeficiency Diseases: Definition, Diagnosis and Management. Second Edition. Edited by N. Rezaei, A. Aghamahammadi, LD Notarangelo. Springer, 2015