

Jolan E. Walter, MD, PhD

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Citizenship

U.S. Citizen

Education

University of Pecs, Hungary, Virology, Ph.D.	10/96-10/02
University of Pecs, Hungary, Medicine, M.D.	09/90-09/96

Postgraduate Training

Boston Children's Hospital, Boston, MA, Pediatric Allergy Immunology Fellowship	07/06-06/09
Children's Hospital of the King's Daughters, EVMS, Norfolk, VA Pediatric Residency, Pediatrics	07/03-06/06
Center of Pediatric Research, EVMS, Norfolk, VA, Research Fellow in Infectious Diseases	06/98-04/01

Licensure and Board Certification

Florida Medical License (active) (#ME127615)	01/18-01/20
Massachusetts Medical License (inactive) (#233737)	07/06-06/16
Virginia Medical License (inactive) (#0116014429)	03/02-06/06
U.S. Foreign Medical Graduate Examination (active) (#0-539-027-3)	10/98-Present
Hungarian Medical License (inactive)	10/96
American Board of Pediatrics (recertified, 2023) License #666979	2006
American Board of Allergy and Immunology (recertified, 2028) License #91647	2008

Awards, Honors, Honorary Society Memberships

Fellow of Clinical Immunology Society, Elected Member	2021
Advocacy Committee, Society of Pediatric Research	2021
Member, Variant Curation Expert Panel (VCEP) for SCID	2020
Steering Committee Member, SCID Compass, Immune Deficiency Foundation	2019
Steering Committee Member, NICER	2019
"Americas Top Doctors" – Castle Connolly	2019
John W. Hadden MD Young Investigator Award, University of South Florida, Research	2017
Faculty Inductee, Robert A. Good Honor Society, University of South Florida, Research	2017

“Excellence in Action”, Massachusetts General Hospital, Clinical Care	2015
Society for Pediatric Research, Elected Member	2014
K08 Career Award, NIAID, National Institutes of Health, Research	2012
Nathan Talbot Pediatric Research Award, MassGeneral Hospital <i>for</i> Children, Research	2012
Junior Faculty Award, Clinical Immunology Society, Research	2011
Partners in Excellence, Massachusetts General Hospital, Clinical care	2010
Travel Award, Advanced Course of Basic and Clinical Immunology, Scottsdale, AZ, Clinical Care	2009
Fellows Career Development Award, GSK-American Academy of Allergy, Asthma and Immunology, Research	2008
Young Faculty Research Award, Southern Society for Pediatric Research, Research	2002
Travel Award, Seventh European Workshop on Virus Evolution and Molecular Epidemiology, Leuven, Belgium, Research	2001
Perry Morgan Fellow, Center for Pediatric Research, EVMS, Norfolk, VA, Research	2000
Travel Award, Annual Conference of America Society of Virology, Research	1999, 2000
First Prize Award by the Dean, Pécs University Medical School, Thesis	1995
Scholarship of the Republic of Hungary, Government (Republic of Hungary), Outstanding academic performance	1994-96

Appointments

Academic Appointments

Associate Professor, Genomics Program, University of South Florida	05/19-Present
Associate Professor, Molecular Medicine, University of South Florida	12/18-Present
Faculty (joint BMT/Immunology clinic), Moffitt Cancer Center	10/18-Present
Associate Professor of Pediatric (adjunct), Johns Hopkins Medical Institute	10/16-Present
Director, Jeffrey Modell Diagnostic and Research Center for Primary Immunodeficiencies, Johns Hopkins All Children’s Hospital	03/16-Present
Robert A. Good Endowed Chair	03/16-Present
Associate Professor, Department of Pediatrics, University of South Florida	03/16-Present
Consultant in Immunology Research, Pediatric (Allergy/Immunology) Massachusetts General Hospital <i>for</i> Children	03/16-Present
Associate in Pediatrics, Pediatrics (Allergy/Immunology) Massachusetts General Hospital	08/15-02/16
Associate Investigator, Nutrition Obesity Research Center, Harvard Medical School	09/14-02-16
Principal Investigator, Center for Immunology and Inflammatory Disease (CIID), Massachusetts General Hospital	11/13-02/16
Assistant in Pediatrics, Pediatrics (Allergy/Immunology), Massachusetts General Hospital	07/09-08/15

Administrative Appointment

Vice Chair, AAAAI	3/22-present
Division Chief, Pediatric (Allergy/Immunology), John Hopkins All Children’s Hospital	05/16-Present
Division Chief, Pediatric (Allergy/Immunology) University of South Florida	03/16-Present
Director, Immunodeficiency Program, Division of Allergy/Immunology, Massachusetts General Hospital <i>for</i> Children	04/14-02/16

Hospital

Staff Physician, Immunology, Moffitt Cancer Center	08/18-Present
Staff Physician, Pediatrics, Johns Hopkins All Children's Hospital	10/16-Present
Staff Physician, Pediatrics, Franciscan Hospital for Children, Brighton, MA	10/07-10/16

Teaching, Lecture**Undergraduate Medical**

Immune Deficiency Cases, 2-hr sessions, Morsani College of Medicine, USF	03/19-present
Cellular and Molecular Immunology (HST-175), Harvard Medical School, three 1-hr sessions per year on topics related to Clinical Immunology	2010-2016

Graduate Medical (residents, fellows)

Noon teaching conference for residents and medical students, Massachusetts General Hospital <i>for</i> Children, three 1-hr sessions per year on topics related to Immunology	08/2018
Morning teaching conference for Allergy/Immunology fellows, University of South Florida and Johns Hopkins All Children's Hospital, five 1-hr sessions per year on topics related to Immunology: (1) Immunodeficiency 101, (2) B cell phenotypes and function, (3) T cell phenotypes and function, (4) Gene therapy, (5) Immune phenotyping and genetic approach in primary immunodeficiencies	2016-present
Noon teaching conference for residents (USF, JHACH) University of South Florida and Johns Hopkins All Children's Hospital, one to two 1-hr sessions per year on topics related to Immunology (Primary Immunodeficiency 101)	2016-present

Hospital

Annual Grand Rounds presentation for USF Medicine (Dermatology, Pediatrics), Johns Hopkins All Children's Hospital (Medicine, Neonatology) on newborn screening for SCID and primary immunodeficiencies	2016-present
Moffitt Cancer Center, ad hoc talks on primary immunodeficiencies and HSCT	2018-present

Teaching, Supervisory**Clinical supervisory and training responsibilities**

Supervision of residents in Pediatric Allergy/Immunology clinics, 10 residents per annum. USF and JHACH Ped and Med/Peds Programs (5% effort)	2016-present
Supervision of Allergy/Immunology fellows on inpatient wards rotations and in Pediatric Allergy/Immunology clinics (1 or 2 fellows from USF Peds AI and 3 fellows from USF Medicine AI per annum) (10% effort)	2016-present

Clinical trainees in Pediatric Allergy/Immunology:

Several of our trainees were mentored to present abstract at national meetings and participate in CIS-PID school

Jenny Yoon, Sheila Bina, Adeeb Bulkhi	2016
Bhumika Patel, Joanna Castro-Wagner, Peter Ricketti, Chen Lin, Sultan Alandijani	2016-2017
David Lindsay, Sonia Joychan, Kirk Shepard, Amber Pepper, Emma Westermann	2016-2018
Fatima Khan, Warit Jithpratuck, Tara Saco, Shiven Patel	2017-2019
Irina Dawson, Jacqueline Squire, Raul Villareal, Claudia Gaefke	2018-2020

Maria Chitty Lopez, Stephanie Hudey, Nicholas Kolinsky, David Gubernick	2019-2021
Mary Ann Miranda, Priya Timothy, Donya Imanirad, Natalie Diaz-Cabrera	2020-present
Shannon Sullivan, Silpa Taunk	2021-present

<u>International clinical trainees in Pediatric Allergy/Immunology (visiting):</u>	2018-2020
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Visiting international fellows and faculty:

Dr Benz(Pailin Yooma) and Dr Oui (Thitaya Sangsawang) Yada Treepaiboon.	2020
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Nattiya Kitiphapat, Jose Roberto M. Pegler, Cristiano Góes do Nascimento and Maira Mastrocola de Campos.	2019
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Nuntanut Rutrakool, Thunyatorn Phisitbuntoon, Teerapol Chantveerawong, Isree Leelayuwattanakul, Pamela F. Alves Barbosa, Mariana Castiglioni, Nathalia Mota, Maram Albanyan, Renan Perera, Renata Guardiano Dias Lais Millan and Mariana Fernandes Barp Melnik.	2018
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Laboratory or other research supervisory, training Formally supervised trainees

Lael Yonker MD , Clinical fellow in Pulmonology / Massachusetts General Hospital <i>for</i> Children. Provide formal mentorship, supervision and career guidance in research in the field of common variable immunodeficiency. She presented our findings at CIS (2011) and co-author on the summary of these findings published in <i>Frontiers of Immunology</i> (2017). Current position: Assistant Professor at Harvard (MGH)	2010-2013
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Melissa Walker MD PhD , Resident / Massachusetts General Hospital <i>for</i> Children (now Instructor at Harvard). Provide formal mentorship, supervision and career guidance in research in the field of immunodeficiency in mitochondrial disorders. She presented our findings at CIS (2013) and her manuscript was accepted in <i>JACI: In Practice</i> (2014). She completed a review on mitochondrial disease and the immune system at <i>Journal of Immunology Research</i> (2014). She is co-author on an additional case review. I was a grant mentor for Dr. Walker who has received a Publication Research Grant from CSL-Behring to assess cost-benefit of immunoglobulin replacement for patients with recurrent infections, immune dysfunction and mitochondrial disorder. Current position: Instructor, Harvard (MGH)	2012-2016
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Tim Lax, MD , Clinical Fellow in Allergy/Immunology / Massachusetts General Hospital <i>for</i> Children (now Instructor at Harvard). Provide formal mentorship, supervision and career guidance in research in the field of dyskeratosis congenita. Dr. Lax presented our findings at CIS (2013) and CIS-PID School (2013). Case report has been presented as CPC series at Massachusetts General Hospital in May 2014 and was invited for publication at <i>The New England Journal of Medicine</i> (2015). Current position: Instructor, Harvard (BIDMC)	2012-2014
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- Samira Farough MD**, Foreign graduate (now Research Fellow in Radiology, MGH). 2012-2014
 Provided formal mentorship, supervision and career guidance in research in the field of immunodeficiency in mitochondrial disorders
 She presented our findings at CIS (2013) and has a case report with Coenzyme Q10 deficiency and immune dysfunction (*Clinical Immunology*, 2014). Current position: Director of Medical Review, Imaging Endpoints (AZ)
- Ottavia Delmonte MD PhD**, Resident/ Boston Children's Hospital (now Staff Clinician, NIAID, NIH) PhD thesis advisor, and providing supervision and career guidance in research in the field of immunodeficiency, in particular AID deficiency. 2013-2016
 She presented her data as oral abstract at AAAAI 2014 meeting and defended her PhD thesis on AID deficiency in Italy in April 2014. Current position: Assistant Professor, National Institutes of Health.
- Boglarka Ujhazi MS**, PhD candidate, Research technician II, Walter laboratory, MGH. Studies on celiac-like enteropathies in primary immunodeficiencies in human and mice models. She oversees laboratory operations, computational analysis of B cell repertoire. She has several platform and poster presentations at national meetings and coauthor on several publications. Co-mentor for PhD training. Current position: Biological Scientist, Walter laboratory, USF 2013-present
- Katrina Abernethy MD**, Co-supervisor in her Master in Research entitled "Measurement and characterisation of autoantibodies by high capture fluorescence microarray in X-linked chronic granulomatous disease carriers" visiting medical student from Institute of Cellular Medicine, Newcastle University, Newcastle-upon-Tyne, UK. She completed her thesis with distinction and had poster accepted to the biannual meeting of European Society of Primary Immunodeficiencies (ESID) in October 2014. Current position: Unknown 2014 Apr-May
- George Zhang**, 1st year undergraduate student, Harvard University (Pre-Med). 2014 Jun
 Walter laboratory, MGH. Human studies of autoreactive cells in Rag deficiency: cloning and sequencing monoclonal heavy and light chain from patient with Rag deficiency. Current position: Software engineer, Google (CA)
- Zachary Fitzpatrick**, graduate student, Louisiana State University (Biochemistry, Political Science). Walter laboratory, MGH. Clinical studies: Genetic studies in primary immunodeficiencies, in particular TRECS and KRECS in SCID, T cell lymphopenia and combined immunodeficiencies (CID). Laboratory experiment: Sequence analysis of B cell repertoire of patients with RAG deficiencies and analysis of epigenetic modifiers in disease. Zach is currently a Fulbright Scholar, studying therapeutic gene transfer and immune deficiencies at the Pasteur Institute in Paris, France and will continue graduate work at the University of Cambridge as a Gates Cambridge Scholar. Current position: Fulbright Scholar, Université Pierre-et-Marie-Curie and the Pasteur Institute, France 2015-2016
- Omri Rotem**, 4th year medical student, Debrecen University Medical School, Hungary. Walter laboratory, MGH. Human studies of autoreactive cells in RAG deficiency. Amplification of heavy and light chain of single B cells of naïve and 2014 Jul

mature compartment from patients with RAG deficiencies. Current position
Emergency Medicine Resident, Sheba Tel HaShomer City of Health, Israel

Carin Dahlberg, Graduate student, Karolinska Institute, Stockholm, Sweden. Walter 2014 Aug
laboratory, MGH. Dysregulation of innate lymphocytes and induction of
autoimmunity in murine model of RAG deficiency. We published jointly on immune
dysregulation and autoantibodies in Wiskott-Aldrich mouse model. Current position:
Project Manager, Karolinska Institute, Sweden

Zsofia Foldvari MD, 6th year medical student, Debrecen University Medical School, 2015-2016
Hungary. Walter laboratory, MGH. Human studies of autoreactive cells in RAG
deficiency. Cloning of heavy and light chain of single B cells of naïve and mature
compartment from patients with RAG deficiencies. Assistance with murine models of
RAG deficiency. Zsofia is first co-author on treatment outcomes of autoimmune
cytopenias and other autoimmune disorders in RAG deficiency published in *JACI: In
Practice* (2019). Current position: PhD student, K.G. Jebsen Center for Cancer
Immunotherapy, Oslo University Hospital, Norway

Christoph Geier MD PhD, medical student from Medical School of Vienna, 2015-present
Austria. Christoph has collected and presented our data on adult RAG patient with as
a moderated poster presentation at the European Society of Immunodeficiency
(ESID) meeting in 2016 and published our findings as first co-author in the *Journal
of Allergy and Clinical Immunology* (2018). This was an extensive global
collaboration between investigators from the United States of America and Europe
and Christoph has been instrumental in communication with the sites. We also
collaborated with Christoph and his mentors, Dr. Wolf and Dr. Eibl on common
variable immunodeficiency (CVID) and Christoph published a first author paper on
“deficiency of group 2 innate lymphoid cells in patients with common variable
immunodeficiency” (*European Journal of Immunology*, 2017). In 2019, Christoph
presented on complications of vasculitis in RAG deficiency at AAAAI and published
our findings in the *Frontiers of Immunology* (2020). Current joing work includes the
natural history of WHIM syndrome and B cell studies in WHIM patients. He will
complete a manuscript and assist with genetic studies on SCID patients from Brazil.
Current position: Postdoctoral Fellow and Clinician

Bhumika Patel MD, Fellow in Allergy Immunology, University of South Florida, 2016-2017
supervision and career guidance in research in the field of immunodeficiency. Dr.
Patel submitted an abstract on RAG deficient patient cohort with autoimmune
cytopenia and is a co-author on a publication in *JACI: In Practice* (2018). Current
position: Private Allergist Immunologist, Tampa, FL

Anna Meyer MD PhD, Pediatric Resident, Johns Hopkins All Children’s Hospital, 2016-2018
supervision and career guidance in research in the field of autoimmunity and
immunodeficiency. Dr. Meyer presented our retrospective dataset on autoimmune
cytopenia with or without primary immunodeficiencies at a national meeting and is
primary author on a publication in draft on this dataset. She also has submitted
abstracts to 2018 CIS meeting on leaky SCID patients identified by newborn
screening that was published in *Frontiers of Pediatrics* (2019). Current position:
Assistant Professor, Allergy & Immunology, National Jewish Health

Bence Zakota, Undergraduate Student, University of South Florida. Bence has worked for over a year in our laboratory as a student researcher. He has won a Poster Award for Undergraduate Research in Allergy, Immunology and Infectious Diseases in 2018 USF Health Research Day. Current position: Medical Student 2017-2018

Cristina Meehan, Graduate Student, University of South Florida. Cristina has worked for the past two years in our laboratory as a clinical coordinator and student researcher. She contributed to writing grants to Foundations and have published two papers with our team (*Rev Paul Pediatr* (2018), *Frontiers of Pediatrics* (2019)). Her work was focusing on immunological and genetic studies in our retrospective and prospective dataset on autoimmune cytopenia with or without primary immunodeficiencies; description of leaky SCID patients identified by newborn screening and global awareness of SCID. She contributed to several grant applications. She has also represented our team at several local conferences (USF, JHACH). Current position: MD PhD student, at University of Alabama, Birmingham 2017-2019

Joseph Dasso MD PhD, Staff Scientist, Walter laboratory, USF. Dr. Dasso's main focus is the microbiome in humans and animal models of PID. I have been mentoring Dr. Dasso on human studies in RAG deficiency and immune dysregulation. He has presented posters at meetings, has been assisting with eight grant applications (institutional, private and federal [R01] sources) and is a co-author on 6 publications in the Walter lab. In particular, he co-wrote and successfully applied with me for the Rothman grant (2019) where we plan to correlate the microbiome of children with PANDAS to their immune status (IgA deficiency) and an R01 on age-associated B cells. Current position: Staff Scientist, Walter laboratory 2017-present

Rachel Cruz, Undergraduate Student, University of South Florida. Rachel has worked for the past two years in our laboratory as a student researcher and in May 2019 started as research coordinator. She has presented at 2018 CIS meeting a case series of patients with cardiac transplant and parvovirus viremia and published her findings in the *Open Forum of Infectious Diseases*. Currently she is working on the manuscript on natural history of WHIM syndrome and ovarian cancer in CVID. She has also represented our team at several local conferences (USF, JHACH). Current position: Medical Student, USF 2017-present

Maryssa Ellison, BSc, Research Coordinator, University of South Florida. Maryssa has worked for 4 years in our laboratory as as research coordinator. She has presented at several local and national meetings on WHIM syndrome. Currently she is working on the manuscript on natural history of WHIM syndrome Current position: PhD student, Wake-Forest University, Molecular and Cellular Biological Sciences, Winston-Salem, NC 2017-2021

Kevin Wu, Medical Student, University of South Florida. Supervisor for summer elective Scholarly Concentration Program (SCP) administrated by the office of Research, Innovation & Scholarly Endeavors (RISE) (2018). Kevin has worked all summer in our laboratory on impaired tolerance checkpoints and polyreactive B cells, represented our team at several local conferences with platform presentations (USF, 2017-2020

JHACH) and in webinar setting for CIS. He received the “Best Oral Presentation Award” at 2019 USF Research Day and has a first author paper published on treatment approach for two patients with RAG deficiency and arthritis (*Frontiers of Pediatrics* (2019)). He was selected to the Robert A. Good Honor Society as a graduating medical student. Current position: Diagnostic Radiology, USF

Claudia Gaefke MD, Fellow in Allergy Immunology, University of South Florida, Supervisor and career mentor in research in the field of immunodeficiency. Dr. Gaefke presented at several meetings on our patients with primary immunodeficiency (AAAAI 2019, CIS 2019) and currently preparing publications on patients with ALPS. Current position: Specialist in Allergy Immunology, Tucson, AZ 2018-2020

Pooja Purswani MD, Resident and JHACH. Supervisor and career mentor in research in the field of immunodeficiency. Dr. Purswani has published two case reports on immunodeficiencies in the *Frontiers of Immunology* and submitted abstracts to our national AAAAI meeting. Current position: Clinical Fellow in Pediatric Allergy Immunology, Duke Medical School 2018-2021

Priya Patel MD, Resident and JHACH. Career mentor research in the field of immunodeficiency and immune dysregulation, specifically with partial DiGeorge syndrome and autoimmune cytopenias. Dr. Patel has presented several abstracts at our national AAAAI and CIS meeting. Manuscript was invited and are in process for submission to *Frontiers of Immunology*. Current position: Resident at JHACH 2020-present

Priya Timothy MD, Fellow in Allergy Immunology, University of South Florida, supervision and career guidance in research in the field of immune evaluation in food allergy and immune dysregulation. Dr. Patel submitted an abstract on CTLA4 deficiencies. She is spearheading a funded internal grant on immune evaluation among patients with peanut oral immunotherapy. Current position: Fellow in Allergy and Immunology, USF, Tampa, FL 2020-present

(David) Evan Potts, Medical Student, University of South Florida. Supervisor for summer elective Scholarly Concentration Program (SCP) administrated by the office of Research, Innovation & Scholarly Endeavors (RISE) (2021). Evan has worked all summer in our laboratory on three SCID genes discovered among Irish Travelers in the US. He is key personnel on the project above that is funded by the SCID Compass, Immune Deficiency Foundation. Also assists in studies in other SCID variants (IL2RG), WHIM patient cohorts, Immune responses in patients infected or immunized with SARS-CoV2 and RAG recombinase activity measurements. Current position: Medical Student, USF 2021-present

Kady Palmer, Medical Student, Drexler University, Philadelphia, Pennsylvania. Supervisor for summer elective on pathomechanism for clinical variability in the Mennonite population with hypomorphic RAG variant, cohort of patients with SCID variants (IL2RG) and human microbiome studies in primary immunodeficiencies. Current position: Medical Student, Drexler University, Philadelphia, Pennsylvania 2021-present

Melis Yilmaz, volunteer intern, Walter laboratory, USF. Mentor on projects on WHIM syndrome including natural history and pathomechanisms for B cell 2021-present

abnormalities. Current position: Medical Student, Drexler University, Philadelphia, Pennsylvania

Peter Illes MD (from Hungary), clinical coordinator, Walter laboratory, USF. 2021-present
Mentor on projects on WHIM syndrome including natural history and pathomechanisms for B cell abnormalities. Current position: Research Coordinator, Allergy and Immunology, Tampa, FL

Mentoring faculty

Krisztian Csomos PhD, Research Associate, Walter laboratory (USF). I started 2013-present
mentoring Dr. Csomos as Postdoctoral Fellow at MGH in the area of human and animal studies of autoreactive cells in RAG deficiency. Krisztian is co-author is on several publications in *Journal of Clinical Investigations*, *JACI*, *JACI: In Practice*. He established multiple techniques in our laboratory and prepares data for first author publication on impaired tolerance mechanisms in RAG deficiency. He has several platform and poster presentations at national meetings. Highly productive team member and leader of the Walter laboratory operations. He had received grant internal funding from USF in 2019. Current position: Research Associate (USF).

Jocelyn Farmer MD PhD, Supervision and career guidance in research in the field 2013-present
of immunodeficiency, in particular CVID. She assisted in preparing my presentation for the USIDNet Face-to-Face meeting at NIH (Jan 2014). Grant mentor for Dr. Farmer who has received a Publication Research Grant from CSL-Behring to assess immunoglobulin dosing among patients with infectious and non-infectious complications and CVID (1) and Grant from USIDNet/Baxter on comparison of population-based MGH CVID cohort to USIDNet data. She is invited to USIDNet Face-to-Face meeting at NIH on CVID data at MGH (Jan 2016). First and co-author on several papers and chapters in *Frontiers of Immunology*, *JACI* and *JACI: In Practice*, *JAMA* and *Cell Signaling* (see below). Jocelyn submitted a K08 application to NIAID in 2020 under co-mentorship with Dr. Shiv Pillai and me. Current position: Assistant Professor at Harvard Medical School and Massachusetts General Hospital

Sara Barmettler MD, Supervision and career guidance in research in the field 2015-present
of immunodeficiency. First author publication in *JAMA Open Access* adverse outcome of Rituximab at Massachusetts General Hospital. Currently, she is working on AAAAI guidelines on hypogammaglobulinemia after Rituximab. She also published an XLA cohort study with USIDNet, approach to patients with ataxia telangiectasia identified by newborn screening for SCID. Currently we are working on joint projects on WHIM syndrome. I am mentoring Dr. Barmettler on a funded K23 on CAR-T cells and B cell immune reconstitution. Current Position: Assistant Professor at Harvard Medical School and Massachusetts General Hospital

Panida Sriaroon MD, Supervision and career guidance in for independence in 2016-present
research and leadership role. Under my mentorship she is leading a successful Pediatric Allergy & Immunology Fellowship Program. She established and currently leads the Food Allergy Center at JHACH. She serves as local co-Director of the FARE program, became PI in several sponsored studies related to food allergy (e.g. eosinophilic esophagitis) and in collaboration with academic centers, such as Baylor

Medical School, she is now participating in a R32 grant and NIH U grant applications. She submitted a grant on mechanistic studies in peanut OIT to JHACH. We have co-authored several papers. Current Position: Associate Professor, Division of Allergy & Immunology, USF.

Adeeb Bulkhi, MD, Supervision and career guidance in for building his practice in clinical immunology and to become a national leader in Immunology in Saudi Arabia. We have abstracts at national meetings, co-authored several peer reviewed papers, jointly edited chapter for the Encyclopedia of Immunology and he is participating in our book “Genetics 101 for the Practicing Immunologist”. Current Position: Adjunct Professor at USF, Assistant Professor, Umm Al Qura University and Consultant, International Medical Center, Jeddah, Saudi Arabia. Current Position: Assistant Professor, Umm Al Qura University 2017-present

Vera Goda, MD, Supervision and career guidance, outside mentor for PhD thesis in translational medicine. Dr. Goda is an Immunologist with strong interest in primary immunodeficiencies. Dr. Goda is the first author on our joint publication in JACI-IP (2018). We have active collaboration on variants of SCID and PI3K deficiencies. Current position: Assistant Professor, Bone Marrow Transplant Unit, St Laszlo Hospital 2018-present

Svetlana Sharapova PhD, Supervision and career guidance in human studies in RAG deficiency and variants of SCID. Dr. Sharapova had a poster presentation at CIS 2019 with us. We are working on several papers. I have also mentored Dr. Sharapova in a successful grant application to the Jeffrey Modell Foundation in 2019. As part of this grant, she has visited our laboratory in summer 2019. She published as two author paper under my mentorship in the *Frontiers in Immunology* (2020). Current Position: Visiting Faculty Scientist, Walter laboratory, USF and Principal Investigator, Cancer Institute, Minsk, Belarus. 2018-present

Emma Westermann-Clark MD, Supervision and career guidance in primary immunodeficiencies with autoimmune cytopenias. She presented at several national meetings (CIS, PIDTC). Emma oversees our retrospective and prospective studies on patients with autoimmune cytopenias with specific emphasis for those with underlying immunodeficiency. She has published a first author publication and served as co-author on papers with RAG deficiency and autoimmune cytopenias (*Journal of Clinical Immunology* 2018, *JACI: In Practice* 2019). She has also been working with us on NIH grant applications. Current Position: Adjunct Research Faculty in Allergy/Immunology 2018-present

Marton Keszei PhD, Adjunct Faculty, Walter laboratory, USF. Dr. Keszei was visiting us from the Karolinska Institute in Sweden. He is being mentored by me on human studies in WHIM syndrome. Dr. Keszei has written an Investigator Initiated Grant to X4 with us and will continue collaboration on fitness of T and B cells in WHIM syndrome 2018-present

Anne Szymanski MD, Clinical Instructor, Johns Hopkins All Children’s Hospital. Dr. Szymanski has trained at the NIH and currently is an academic Rheumatologist with expertise in translational research. Under my joint mentorship with Dr. Diana 2019-present

Milojevic, Dr. Szymanski has submitted an Institutional Grant at JHACH for research on antibody response in patients with immunodeficiency or immune modulation at JHACH.

Joud Hajjar, MD, Assistant Professor, Baylor Medical School (PhD student) 2020-present
Dr. Hajjar is an Allergist/Immunologist with strong interest in translational medicine. She is currently preparing a K23 as we support her with samples and consultation on the microbiome of patients with CVID and non-infectious complications.

Formal teaching of peers (e.g. CME, other continuing ed) (CME)

Food allergies: A growing epidemic Primary Care Pediatric Conference, Harvard Medical School	2009
Severe case of systemic mastocytosis, MGH Chief of Service Round. MGH staff	2010
SCID and newborn screening update. Porter Lecture MGH/C Pediatric Grand Rounds. MGH staff	2011
Screening for Primary Immunodeficiencies: When and How? First Steps with SCID Newborn Screening. Primary Care Pediatric Conference, Harvard Medical School	2011
Human RAG deficiency: not just immunodeficiency. Novel insights into mechanisms of immune dysregulation. Presenter at Plenary Session, MGH/C Research Day	2012
Severe Combined Immunodeficiency Allergy/Immunology Grand Rounds, MGH	2013
Autoimmunity in Primary Immunodeficiencies Pediatric Grand Rounds, MGH/C	2013
A 14-year-old boy with immune dysfunction and cognitive, endocrine and liver abnormalities. Case Records of Massachusetts General Hospital: Clinicopathological Conference at Pediatric Grand Rounds	2014
Primary Immunodeficiencies – in the era of whole exome sequencing Analytic and Translational Genetics Unit (ATGU) Coffee hour	2014
Annual Symposium & Rhinolaryngoscopy Hands-on Workshop Diagnosis of Immunodeficiency Diseases. Tampa, FL	2016-present
Moderator and organizer of clinical case and research conference in Immunology for clinical fellows and faculty (Immune Dysregulation Think Tank Meeting) 1-hr presentation per month when we discussed cases and research ideas in Immunology in multidisciplinary settings at JHACH. Outside speakers were invited and their visit was funded with advocacy funds from the Jeffrey Modell Diagnostic and Research Center	2017-present

Lectures by InvitationInternational

- “The growing spectrum of RAG deficiencies” 2013
Invited speaker, II São Paulo Advanced School on Primary Immunodeficiencies:
“The interface of Autoimmunity and Immunodeficiency (II ESPCA-PID), São Paulo,
Faculdade de Medicina da Universidade de São Paulo (FMUSP), Brazil
- “Dysregulated innate immune system feeds hyperinflammation in RAG deficiency” 2013
Invited speaker, Annual Meeting of Hungarian Immunology Society, Pecs, Hungary
- “Combined Immunodeficiencies with immune dysregulation” 2015
Cancer Research Institute rounds Immunology Rounds, The Chaim Sheba Medical
Center, Tel-Aviv University, Sackler School of Medicine, Tel-Aviv, Israel
- “RAG and autoimmunity” 2015
Invited speaker, Second International Workshop on ALPS and associated disorders,
Genova, Italy
- “Review of Novel Primary Immunodeficiencies: DNA PK deficiency” 2016
Euro-CID/P-CID/CELL-PID meeting, Rome, Italy
- “SCID identified by newborn screening: diagnostic approach and targeted therapy”, 2019
Annual meeting of Hungarian Immunology Society (MIT), Hungary
- “Cytopenias of Many Kinds: An Emerging Face of Primary Immunodeficiencies”, 2019
Immunodeficiency Canada 7th SCID Symposium, Montreal, Canada
- “Most frequent molecular defects and gene therapy for SCID”, XLVIII Brazilian 2021
Congress of Allergy and Immunology, virtual
- “Crossroads between SID, PID and Autoimmunity” Symposium on Secondary 2021
Immune Deficiency and Primary Immune Deficiency Crossovers, European Society
for Immunodeficiencies (ESID), virtual

National

- “B cell dysregulation in RAG-dependent PID in human and mice” 2011
Department of Pediatrics, Stanford Medical School
- “Autoimmunity in RAG-dependent Primary immunodeficiency” 2011
Grand Rounds, NIAID, National Institutes of Health, Bethesda
- “Understanding Autoimmunity in Immunodeficiencies” 2012
Discussion Leader, AAAAI Annual Meeting, Miami, FL
- “Challenges before and after newborn screening for SCID” 2012
Grand Rounds. Children’s Hospital of The King’s Daughters, Eastern Virginia
Medical School

“Clinical Challenges in the Diagnosis of Primary Immunodeficiencies” Annual meeting for Association of Medical Laboratory Immunologists (AMLI), Minneapolis, MN	2012
“Clinical Challenges in the Diagnosis and Treatment of Autoimmunity in Primary Immunodeficiencies”, Immunology Seminars, Mayo Clinic, Rochester, MN	2012
“Secondary Evaluation of SCID Screen-Positive Infants” AAAAI Annual Meeting, San Diego, CA	2014
“Comparative analysis of patients followed at the Massachusetts General Hospital (MGH) versus the national CVID registry (USIDNET)” USIDNET Annual Face-to-Face Meeting, Washington, DC	2014
“The broadening spectrum of RAG deficiencies”. Grand Rounds, Division of Pediatric Allergy and Immunology, Northwestern University, Chicago, IL	2014
“Clinical spectrum and challenges in autoimmune complications in PID” (Symposium supported by CSL-Behring), AAAAI Annual Meeting, Houston, TX	2015
“Autoimmunity in RAG deficiency: Lessons from our patients” Grand Rounds, University of South Florida, Tampa, FL	2015
“Autoimmunity in RAG deficiency: Lessons from our patients” Grand Rounds, Mattel Children’s Hospital, UCLA, Los Angeles, CA	2015
“Clues for early detection, proper immune suppression and support of patients with autoimmune disease and underlying immune deficiency” Annual Meeting of American College of Rheumatology, San Francisco, CA	2015
“Mechanisms of allergic inflammation in Omenn Syndrome” Discussion Leader, Annual Meeting of AAAAI, Los Angeles, CA	2016
“The link to immunodeficiency: Targeted therapy” Discussion Leader, Annual Meeting of AAAAI, Los Angeles, CA	2016
Session on “Immune dysregulation” Moderator, Annual Meeting of Clinical Immunology Society, Boston, MA	2016
“Mechanism-based treatment strategies of immune dysregulation in PIDs” Allergy & Clinical Immunology Seminar, Yale University School of Medicine, New Haven, CT	2016
“Congenital Immunodeficiency: Primary Immunodeficiencies hints for diagnosis and treatment” Grand Rounds, Cleveland Clinic, Cleveland, OH	2016

“Advancing Ig Nursing and Pharmacy Practice. The Role of Rituximab and Ig Therapy” IgNS 2016 5th National Conference, Miami, Florida	2016
“T-cell receptor excision circle (TREC) cutoff in newborn screening for SCID: when and how to act” 48th Annual Meeting of Hungarian Medical Association of America. Sarasota, FL	2016
“Severe Combined Immunodeficiency” HST-Session for medical students, Harvard Medical School, Boston, MA	2016
“Year in Review Workshop: Hot Topics in Pediatric Allergy/Immunology” Annual Meeting of AAAAI, Atlanta, GA	2017
“Anti-cytokine autoantibodies in human disease” Discussion Leader, Annual Meeting of AAAAI, Atlanta, GA	2017
“Hot Topics in Immunology” Pediatric Allergy & Immunology Teaching Conference, Duke University, Durham, NC	2017
“Clinical variability and immune dysregulation in Rag deficiency” Baylor College of Medicine /Texas Children’s Hospital, Houston, TX	2017
“Primary Immunodeficiency Diseases in Adults”, Annual meeting of Oklahoma Allergy and Asthma Society, Oklahoma City, Oklahoma	2018
“Patient Perspectives on Primary Immunodeficiency (PI): Shared Decision-Making for Personalized PI Management” Annual Meeting of AAAAI, Symposium sponsored by Shire, Orlando, FL	2018
“Impaired tolerance mechanisms and targeted therapies for autoimmunity in PID” AIR Grand Rounds, University of Rochester School of Medicine & Dentistry Rochester, NY	2018
“Gene Therapy Technology in Clinical Trials for Hereditary Immune Deficiency”, Annual Meeting of AAAAI, San Francisco, CA	2019
“Genetics 101 for the Practicing Immunologist” Discussion Leader, Annual Meeting of AAAAI, San Francisco, CA	2019
“Adult Primary Immunodeficiencies” Discussion Leader, Annual Meeting of AAAAI, San Francisco, CA	2019
Practical application of “Why How Where” Annual Meeting of Clinical Immunology Society, Atlanta, GA	2019
“Primary Immunodeficiency Diseases in Adults” Oklahoma Allergy and Asthma Society, Tulsa, Oklahoma	2019

“Updates in Diagnostics for Neutrophil Disorders” National Neutropenia Network, Virtual Conference, Webinar	2020
“Rituximab and Eculizumab when Treating Non-Malignant Hematologic Disorders (NMHD): Infection Risk, Immunization Recommendations, Antimicrobial Prophylaxis Needs” American Society of Hematology, Annual Meeting, Virtual	2020
“Advances in clinical and mechanic understanding in WHIM syndrome” NICER monthly meeting, virtual	2021
“Inborn errors of immunity and metabolic diseases in Irish Travelers community” SCID Compass Key Opinion Leader Roundtable, virtual	2021
“NBS for SCID: perspectives of a clinician and researcher” Newborn Screening Research: Innovations from Technology, Advocacy and Clinical Care. Newborn Screening Summit by Newborn Screening Translational Research Network, virtual	2021
“Diagnosis and Management of Autoimmune Cytopenias (AIC) in Primary Immunodeficiency (PID)” National Meeting of Immune Deficiency Foundation, virtual	2021
WHIM Syndrome (Warts, Hypogam, Infections, Myelokathexis) Rare of The Rare Summit, Immune Deficiency Foundation, virtual	2021
<u>Regional</u>	
“Everyday management of primary immunodeficiency for parents” Immunodeficiency Foundation Education Conference, Billerica, MA	2009
“Ask the Expert” Session Immunodeficiency Foundation Education Conference, Dedham, MA	2011
Immune deficiency and immune dysregulation 1. “Ask the Expert” Session 2. Navigating between specialists Immunodeficiency Foundation Education Conference, Dedham, MA	2012
“Teen Escape” – conference focusing on needs to adolescent with PID 1. “Ask the Expert” Session 2. “Take control of your health” – round table discussion with teens Immunodeficiency Foundation Education Conference, Dedham, MA	2012
Frontiers in Pediatric Gastroenterology, Hepatology & Nutrition. Immune modulation in Pediatric Neuropsychiatric disorders	2014
An Educational Day to Learn about Primary Immunodeficiency Diseases” 1. Autoimmunity & Primary Immunodeficiency – What’s the Link? – oral presentation 2. “Ask the Medical Expert” Session	2014

Immune Deficiency Foundation Family Conference Day, Dedham, MA	
“Autoimmunity & Immunodeficiency: What’s the link?” “Ask the Medical Experts” Session Immune Deficiency Foundation Retreat, Newton, MA	2015
“Autoimmunity & Immunodeficiency: What’s the link?” Immune Deficiency Foundation Education Meeting. Sarasota, FL	2016
“Diagnosis of Immunodeficiency Diseases” Florida Allergy, Asthma & Immunology Society Annual Meeting. Orlando, FL	2016
“Targeted treatment strategies for autoimmunity in PID” Conference and meeting with Rheumatology and Immunology, Gainesville, FL	2017
“Where we are & Where we are going”, Immune Deficiency Foundation. Immune Deficiency Foundation Education Meeting, St Augustine, FL	2018
“Genetic Landscape and diagnostic approach for SCID identified by Newborn screening” Genetics and Newborn Screening Advisory Council meeting, Tallahassee, FL	2018
“Two atypical SCID cases from Florida” Florida Pediatric BMT and Cell Therapy Consortium, Webinar	2019
“Cytopenias of Many Kinds: An Emerging Face of Primary Immunodeficiencies” Florida Pediatric BMT and Cell Therapy Consortium, Webinar	2020
“Autoimmunity in PID”. National Jewish Hospital, Pediatric Allergy and Immunology Morning Conference, Denver, CO	2020
<u>Local</u>	
“Immune dysregulation in RAG deficiencies” MGH Immunology Seminar Series Charlestown Naval Yard Research Building, MA	2013
“Challenges before and after newborn screening for Severe Combined Immunodeficiency” Grand Rounds, Department of Pediatrics, Cambridge Health Alliance	2013
“Autoimmunity in Primary Immunodeficiencies” Harvard Introductory Course for Allergy and Immunology Fellows, Brigham and Women Hospital, Boston, MA	2014
“Inflammation and immune modulation in Pediatric Autoimmune Neuropsychiatric Disorder Associated with Streptococcus (PANDAS)”, Grand Rounds, Emerson Hospital, Concord, MA	2014-2018

“Autoimmunity in CVID and other Primary Immunodeficiencies” Harvard Introductory Course for Allergy and Immunology Fellows, Brigham and Women Hospital, Boston, MA (annual talk)	2014
“Autoimmunity in Primary Immunodeficiencies: Lessons from RAG deficiency in human and mice” Rheumatology Grand Rounds, Boston Children’s Hospital, Boston, MA	2015
“Autoimmunity in Primary Immunodeficiencies: Lessons from RAG deficiency in human and mice” Rheumatology Grand Rounds, Brigham and Women Hospital, Boston, MA	2015
“Clues for early detection, proper immune suppression and support of patients with autoimmune disease and underlying immune deficiency” Rheumatology Grand Rounds, Brigham and Women Hospital, Boston, MA	2015
“Mechanism of inflammatory enteropathy through the lens of RAG deficiency” Annual Meeting of Center for Study of Inflammatory Bowel Disease Massachusetts General Hospital, Boston, MA	2015
“Challenges of a new era: combined immune deficiencies with immune dysregulation” Allergy Grand Rounds, Boston Children’s Hospital, Boston, MA	2016
“Precision Medicine for Autoimmunity in Primary Immunodeficiencies” Pediatric Grand Rounds, Tampa General Hospital, Tampa, FL	2016
“Impaired tolerance checkpoints drive autoimmunity in immune deficiency” Johns Hopkins All Children’s Hospital 5th Annual Research Symposium, St. Petersburg, FL	2017
“Personalized Medicine for Late-Onset Primary Immunodeficiencies: From Biologicals to HSCT”, Grand Rounds, Moffitt Cancer Center, Tampa, FL	2017
“Precision Medicine for Autoimmunity in Primary Immunodeficiencies” Grand Rounds, St. Joseph Children’s Hospital, Tampa, FL	2018
“Newborn Screening, Genetic Testing & New Avenues in Therapy” Immune Deficiency Foundation, Education Meeting, Tampa, FL	2018
“Role of microbiome in immune dysregulation and neurological manifestations” Gut-Brain Research Program, Symposium & Planning Retreat, Johns Hopkins All Children’s Hospital, St. Petersburg, FL	2019
“SCID/CID Screening 101: diagnostic approach and targeted therapy” Johns Hopkins All Children’s Hospital, St. Petersburg, FL	2019
Immune Mediated Cytopenias -A Sign of PIDD or Not? NICER Immuno-Hematology Symposium Agenda, OH	2019

“Autoimmunity in PID” 2020
Johns Hopkins All Children’s Hospital, St. Petersburg, FL

Clinical Activities or Innovations

Practice Activities

Pediatric inpatient care, Franciscan Hospital for Children, Brighton, MA, 3 night and 1 weekend day per month	2009-2016
Sub-specialty ambulatory care, Allergy/Immunology, MGHfC, Boston, MA 4-5 half-day clinic sessions per week	2009-2012
Sub-specialty ambulatory care, Allergy/Immunology, MGHfC, Boston, MA 2 half-day clinic sessions per week	2012-2016
Sub-specialty inpatient care, Allergy/Immunology, MGHfC, Boston, MA 12 weeks of inpatient service per year	2009-2016
Sub-specialty ambulatory care, Allergy/Immunology, USF, St. Petersburg, FL 1.5 half-day clinic sessions per week	2016-present
Sub-specialty inpatient care, Allergy/Immunology, JHACH, St. Petersburg, FL 16 weeks of inpatient service per year	2016-present
Sub-specialty ambulatory care, Immunology, Moffitt Cancer Center, Tampa, FL 1 half-day clinic sessions per month	2019-present

Clinical Innovations

Dr. Walter’s work contributed to the increased awareness of autoimmune disease among pediatric and adult patients with primary immunodeficiency as a sign of immune dysregulation. She closely works with other disciplines to promote early diagnosis. In her clinical care, Dr. Walter promotes mechanism-based immune modulation as bridge therapy for this patient population. If bridge therapy fails, her patients are diligently prepared for discussion for hematopoietic stem cell transplant in conjunction with the BMT team. Lastly, she had success referring her patients to gene therapy trials if conventional HSCT options are exhausted. Her observations have been reported at the Clinical Immunology Society Meetings in multiple abstracts and in publications on combined immunodeficiencies. Her work will continue in clinical care and translational research in CID in children and adults.

Research Innovation

Dr. Walter is a translational physician-scientist with specific training and expertise in basic science (cellular immunology, murine studies), clinical immunology (primary immune deficiency [PIDs] and regulatory disorders) and virology (epidemiology of viruses in the gut).

Dr. Walter's research concerns fundamental and therapeutic studies of immune dysregulation in combined immunodeficiencies (CIDs), with emphasis on B cell dysfunction. As a recipient of an NIAID-K08 grant (2012), I developed and carried out studies in B cell tolerance in PIDs and assembled a research team at Harvard (2013-2016). Currently I am Division Chief of Pediatric Allergy/Immunology at University of South Florida and at Johns Hopkins All Children's Hospital with my time divided between research (75%), teaching and clinical care (15%), and administration (10%).

At present, targeted therapies for elimination of autoreactive B cells are limited and may result in hypogammaglobulinemia, especially in inborn errors of immunity (IEI) (*Barmettler JAMA-NO 2018*). Mechanistic understanding of impaired tolerance checkpoints and drivers of autoreactive B cells could guide innovation in treatment approaches. Dr. Walter's group examined impaired tolerance checkpoints at specific B cell developmental stages in several IEI including partial RAG deficiency (pRD) (*Walter, JCI 2015, Walter, JEM 2010*) and in collaboration in human PI3K deficiency (*Farmer, Science Signaling 2019*), human activation-induced cytidine deaminase (AID) deficiency (*Cantaert, Immunity 2015*) and B cell intrinsic mouse models of Wiskott-Aldrich syndrome. Treatment outcomes for immune dysregulation are often guarded and mechanism-based therapy is available for selected CIDs but not for pRD (*Farmer, JACI-IP 2018*).

Dr. Walter investigate pathomechanisms of immune dysregulation in inborn errors of immunity. In the past 10 years, novel polyreactive-prone B cell subsets were identified both on clinical grounds and in animal models that accumulate in the extrafollicular (EF) space and contribute to the pre-plasma cell pool for autoantibody secreting cells. Specifically, my research team focuses on extrinsic and intrinsic factors related to age-associated B cell (ABC) development, that may affect the marked phenotypic diversity and tendency for autoimmunity in a genetically-coded IEIs (G-CIDs). These factors include microbial antigen load and B cell activating factor (BAFF). The results of our investigations will likely have important clinical implications for future research on disease mechanisms and preventive/therapeutic strategies.

Dr. Walter's team has a strong collaborative network for gene expression studies in mouse models and leading B cell experts including Michael Cancro (UPenn) (one of two lead investigators to first describe ABCs [2011]), Klaus Warnatz (Freiburg, Germany) (B cell expert in ABC-like cells in CVID), and Chis Tipton from Sanz group (Emory University) (B cell repertoire studies in human autoimmune diseases). Overall, my long-term goal is to translate the results of basic science studies of B cell dysfunction to impact clinical practice, part of which focuses on a highly informative albeit challenging group of patients with CID and immune dysregulation.

Scholarly Activity (grant history)

A) Current Grants

Agency: NIH/NIAID

ID#: 1R01AI153830-01A1

Title: "Mechanisms driving extrafollicular polyreactive B cell lineages in partial RAG deficiency"

P.I.: Jolan Walter, MD PhD

Role on Project: Principal Investigator

Percent effort: 30%

Direct costs per year: \$440,669/year (5 years)

Total costs for project period: \$2,269,039.27

Project period: 09/22/2021 – 08/31/2026

Agency: Johns Hopkins All Children's Hospital
ID#: All Children's Research Institute
Title: "Mechanistic studies of accelerated aging of B cells in combines immunodeficient patients and animal models"
P.I.: Jolan Walter, MD PhD
Role on Project: Principal Investigator
Percent effort: 5%
Direct costs per year: ~\$80,000/year (3 years)
Total costs for project period: \$250,000.00
Project period: 11/01/2021 – 09/30/2024

Agency: SCID Angels for Life Foundation
ID#: n.a.
Title: "Severe Combined Immunodeficiency(SCID) Screening and Education"
P.I.: Jolan Walter, MD PhD
Role on Project: Principal Investigator
Percent effort: 2%
Direct costs per year: \$50,000/year (1 year)
Total costs for project period: \$ 50,000
Project period: 08/01/2021 – 07/31/2022

Agency: Florida Department of Health and Orca Biosystems, Inc.
ID#: n.a.
Title: "Phase I/II Trial: Engineered Donor Graft (Orca Q) for Pediatric Hematopoietic Cell Transplant HCT"
P.I.: Biljana Horn, MD
Role on Project: Site-PI
Percent effort: 1%
Direct costs per year: \$16,000/year (3 year)
Total costs for project period: \$48,000
Project period: 04/01/2021 – 03/31/2026

Agency: Johns Hopkins All Children's Hospital
ID#: Department of Medicine Research Council Grant
Title: "Evaluating T cell subsets during oral immunotherapy for peanut allergy"
P.I.: Panida Sriaroon, MD
Role on Project: Co-Principal Investigator
Percent effort: 2%
Direct costs per year: \$30,000/year (1 year)
Total costs for project period: \$30,000.00
Project period: 09/01/2021 – 08/31/2022

Agency: Immune Deficiency Foundation
ID#: 4 SC1MC31881-02-02
Title: "Severe Combined Immunodeficiency(SCID) Screening and Education"
P.I.: Alissa Creamer

Role on Project: Co-Investigator (Subcontract)

Percent effort: 2%

Direct costs per year: \$50,000/year (1 year)

Total costs for project period: \$ 74,750

Project period: 08/01/2021 – 07/31/2022

Agency: Johns Hopkins All Children's Hospital

ID#: Foundation Institutional COVID-19 Related Research Grant

Title: "Studies on natural history of and immune response in COVID-19 in the immunocompromised population"

P.I.: AnnMarie Szymanski, MD PhD

Role on Project: Co-Principal Investigator

Percent effort: 3%

Direct costs per year: \$100,000/year (1 year)

Total costs for project period: \$100,000.00

Project period: 08/01/2021 – 07/31/2022

Agency: University of South Florida

ID#: Project Enhancement Grant

Title: "Dissecting the role of T and B cells in immune dysregulation partial RAG deficiency and autoimmunity"

P.I.: Jolan Walter, MD PhD

Percent effort: 5%

Direct costs per year: \$25,000/year (1 year and NCE)

Total costs for project period: \$25,000

Project period: 05/01/2019 – 04/30/2022

Agency: Medical Center-University of Freiburg, Germany

I.D.#: Multicenter Grant

Title: "A prospective outcome study on patients with profound combined immunodeficiency"

P.I.: Jolan Walter, MD PhD (US Site)

Role: US-Site Coinvestigator

Percent effort: 1%

Total costs for project period: paid per referral

Project period: 01/01/2017 – 12/31/2022

Sponsored Research Study:

Agency: Shire (now Takeda)

ID#: Sponsored study

Title: "Efficacy, Safety, Tolerability, Immunogenicity and Pharmacokinetic Evaluation of HYQVIA in Pediatric PIDD Subjects"

Site-P.I.: Jolan Walter, MD PhD

Percent effort: 5%

Direct costs per year: \$31,000/year (4 years)

Total costs for project period: \$127,000
Project period: 11/17/2017 – 11/16/2022

Agency: X4 Pharmaceuticals, Inc.
ID#: Investigator Initiated Study
Title: “Immune Cell Profiling Protocol for WHIM Syndrome”
P.I.: Jolan Walter, MD PhD
Percent effort: 1%
Direct costs per year: \$45,000/year (1 year and NCE)
Total costs for project period: \$45,000
Project period: 03/01/2019 – 02/29/2024

Agency: Octapharma
ID#: Sponsored study
Title: “Study to Monitor Subcutaneous Human Immunoglobulin Administered at Modified Dosing Regimens in Patients with Primary Immunodeficiency Diseases”
Site-P.I.: Jolan Walter, MD PhD
Percent effort: 2%
Direct costs per year: \$30,000/year (4 years)
Total costs for project period: \$120,000
Project period: 01/03/2020 – 01/02/2025

Agency: Leadiant
ID#: Sponsored study
Title: “Registry Study of Revkovi Treatment in Patients with ADA-SCID”
Site-P.I.: Jolan Walter, MD PhD
Percent effort: 2%
Direct costs per year: \$20,000/year (4 years)
Total costs for project period: \$80,000
Project period: 08/07/2019 – 08/06/2024

1-Name: Norm-01(Newnorm)(PID)- 20% SC product
Sponsor: Octapharma PI: Dr. Jolan Walter Sub investigator: Daime Nieves

2-Name: NGAM-12(CLL) secondary immunodeficiency- Panzyga 10% IV- USF IC
Sponsor: Octapharma PI: Dr. Jolan Walter Sub investigator: Daime Nieves

4-Name: X4P 001-104(Severe Congenital Neutropenia >12y ANC less than 1000 – genetics done, diagnosis 6 months prior to screening Sponsor: X4Pharmaceuticals
PI: Dr. Jolan Walter. Sub investigator: Daime Nieves Collaboration: Dr. Sotomayor TGH

- Name: ABCVILD
Sponsor: Bristol Mayer/(BM) FDA/ Cincinnati Children- Michael Jordan MD

PI: Cincinnati- Michael Jordan MD Sub investigator: Dr. Jolan Walter/ Daime Nieves

Sponsor: Janssen PI: Dr. Jolan Walter Sub investigator: Daime Nieves
Status: Enrolling- Amendment 4 - renegotiating budget –

Name: AZD7442- Evusheld- monoclonal for kids
Sponsor: Astrazeneca PI: Dr. Jolan Walter Sub investigator: Daime Nieves

Name: Revcov- Registry- LBI-2279-004
Sponsor: Lediand/ Chiesi. PI: Dr. Jolan Walter Sub investigator: Daime Nieves

9- Name: ADMA- 004- Hyperimmune IgG Asceniv- patient in IV for 12 weeks (2-6) (7-11)
Sponsor: ADMA PI: Dr. Jolan Walter Sub investigator: Daime Nieves

10- Name: Regeneron R686-EE 1877- Dupixent and EOE
Sponsor: Regeneron PI: Dr. Panida Sriaroon Sub investigator: Daime Nieves

B) Past Grants

Agency: NIH/NIAID
ID#: 5R01AI10088707
Title: “Immune repertoire and function in typical and atypical SCID”
P.I.: John Manis, MD
Role on Project: Collaborator (Subcontract)
Percent effort: 5%
Direct costs per year: \$30,000/year (5 years)
Total costs for project period: \$7,097,957
Project period: 07/01/2016 – 06/30/2021

Agency: Jeffrey Modell Foundation
ID#: Research Grant
Title: “Genetic Screening Protocol for WHIM syndrome: Diagnostic Protocol for Non-Immunologists”
P.I.: Jolan Walter, MD PhD
Percent effort: 1%
Direct costs per year: \$50,000/year (2 years and NCE)
Total costs for project period: \$100,000
Project period: 05/01/2018 – 04/30/2021

Agency: Jeffrey Modell Foundation
ID#: C.H.I.L.D.R.E.N! Research Program Grant
Title: “Genetic Screening for Primary Immune Deficiency Among Patients with Autoimmune Cytopenias”
P.I.: Jolan Walter, MD, PhD

Percent effort: 5%
Direct costs per year: \$25,000/year (2 years and NCE)
Total costs for project period: \$50,000
Project period: 04/01/2018 – 03/31/2021

Agency: University of South Florida
ID#: New Researcher Grant
Title: “Gut microbiota translocation drives autoimmunity in RAG deficiency”
P.I.: Krisztian Csomos PhD (Co-PI: Jolan Walter MD PhD)
Direct costs per year: \$25,000/year (1 year and NCE)
Total costs for project period: \$25,000
Project period: 05/01/2019 – 04/30/2021

Agency: Johns Hopkins All Children’s Hospital
I.D.# Institutional Grant (“Rothman”)
Title: “The microbiome-gut-brain axis in pediatric neuropsychiatric syndromes with OCD and/or tics as affected by antibiotic treatment and native IgA levels—a pilot study”
P.I.: Jolan Walter, MD PhD
Total costs for project period: \$100,000
Percent effort: 1%
Project period: 06/01/2019 – 05/31/2021

Agency: Johns Hopkins All Children’s Hospital
I.D.# Institutional Grant
Title: “Feasibility study to assess the role of T and B cells in refractory cytopenias in children”
P.I.: Jolan Walter, MD PhD
Percent effort: 1%
Total costs for project period: \$240,000
Project period: 01/01/2017 – 06/01/2020

Agency: NIH/NIAID
I.D.# K08 Career Award
Title: “Autoimmunity in Rag deficiency: in the nexus of immunodeficiency and immune dysregulation”
P.I.: Jolan Walter, MD PhD
Percent effort: 75%
Direct costs per year: \$125,000
Total costs for project period: \$625,000
Project period: 08/01/2012 – 07/31/2017

Agency: John Hopkins All Children’s Hospital
I.D.# Institutional Grant
Title: “The Gut Microbiota of Primary Immunodeficiency Diseases”
P.I.: Larry Dishaw, PhD

Role on Project: Co-Investigator
Percent effort: 1%
Total costs for project period: \$80,000
Project period: 01/01/2015 – 12/31/2019

Agency: USIDNet
I.D.# Baxter Grant
Title: “Define CVID epidemiology, with a specific focus
on non-infectious disease complications”
P.I./Mentor: Jolan Walter, MD PhD
Mentee: Jocelyn Farmer, MD PhD
Percent effort: 10%
Total costs for project period: \$8,000
Project period: 07/01/2015 – 06/30/2016

Agency: CSL-Behring
I.D.# Publication Research Grant
Title: “Defining the epidemiology and individualized Ig Dosing for
CVID in a large tertiary care centers”
P.I./Mentor: Jolan Walter, MD PhD
Mentee: Jocelyn Farmer, MD PhD
Percent effort: 10%
Total costs for project period: \$25,000
Project period: 01/01/2015 – 12/31/2016

Agency: CSL-Behring
I.D.# Publication Research Grant
Title: “Cost-benefit analysis of immunoglobulin replacement for
mitochondrial patients with immune dysfunction”
P.I./Mentor: Jolan Walter, MD PhD
Mentee: Melissa Walker, MD PhD
Percent effort: 10%
Total costs for project period: \$20,000
Project period: 07/01/2015 – 06/30/2016

Agency: NIH-NIDDK
Center for Study of Inflammatory Bowel Disease, MGH
I.D.# Pilot Feasibility Grant (in 5P30DK043351-25)
Title: “Mechanism of inflammatory enteropathy through the lens of
RAG deficiency”
P.I.: Jolan Walter, M.D PhD
Percent effort: 10%
Total costs for project period: \$10,000/year
Project period: 01/01/15 – 12/30/15

Agency: NIH-NIAID
I.D.# Junior Faculty Pilot Grant (IOF in U19 AI095261-01)
Title: “ILC2-driven allergic inflammatory responses in a murine

model of Omenn syndrome”
P.I.: Jolan Walter, MD PhD
Percent effort: 10%
Total costs for project period: \$50,000
Project period: 07/01/2013 – 06/30/2014

Agency: Nathan Talbot
I.D.# Pilot Feasibility Grant
Title: “Dissecting mechanisms of central and peripheral
dysregulation among patients with RAG deficiencies”
P.I.: Jolan Walter, MD PhD
Percent effort: 10%
Total costs for project period: \$50,000
Project period: 07/01/2012 – 06/30/2013

Agency: Clinical Immunology Society/Baxter
I.D.# Junior Faculty Award
Title: “Unraveling the spectrum of B cell mediated autoimmunity in
Rag deficiencies”
P.I.: Jolan Walter, MD PhD
Percent effort: 50%
Total costs for project period: \$50,000
Project period: 07/01/2011 – 06/30/2012

Agency: GSK-AAAAI
I.D.# Fellow Career Development Award
Title: “Role of B cells in the pathophysiology of Omenn syndrome”
P.I.: Jolan Walter, MD PhD
Percent effort: 50%
Total costs for project period: \$50,000
Project period: 01/01/2008 – 12/31/2010

Agency: NIH-NIAID
I.D.# NIH 5T32 AI007512
Title: “Molecular Basis of Allergic and Immunologic Diseases”
P.I.: Raif Geha, MD
Percent effort: 100%
Total costs for project period: n.a.
Project period: 01/01/2007 – 12/31/2009

Past Sponsored Research Studies:

Agency: X4 Pharmaceuticals, Inc
ID#: Sponsored study
Title: “A Trial of X4P-001 in Patients with WHIM Syndrome (X4P-
001-MKKA)”
Site-P.I.: Jolan Walter, MD PhD

Percent effort: 1%
Total costs for project period: \$20,000
Project period: 05/01/2016 – 04/30/2020

Agency: Leadiant Biosciences, Inc.
ID#: Sponsored study
Title: “EZN-2279 in Patients with ADA-SCID”
Site-P.I.: Jolan Walter, MD PhD
Percent effort: 5%
Total costs for project period: \$10,000
Project period: 05/01/2016 – 04/30/2017

Published Bibliography

1. Szánya, J.E., Szakály, P., Magyarlaki, T., Balogh, Z., Nagy, J., Kalmár Nagy, K. (1997). Predictive morphological findings in 0 hour biopsies of renal allografts. *Acta Chirurgica Hungarica*, 36(1-4): 346-348. PMID: 11218129
2. Walter, J.E., Mitchell, D.K., Guerrero, M.L., Berke, T., Matson, D.O., Monroe, S.S., Pickering, L.K., Ruiz-Palacios, G. (2001). Molecular Epidemiology of Human Astrovirus among Children from a Periurban Community of Mexico City. *Journal of Infectious Diseases*, 183(5) 681-686. PMID: 11181143
3. Taylor, M.B., Walter, J.E., Berke, T., Cubitt, D.W., Mitchell, D.K., Matson, D.O. (2001). Characterisation of a South African human astrovirus as type 8 by antigenic and genetic analyses. *Journal of Medical Virology*, 64(3) 256-261. PMID: 11424112
4. Walter, J.E., Briggs, J., Guerrero, M.L., Matson, D.O., Pickering, L.K., Ruiz-Palacios, G., Berke, T., Mitchell, D.K. (2001). Molecular Characterization of a Novel Recombinant Strain of Human Astrovirus Associated with Gastroenteritis in Children. *Arch Virol* 146(12):2357-67. PMID: 11811685
5. Jakab, F., Walter, J.E., Szűcs, G.Y. (2001). Human astroviruses in children with gastroenteritis. *Infektologia es Klinikai Mikrobiologia* 7(3) 118-122.
6. Walter, J.E., Matson, D.O., Berke, T., Mitchell, D.K. (2001). Reclassification of the family Astroviridae based on Comparative Phylogenetic Analysis of Capsid Genes. *The Infectious Disease Review* 3(3) 155-6.
7. Nadan, S., Walter, J.E., Grabow, W.O.K., Mitchell, D.K., Taylor, M.B. (2003). Molecular characterization of astroviruses: comparison between clinical and environmental isolates from South Africa. *Applied and Environmental Microbiology*, 69(2) 747-753. PMID: 12570991, PMCID: PMC143596
8. Jakab, F., Walter, J.E., Berke, T., Matson, D.O., Mitchell, D.K., Szucs, G. (2003). Molecular characterization and sequence analysis of human astroviruses circulating in Hungary. *FEMS Immunology and Medical Microbiology*, 39(2) 97-102. PMID: 14625092
9. Al-Mutairy, B., Walter, J.E., Pothén, A., Mitchell, D.K. (2005). Genome Prediction of Putative Genome-Linked Viral Protein (VPg) of Astroviruses. *Virus Genes*, 31(1) 21-30. PMID: 15965605

10. Walter, J.E., Miller, J., Fisher, R.G., Byrd, R. (2006). Subcutaneous nodule in a young girl: a diagnostic dilemma. *Clinical Pediatrics*, 45: 661-4. PMID: 16928845
11. Zhang, Z., Mitchell, D.K., Afflerbach, C., Jakab, F., Walter, J.E., Zhang, Y.J., Staat, M.A., Azimi, P., Matson, D.O. (2006). Quantitation of human astrovirus by real-time reverse-transcription-polymerase chain reaction to examine correlation with clinical illness. *J Virol Methods*, 134(1-2):190-6. PMID: 16490263
12. Permaul, P., Stutius, L.M., Sheehan, W.J., Rangsithienchai, P., Walter, J.E., Twarog, F.J., Young, M.C., Scott, J.E., Schneider, L.C., Phipatanakul, W. (2009). Sesame allergy: role of specific IgE and skin-prick testing in predicting food challenge results. *Allergy Asthma Proc*, 30(6):643-8. PMID: 20031010, PMCID: PMC3131114
13. Rucci, F., Notarangelo, L.D., Fazeli, A., Partizi, L., Hickernell, T., Paganini, T., Coakley, K., Detre, C., Keszei, M., Walter, J.E., Feldman, L., Cheng, H.L., Poliani, L., Wang, J.H., Balter, B., Recher, M., Andersson, E., Zha, S., Giliani, S., Terhorst, C., Alt, F.W., Yan, C. (2010). Homozygous DNA Ligase IV R278H mutation in mice leads to leaky SCID and represents model for human LIG4 syndrome. *Proc Natl Acad Sci USA*, 107(7):3024-9. PMID: 20133615, PMCID: PMC2840307
14. Walter, J.E., Rucci, F., Detre, C., Patrizi, L., Paganini, T., Recher, M., Keszei, M., Pessach, I., Lang, P.A., Regenass, S., Giliani, S., Andersson, E., Sekiguchi, J., Al-Herz, W., Cowan, M.J., Dbaibo, G., ElGhazali, G., Gennery, A.R., Pasic, S., Puck, J.M., Malech, H., DeRavin, S.S., Niehues, T., Schuetz, C., Blessing, J., Terhorst, C., Alt, F.W., Notarangelo, L.D. (2010). Defects of B cell tolerance and expansion of immunoglobulin-secreting cells in Rag-dependent immunodeficiency. *Journal of Experimental Medicine*, 207(7):1541-54. PMID: 20547827, PMCID: PMC2901061
15. de la Fuente, M.A., Recher, M., Rider, N.L., Strauss, K.A., Morton, D.H., Adair, M., Bonilla, F.A., Ochs, H.D., Gelfand, E.W., Pessach, I.M., Walter, J.E., King, A., Giliani, S., Pai, S.Y., Notarangelo, L.D. (2011). Reduced thymic output, cell cycle abnormalities, and increased apoptosis of T lymphocytes in patients with cartilage-hair hypoplasia. *J Allergy Clin Immunol*, 128(1):139-46. PMID: 21570718, PMCID: PMC4287238
16. Meyers, G., Ng, Y.S., Bannock, J.M., Lavoie, A., Walter, J.E., Notarangelo, L.D., Kilic, S.S., Aksu, G., Debré, M., Rieux-Laucat, F., Conley, M.E., Cunningham-Rundles, C., Durandy, A., Meffre, E. (2011). Activation-induced cytidine deaminase (AID) is required for B-cell tolerance in humans. *Proc Natl Acad Sci USA*, 108(28):11554-9. PMID: 21700883, PMCID: PMC3136251
17. Recher, M., Berglund, L.J., Avery, D.T., Cowan, M.J., Gennery, A.R., Smart, J., Peake, J., Wong, M., Pai, S.Y., Baxi, S., Walter, J.E., Palendira, U., Tangye, G.A., Rice, M., Brothers, S., Al-Herz, W., Oettgen, H., Eibel, H., Puck, J.M., Cattaneo, F., Ziegler, J.B., Giliani, S., Tangye, S.G., Notarangelo, L.D. (2011). IL-21 is the primary common gamma chain-binding cytokine required for human B-cell differentiation in vivo. *Blood*, 118(26):6824-35. PMID: 22039266, PMCID: PMC3338166
18. Recher, M., Burns, S.O., de la Fuente, M.A., Volpi, S., Dahlberg, C., Walter, J.E., Moffitt, K., Mathew, D., Honke, N., Lang, P.A., Patrizi, L., Falet, H.,

- Keszei, M., Mizui, M., Csizmadia, E., Candotti, F., Nadeau, K., Bouma, G., Delmonte, O.M., Frugoni, F., Fomin, A.B., Buchbinder, D., Lundequist, E.M., Massaad, M.J., Tsokos, G.C., Hartwig, J., Manis, J., Terhorst, C., Geha, R.S., Snapper, S., Lang, K.S., Malley, R., Westerberg, L., Thrasher, A.J., Notarangelo, L.D. (2012). B cell-intrinsic deficiency of the Wiskott-Aldrich syndrome protein causes severe abnormalities of the peripheral B-cell compartment in mice. *Blood*, 119(12):2819-28. PMID: 22302739, PMCID: PMC3327460
19. Abraham, R.S., Recher, M., Giliani, S., Walter, J.E., Lee, Y.N., Frugoni, F., Maddox, D.E., Kirmani, S., Notarangelo, L.D. (2012). Adult-Onset Manifestation of Idiopathic T Cell Lymphopenia Due To A Heterozygous RAG1 Mutation. *JACI*, S0091-6749(12)01536-9. PMID: 2312263,1 PMCID: PMC3725988
 20. Bankova, L.G., Walter, J.E., Iyengar, S.R., Lorenzo, M.E., Hornick, J.L., Castells, M.C. (2013). Generalized bullous eruption after routine vaccination in a child with diffuse cutaneous mastocytosis. *JACI in Practice*, 1(1):94-6. PMID: 24229828
 21. Recher, M., Fried, A.J., Massaad, M.J., Kim, H.Y., Rizzini, M., Frugoni, F., Walter, J.E., Mathew, D., Eibel, H., Hess, C., Giliani, S., Umetsu, D.T., Notarangelo, L.D., Geha, R.S. (2013). Intronic SH2D1A mutation with impaired SAP expression and agammaglobulinemia. *Clin Immunol*, 146(2):84-9. PMID: 2328049,1 PMCID: PMC3742382
 22. Henderson, L.A., Frugoni, F., Hopkins, G., DeBoer, H., Pai, S.Y., Lee, Y.N., Walter, J.E., Hazen, M.M., Notarangelo, L.D. Expanding the Spectrum of RAG-1 Deficiency: A Family with Early Onset Autoimmunity. (2014). *JACI* 132(4):969-71. PMID: 23891352, PMCID: PMC3874115
 23. Felgentreff, K., Siepe, M., Kotthoff, S., von Kodolitsch, Y., Schachtrup, K., Notarangelo, L.D., Walter, J.E., Ehl, S. (2014). Severe eczema and Hyper-IgE in Loeys-Dietz-syndrome - Contribution to new findings of immune dysregulation in connective tissue disorders. *Clin Immunol*, 150(1):43-50. PMID: 24333532.
 24. Chen, K., Wu, W., Mathew, D., Zhang, Y., Browne, S.K., McManus, M.P., Pulsipher, M.A., Yandell, M., Bohnsack, J.F., Jorde, L.B., Notarangelo, L.D., Walter, J.E. (2014). Autoimmunity due to RAG deficiency and estimated disease incidence in RAG1/2 mutations. *J Allergy Clin Immunol*, 133(3):880-2.e10. PMID: 24472623, PMCID: PMC4107635
 25. Lee, Y.N., Frugoni, F., Dobbs, K., Walter, J.E., Giliani, S., Gennery, A., Al-Herz, W., Haddad, E., Blessing, J.H., Henderson, L., Pai, S.Y., Nelson, R.P., El-Ghoneimy, D.H., El-Feky, R.A., Soler-Palacin, P., Fuleihan, R.L., Patel, N.C., Su, H.C., Puck, J.M., Palma, P., Cancrini, K., Chen, K., Vihinen, M., Alt, F.W., Notarangelo, L.D. (2014). A systematic analysis of recombination activity and genotype-phenotype correlation in human RAG1 deficiency. *J Allergy Clin Immunol*, 133(4):1099-108. PMID: 24290284, PMCID: PMC4005599
 26. Recher, M., Karjalainen-Lindsberg, M.L., Lindlöf, M., Söderlund-Venermo, M., Lanzi, G., Väisänen, E., Kumar, A., Sadeghi, M., Berger, C.T., Alitalo, T., Anttila, P., Kolehmainen, M., Franssila, R., Chen, T., Siitonen, S., Delmonte, O.M., Walter, J.E., Pessach, I., Hess, C., Simpson, M.A., Navarini, A.A., Giliani, S., Hedman, K., Seppänen, M., Notarangelo, L.D.

- (2014). Genetic variation in schlafen genes in a patient with a recapitulation of the murine 'Elektra' phenotype. *J Allergy Clin Immunol*, 133(5):1462-5, 1465.e1-5. PMID: 24373355
27. Walker, M.A., Slate, N., Alejos, A., Volpi, S., Iyengar, R.S., Sweetser, D., Sims, K.B., Walter, J.E. (2014). Predisposition to infection and SIRS in mitochondrial disorders: 8 years' experience in an academic center. *JACI in Practice*, 2(4):465-468, 468.e1. PMID: 25017538
28. Kwan, A., Abraham, R.S., Currier, R., Brower, A., Andruszewski, K., Abbott, J.K., Baker, M., Ballow, M., Bartoshesky, L.E., Bonilla, F.A., Brokopp, C., Brooks, E., Caggana, M., Celestin, J., Church, J.A., Comeau, A.M., Connelly, J.A., Cowan, M.J., Cunningham-Rundles, C., Dasu, T., Dave, N., De La Morena, M.T., Duffner, U., Fong, C.T., Forbes, L., Freedenberg, D., Gelfand, E.W., Hale, J.E., Hanson, I.C., Hay, B.N., Hu, D., Infante, A., Johnson, D., Kapoor, N., Kay, D.M., Kohn, D.B., Lee, R., Lehman, H., Lin, Z., Lorey, F., Abdel-Mageed, A., Manning, A., McGhee, S., Moore, T.B., Naides, S.J., Notarangelo, L.D., Orange, J.S., Pai, S.Y., Porteus, M., Rodriguez, R., Romberg, N., Routes, J., Ruehle, M., Rubenstein, A., Saavedra-Matiz, C.A., Scott, G., Scott, P.M., Secord, E., Seroogy, C., Shearer, W.T., Siegel, S., Silvers, S.K., Stiehm, E.R., Sugerman, R.W., Sullivan, J.L., Tanksley, S., Tierce, M.L 4th., Verbsky, J., Vogel, B., Walker, R., Walkovich, K., Walter, J.E., Wasserman, R.L., Watson, M.S., Weinberg, G.A., Weiner, L.B., Wood, H., Yates, A.B., Puck, J.M. (2014). Newborn screening for severe combined immunodeficiency in 11 screening programs in the United States. *JAMA*, 312(7):729-38. PMID: 25138334,, PMCID: PMC4492158
29. Farough, S., Karaa, A., Walker, M.A., Slate, N., Dasu, T., Verbsky, J., Fusunyan, R., Canapari, C., Van Cleave, J., Sweetser, D., Sims ,K.B., Walter, J.E. (2014). Coenzyme Q10 and Immunity: A case report and new implications for treatment of recurrent infections in metabolic diseases. *Clinical Immunology*,155(2):209-1. PMID: 25264263
30. Farmer, J., Sokol, C., Bonilla, F.A., Murali, M., Kradin, R., Astor, T., Walter, J.E. (2014). Bilateral lung transplantation in a patient with humoral immune deficiency: a case report with review of the literature. *Case Reports Immunol*, 2014:910215. PMID: 25379312
31. Walter, J.E., Lo, M.S., Kis-Toth, K., Tirosh, I., Frugoni, F., Lee, Y.N., Csomos, K., Chen, K., Pillai, S., Dunham, J., Tsokos, G., Luning, P., Notarangelo, L.D. (2015). Impaired receptor editing and heterozygous RAG2 mutation in a patient with systemic lupus erythematosus and erosive arthritis. *J Allergy Clin Immunol*, 135(1):272-3, PMID: 25312763, PMCID: PMC4289116
32. Buchbinder, D., Baker, R., Lee, Y.N., Ravell, J., Zhang, Y., McElwee, J., Nugent, D., Coonrod, E.M., Durtschi, J.D., Augustine, N.H., Voelkerding, K.V., Csomos, K., Rosen, L., Browne, S., Walter, J.E., Notarangelo, L.D., Hill, H.R., Kumánovics, A. (2015). Identification of patients with RAG mutations previously misdiagnosed with common variable immunodeficiency disorders. *J Clin Immunol*, 35(2):119-24. PMID: 25516070
33. Chan, K., Frankovich, J., Copperstock, M., Cunningham, M.W., Murphy, T.K., Pasternack, M., Thienemann, M., Williams, K., Walter, J.E., Swdo,

- S.E. (2015). Clinical evaluation of youth with pediatric acute-onset neuropsychiatric syndrome (PANS): recommendations from the 2013 PANS Consensus Conference. *J Child Adolesc Psychopharmacol*, (1):3-13. PMID: 25325534 PMCID: PMC4340805
34. Mathieu, A.L., Verronese, E., Rice, G.I., Fouyssac, F., Bertrand, Y., Picard, C., Chansel, M., Walter, J.E., Notarangelo, L.D., Butte, M.J., Nadeau, K.C., Csomos, K., Chen, D.J., Chen, K., Delgado, A., Rigal, C., Bardin, C., Schuetz, C., Moshous, D., Reumaux, H., Plenat, F., Phan, A., Zobot, M.T., Balme, B., Viel, S., Bienvenu, J., Cochat, P., van der Burg, M., Caux, C., Kemp, E.H., Rouvet, I., Malcus, C., Méritet, J.F., Lim, A., Crow, Y.J., Fabien, N., Ménétrier-Caux, C., De Villartay, J.P., Walzer, T., Belot, A. (2015). PRKDC mutations associated with immunodeficiency, granuloma, and autoimmune regulator-dependent autoimmunity. *J Allergy Clin Immunol* 135(6):1578-88. PMID: 25842288, PMCID: PMC4487867
35. Dahlberg, C.M., Torres, M.L., Petersen, S.H., Baptista, M.A.P., Keszei, M., Volpi, S., Grasset, E.K., Karlsson, M.C.I., Walter, J.E., Snapper, S., Notarangelo, L.D., Westerberg, L.S. (2015). Deletion of WASp and N-WASp in B cells cripples the germinal center response and results in production of IgM autoantibodies. *Journal of Autoimmunity*, 62:81-92. PMID: 26143192, PMCID: PMC6245946
36. Gorgy, A.I., Jonassaint, N.L., Stanley, S.E., Koteish, A., Walter, J.E., DeZern, A.E., Sopa, S.C., Hamilton, J.P., Hoover-Fong, J., Chen, A.R., Anders, R.A., Kamel, I.R., Armanios, M. (2015). Hepatopulmonary syndrome is a frequent cause of dyspnea in the short telomere disorders. *Chest*. PMID: 26158642, PMCID: PMC4594621
37. Walter, J.E., Rosen, L.B., Csomos, K., Rosenberg, J.M., Mathew, D., Keszei, M., Ujhazi, B., Chen, K., Lee, Y.N., Tirosh, I., Dobbs, K., Al-Herz, W., Cowan, M., Puck, J., Blessing, J., Grimley, M.S., Melch, H., De Ravin, S.S., Gennery, A.R., Abraham, R.S., Joshi, A.Y., Boyce, T.G., Butte, M.J., Nadeau, K.C., Balboni, I., Sullivan, K.E., Akhter, J., Adeli, M., El-Feky, R., El-Ghoneimy, D., Dbaibo, G., Wakim, R., Azzari, C., Palma, P., Cancrini, C., Capuder, K., Condino-Neto, A., Costa-Carvalho, BT., Oliveira, J.B., Roifman, C., Buchbinder, D., Kumanovics, A., Franco, J.L., Niehues, T., Schuetz, C., Kuijpers, T., Yee, C., Chou, J., Masaad, M.M., Geha, R., Uzel, G., Geller, R., Holland, S.M., Recher, M., Utz, P.J., Browne, S.K., Notarangelo, L.D. (2015). Broad spectrum antibodies to self-antigens and cytokines in RAG deficiency. *Journal of Clinical Investigation J Clin Invest*, 125(11):4135-48. PMID: 2645773, PMCID: PMC4639965
38. Tineke Cantaert, T., Bannock, J.M., Ng, Y.S., Massad, C., Schickel, J.N., Chamberlain, N., Kinnunen, T., Menard, L., Lavoie, A., Walter, J.E., Notarangelo, L.D., Bruneau, J., Al-Herz, W., Kilic, S.S., Ochs, H., Nonoyama, S., Cunningham-Rundles, C., Durandy, A., Meffre, E. (2015). Abnormal induction of somatic hypermutation results in defective regulatory T cells and peripheral B-cell tolerance. *Immunity*, 17;43(5):884-95. PMID: 26546282
39. Walter, J.E., Armanios, M., Shah, U., Friedmann, A., Spitzer, T., Sharatz, S., Hagen, C. (2015). Case records of the Massachusetts General Hospital. Case-41-2015. 14-Year-Old Boy with Immune and Liver Abnormalities. *New England Journal of Medicine*, 373(27):2664-76. PMID: 26716919

40. Henderson, L., Volpi, S., Frugoni, F., Janssen, E., Kim, S., Sundel, R.P., Dedeoglu, F., Lo, M.S., Hazen, M.M., Son, M.B., Mathieu, R., Zurakowski, D., Yu, N., Lebedeva, T., Fuhlbrigge, R., Walter, J.E., Lee, Y.N., Nigrovic, P., Notarangelo, L. (2016). Next generation sequencing reveals restriction and clonotypic expansion of regulatory T cells in juvenile idiopathic arthritis. *Arthritis Rheumatol*. PMID: 2681513, PMCID: PMC5398095
41. Notarangelo LD, Kim MS, Walter JE, Lee YN. Human RAG mutations: biochemistry and clinical implications. *Nat Rev Immunol*. 2016 April;16(4):234-46. Epub 2016 Mar 21. PubMed PMID: 26996199.
42. Felgentreff, K., Baxi, S.N., Lee, Y.N., Dobbs, K., Henderson, L.A., Csomos, K., Tsitsikov, E.N., Armanios, M.Y., Walter, J.E., Notarangelo, L.D. (2016). Ligase-4 deficiency causes distinctive abnormalities in lymphocyte subsets, cellular radiosensitivity, and immune repertoire in asymptomatic individuals. *Journal of Clinical Immunology*. PMID: 27063650, PMCID: PMC4842108
43. Kumánovics, A., Lee, Y.N., Close, D.W., Coonrod, E.M., Ujhazi, B., Chen, K., MacArthur, D.G., Krivan, G., Notarangelo, L.D., Walter, J.E. (2016). Estimated disease incidence of RAG1/2 mutations: A case report and querying the Exome Aggregation Consortium. *J Allergy Clin Immunol*. PMID: 27609655, PMCID: PMC5303162
44. Lee, Y.N., Frugoni, F., Dobbs, K., Tirosch, I., Du, L., Ververs, F.A., Heng, R., Ott de Bruin, L., Adeli, M., Blesing, J.H., Buchbinder, D., Butte, M.J., Cancrini, C., Chen, K., Choo, S., Elfeky, R.A., Finocchi, A., Fuleihan, R.L., Gennery, A.R., El-Ghoneimy, D.H., Henderson, L.A., Al-Herz, W., Hossny, E., Nelson, R.P., Pai, S., Patel, N.C., Reda, S.M., Soler-Palacin, P., Somech, R., Palma, P., Wu, H., Giliani, S., Walter, J.E., Notarangelo, L.D. (2016). Characterization of T and B cell repertoire diversity in patients with RAG deficiency. *Science Immunology*, 1(6), 1-12. PMID: 28783691, PMCID: PMC5586490
45. John, T., Walter, J.E., Schuetz, C., Chen, K., Abraham, R.S., Bonfim, C., Boyce, T.G., Joshi, A.Y., Kang, E., Carvalho, B.T., Mahajerin, A., Nugent, D., Puthenveetil, G., Soni, A., Su, H., Cowan, M.J., Notarangelo, L., Buchbinder, D. (2016). Unrelated Hematopoietic Cell Transplantation in a Patient with Combined Immunodeficiency with Granulomatous Disease and Autoimmunity Secondary to RAG Deficiency. *J Clin Immunol*, 36(7), 725-32. PMID: 27539235, PMCID: PMC5286911
46. Cantaert, T., Schickel, J.N., Bannock, J.M., Ng, Y.S., Massad, C., Delmotte, F.R., Yamakawa, N., Glauzy, S., Chamberlain, N., Kinnunen, T., Menard, L., Lavoie, A., Walter, J.E., Notarangelo, L.D., Bruneau, J., Al-Herz, W., Kilic, S.S., Ochs, H.D., Cunningham-Rundles, C., van der Burg, M., Kuijpers, T.W., Kracker, S., Kaneko, H., Sekinaka, Y., Nonoyama, S., Durandy, A., Meffre, E. (2016). Decreased somatic hypermutation induces an impaired peripheral B cell tolerance Checkpoint. *J Clin Invest*. PMID: 27701145, PMCID: PMC5096912
47. Walter, J.E., Farmer, J.R., Foldvari, Z.S., Torgerson, T., Cooper, M.A. (2016). Mechanism-based strategies for the management of autoimmunity and immune dysregulation in primary immunodeficiencies. *Journal of Allergy and Clinical Immunology: in Practice*, 4(6), 1089-1100. PMID: 27836058, PMCID: PMC5289744

48. Barmettler, S., Otani, I.M., Minhas, J., Abraham, R.S., Chang, Y., Dorsey, M.J., Ballas, Z.K., Bonilla, F.A., Ochs, H.D., Walter, J.E. (2017). Gastrointestinal Manifestations in X-linked Agammaglobulinemia. *J Clin Immunol.*, 37(3), 287-294. PMID: 28236219, PMCID: PMC5414010
49. Dobbs, K., Tabellini, G., Calzoni, E., Patrizi, O., Martinez, P., Giliani, S.C., Moratto, D., Al-Herz, W., Cancrini, C., Cowan, M., Bleesing, J., Booth, C., Buchbinder, D., Burns, S.O., Chatila, T.A., Chou, J., Daza-Cajigal, V., Ott De Bruin, L.M., de la Morena, M.T., Di Matteo, G., Finocchi, A., Geha, R., Goyal, R., Hayward, A., Holland, S., Huang, C.H., Kanariou, M.G., King, A., Kaplan, B., Kleva, A., Kuijpers, T., Lee, B.W., Lougaris, V., Massaad, M., Meyts, I., Morsheimer, M., Neven, B., Pai, S.Y., Plebani, A., Prockop, S., Reisli, I., Soh, Y., Somech, R., Torgerson, T.R., Kim, Y.J., Walter, J.E., Gennery, A.R., Keles, Ss, Manis, J.M., Marcenaro, E., Moretta, A., Parolini, S., Notarangelo, L.D. (2017). NK cells from patients with RAG and non-homologous end joining (NHEJ) gene defects comprise a higher frequency of CD56bright NKG2A+++ cells, and yet display increased degranulation and higher perforin content. *Frontiers in Immunology.* 8:798. PMID: 28769923, PMCID: PMC5511964
50. Geier, C.B., Kraupp, S., Bra, D., Eibl, M.M., Farmer, J.R., Csomos, K., Walter, J.E., Wolf, H.M. (2017). Reduced numbers of circulating group 2 innate lymphoid cells in patients with common variable immunodeficiency. *European Journal of Immunology.* PMID: 28718914
51. Rowe, J.H., Stadinski, B.D., Lauren, A.H., Ott de Bruin, L., Delmonte, O., Lee, Y.N., de la Morena, M.T., Rakesh, K.G., Hayward, A., China-Hui, H., Kanariou, M., King, A., Kuijpers, T.W., Soh, J.Y., Neven, B., Walter, J.E., Huseby, E.S., Notarangelo, L.D. (2017). Abnormalities of T cell receptor repertoire in CD4+ regulatory and conventional T cells in patients with RAG mutations: implications for autoimmunity. *J Allergy Clin Immunol.* PMID: 28864286, PMCID: PMC5911433
52. Westermann-Clark, E., Grossi, A., Fioredda, F., Giardino, S., Cappelli, E., Terranova, P., Palmisani, E., Farmer, J.R., Foldvari, Z., Yamazaki, Y.7., Faraci, M., Lanino, E., Notarangelo, L.D., Dufour, C., Ceccherini, I., Walter, J.E.*, Miano, M.* (*shared senior co-authors) (2018). RAG deficiency with ALPS features successfully treated with TCR $\alpha\beta$ /CD19 cell depleted haploidentical stem cell transplant. *Clinical Immunology.* PMID: 29104089, PMCID: PMC5941932
53. Farmer, J.R., Ong, M.S., Barmettler, S., Yonker, L.M., Fuleihan, R., Sullivan, K.E., Cunningham-Rundles, C., Walter, J.E. (2018). Common variable immunodeficiency (CVID) non-infectious disease endotypes redefined using unbiased network clustering in large electronic datasets. *Frontiers of Immunology.* PMID: 29375540
54. Tirosh, I., Yamazaki, Y., Frugoni, F., Ververs, F.A., Allenspach, E.J., Zhang, Y., Burns, S., Al-Herz, W., Noroski, L., Walter, J.E., Gennery, A.R., van der Burg, M., Notarangelo, L.D., Lee, Y.N. (2018). Recombination activity of human recombination-activating gene 2 (RAG2) mutations and correlation with clinical phenotype. *J Allergy Clin Immunol.* PMID: 29772310, PMCID: PMC6295349
55. Miggelbrink, A.M., Logan, B.R., Buckley, R.H., Parrott, R.E., Dvorak, C.C., Kapoor, N., Abdel-Azim, H., Prockop, S.E., Shyr, D., Decaluwe, H., Hanson,

- I.C., Gillio, A., Dávila Saldaña, B.J., Eibel, H., Hopkins, G., Walter, J.E., Whangbo, J.S., Kohn, D.B., Puck, J.M., Cowan, M.J., Griffith, L.M., Haddad, E., O'Reilly, R.J., Notarangelo, L.D., Pai, S.Y. (2018). B-cell differentiation and IL-21 response in IL2RG/JAK3 SCID patients after hematopoietic stem cell transplantation. *Blood*. PMID: 29728406, PMCID: PMC6024640
56. Lawless, D., Geier, C.B., Farmer, J.R., Lango Allen, H., Thwaites, D., Atsckezzei, F., Brown, M., Buchbinder, D., Burns, S.O., Butte, M.J., Csomos, K., Deevi, S.V.V., Egner, W., Ehl, S., Eibl, M.M., Fadugba, O., Foldvari, Z., Green, D.M., Henrickson, S.E., Holland, S.M., John, T., Klemann, C., Kuijpers, T.W., Moreira, F., Piller, A., Rayner-Matthews, P., Romberg, N.D., Sargur, R., Schmidt, R.E., Schröder, C., Schuetz, C., Sharapova, S.O., Smith, K.G.C., Sogkas, G., Speckmann, C., Stirrups, K., Thrasher, A.J., Wolf, H.M., Notarangelo, L.D., Anwar, R., Boyes, J., Ujhazi B; NIH Rare Diseases Consortium, Thaventhiran, J., Walter, J.E.*, Savic, S* (*shared senior co-authors). (2018). Prevalence and clinical challenges among adults with primary immunodeficiency and recombination-activating gene deficiency. *J Allergy Clin Immunol*. PMID: 29477728, PMCID: PMC6058308
57. Goda, V., Malik, A., Kalmar, T., Maroti, Z., Patel, B., Ujhazi, B., Csomos, K., Hale, J.E., Chen, K., Blessing, J., Palma, P., Cancrini, C., Comeau, A.M., Krivan, G., Walter, J.E. (2018). Partial RAG deficiency in a patient with varicella infection, autoimmune cytopenia, and anticytokine antibodies. *J Allergy Clin Immunol Pract*. PMID: 29410113, PMCID: PMC6072614
58. Henrickson, S.E., Walter, J.E., Quinn, C., Kanakry, J.A., Bardakjian, T., Dimitrova, D., Ujhazi, B., Csomos, K., Bosticardo, M., Dobbs, K., Nasrallah, M., Notarangelo, L.D., Holland, S.M., Fadugba, O. (2018). Adult-Onset Myopathy in a Patient with Hypomorphic RAG2 Mutations and Combined Immune Deficiency. *J Clin Immunol*. PMID: 30159811
59. Otani, I.M., Carroll, R.W., Yager, P., Kroshinsky, D., Murphy, S., Hornick, J.L., Akin, C., Castells, M., Walter, J.E. (2018) Diffuse cutaneous mastocytosis with novel somatic KIT mutation K509I and association with tuberous sclerosis. *Clin Case Rep*. PMID: 30214774, PMCID: PMC6469384
60. Peled, A., Sarig, O., Sun, G., Samuelov, L., Ma, C.A., Zhang, Y., Dimaggio, T., Nelson, C.G., Stone, K.D., Freeman, A.F., Malki, L., Vidal, L.S., Chamarthy, L.M., Briskin, V., Mohamad, J., Pavlovski, M., Walter, J.E., Milner, J.D., Sprecher, E. (2018). Loss-of-function mutations in CARD14 are associated with a severe variant of atopic dermatitis. *J Allergy Clin Immunol*. PMID: 30248356
61. Meehan, C., Bonfim, C., Dasso, J.F., Costa-Carvalho, B.T., Condino-Neto, A., Walter, J.E. (2018). In Time: The value and global implications of newborn screening for severe combined immunodeficiency. *Rev Paul Pediatr*. PMID: 30540106
62. Barmettler, S., Ong, M.S., Farmer, J.R., Choi, H., Walter, J.E. (2018). Association of Immunoglobulin Levels, Infectious Risk, and Mortality with Rituximab and Hypogammaglobulinemia. *JAMA Netw Open*. PMID: 30646343, PMCID: PMC6324375
63. Farmer, J.R., Foldvari, Z., Ujhazi, B., De Ravin, S.S., Chen, K., Blessing, J.J.H., Schuetz, C., Al-Herz, W., Abraham, R.S., Joshi, A.Y., Costa-

- Carvalho, B.T., Buchbinder, D., Booth, C., Reiff, A., Ferguson, P.J., Aghamohammadi, A., Abolhassani, H., Puck, J.M., Adeli, M., Cancrini, C., Palma, P., Bertaina, A., Locatelli, F., Di Matteo, G., Geha, R.S., Kanariou, M.G., Lycopoulou, L., Tzanoudaki, M., Sleasman, J.W., Parikh, S., Pinero, G., Fischer, B.M., Dbaibo, G., Unal, E., Patiroglu, T., Karakucuk, M., Al-Saad, K.K., Dilley, M.A., Pai, S.Y., Dutmer, C.M., Gelfand, E.W., Geier, C.B., Eibl, M.M., Wolf, H.M., Henderson, L.A., Hazen, M.M., Bonfim, C., Wolska-Kuśnierz, B., Butte, M.J., Hernandez, J.D., Nicholas, S.K., Stepensky, P., Chandrakasan, S., Miano, M., Westermann-Clark, E., Goda, V., Kriván, G., Holland, S.M., Fadugba, O., Henrickson, S.E., Ozen, A., Karakoc-Aydiner, E., Baris, S., Kiykim, A., Bredius, R., Hoeger, B., Boztug, K., Pashchenko, O., Neven, B., Moshous, D., Villartay, J.P., Bousfiha, A.A., Hill, H.R., Notarangelo, L.D., Walter, J.E. (2019). Outcomes and Treatment Strategies for Autoimmunity and Hyperinflammation in Patients with RAG Deficiency. *J Allergy Clin Immunol Pract.* PMID: 30877075
64. Williams, K., Shorser-Gentile, L., Sarvode Mothi, S., Berman, N., Pasternack, M., Geller, D., Walter, J.E. (2019). Immunoglobulin A Dysgammaglobulinemia Is Associated with Pediatric-Onset Obsessive-Compulsive Disorder. *J Child Adolesc Psychopharmacol.* PMID: 30892924
65. Parsons, K., Cipriano, S.D., Rosen, L.B., Browne, S.K., Walter, J.E., Stone, B.L., Keeshin, S., Chen, K. (2019). Severe Facial Herpes Vegetans and Viremia in NFKB2-Deficient Common Variable Immunodeficiency. *Front Pediatr.* PMID: 30941333, PMCID: PMC6433840
66. Purswani, P., Meehan, C.A., Kuehn, H.S., Chang, Y., Dasso, J.F., Meyer, A.K., Ujhazi, B., Csomos, K., Lindsay, D., Alberdi, T., Joychan, S., Trotter, J., Duff, C., Ellison, M., Bleasing, J., Kumanovics, A., Comeau, A.M., Hale, J.E., Notarangelo, L.D., Torgersen, T.R., Ochs, H.D., Sriaroon, P., Oshrine, B., Petrovic, A., Rosenzweig, S.D., Leiding, J.W., Walter, J.E. (2019). Two Unique Cases of X-linked SCID: A Diagnostic Challenge in the Era of Newborn Screening. *Front Imm.* PMID: 31024866, PMCID: PMC6460992
67. Dorna, M.B., Barbosa, P.F.A., Rangel-Santos, A., Csomos, K., Ujhazi, B., Dasso, J.F., Thwaites, D., Boyes, J., Savic, S., Walter, J.E. (2019). Combined Immunodeficiency with Late-Onset Progressive Hypogammaglobulinemia and Normal B Cell Count in a Patient With RAG2 Deficiency. *Front Pediatr.* PMID: 31058115, PMCID: PMC6477099
68. Sriaroon, P., Chang, Y., Ujhazi, B., Csomos, K., Joshi, H.R., Zhou, Q., Close, D.W., Walter, J.E., Kumánovics, A. (2019). Familial Immune Thrombocytopenia Associated with a Novel Variant in IKZF1. *Front Pediatr.* PMID: 31069201, PMCID: PMC6491668
69. Walter, J.E., Ballow, M. (2019). The WHIM Syndrome Is No Longer a Whim. *J Allergy Clin Immunol Pract.* PMID: 31076063
70. Wu, K.Y., Purswani, P., Ujhazi, B., Csomos, K., Snezhina, M., Elissaveta, N., Stefanov, S., Sharapova, S., Ellison, M., Milojevic, D., Savic, S., Sargur, R., Walter, J.E. (2019). Arthritis in Two Patients With Partial Recombination Activating Gene Deficiency. *Front Pediatr.* PMID:31334206

71. Lawless, D., Lango, A.H., Thaventhiran, J; NIHR BioResource–Rare Diseases Consortium, Hodel, F., Anwar, R., Fellay, J., Walter, J.E., Savic, S. (2019) Predicting the Occurrence of Variants in RAG1 and RAG2. *J Clin Immunol*. PMID: 31388879
72. Suresh, S., Upton, J., Green, M., Pham-Huy, A., Posfay-Barbe, K.M., Michaels, M.G., Top, K.A., Avitzur, Y., Burton, C., Chong, P.P., Danziger-Isakov, L., Dipchand, A.I., Hébert, D., Kumar, D., Morris, S.K., Nalli, N., Ng, V.L., Nicholas, S.K., Robinson, J.L., Solomon, M., Tapiero, B., Verma A, Walter, J.E., Allen, U.D. (2019). Live vaccines after pediatric solid organ transplant: Proceedings of a consensus meeting, 2018. *Pediatr Transplant*. PMID:31497926
73. Bulkhi, A.A., Dasso, J.F., Schuetz, C., Walter, J.E. (2019). Approaches to patients with variants in RAG genes: from diagnosis to timely treatment. *Expert Rev Clin Immunol*. PMID:31535575.
74. Chinn, I.K., Chan, A.Y., Chen, K., Chou, J., Dorsey, M.J., Hajjar, J., Jongco, A.M 3rd., Keller, M.D., Kobrynski, L.J., Kumanovics, A., Lawrence, M.G., Leiding, J.W., Lugar, P.L., Orange, J.S., Patel, K., Platt, C.D., Puck, J.M., Raje, N., Romberg, N., Slack, M.A., Sullivan, K.E., Tarrant, T.K., Torgerson, T.R., Walter, J.E. (2019). Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: A working group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. *J Allergy Clin Immunol*. PMID:31568798
75. Farmer, J.R., Allard-Chamard, H., Sun, N., Ahmad, M., Bertocchi, A., Mahajan, V.S., Aicher T, Arnold J, Benson MD, Morningstar J, Barmettler S, Yuen G, Murphy SJH, Walter, J.E., Ghebremichael, M., Shalek, A.K., Batista. F., Gerszten, R., Pillai, S. (2019). Induction of metabolic quiescence defines the transitional to follicular B cell switch. *Sci Signal*. PMID:31641080
76. Walter, J.E., Ayala, I.A., Milojevic, D. (2019). Autoimmunity as a continuum in primary immunodeficiency. *Curr Opin Pediatr*. PMID:31693597
77. Barmettler, S., Ong, M.S., Farmer, J.R., Yang, N., Cobbold, M., Walter, J.E., Long, A.A., Camargo, C.A. Jr. (2019). Gastrointestinal manifestations in common variable immunodeficiency (CVID) are associated with an altered immunophenotype including B- and T-cell dysregulation. *J Allergy Clin Immunol Pract*. PMID:31704440
78. Walter, J.E., Heimall, J. (2019).CMV-Seropositive Mothers of SCID: To Breastfeed or Not? *J Allergy Clin Immunol Pract*. PMID:31706499
79. Chan, A.Y., Leiding, J.W., Liu, X., Logan, B.R., Burroughs, M., Allenspach, E.J., Skoda-Smith, S., Uzel, G., Notarangelo, L.D., Slatter, M., Gennery, A.R., Smith, A.R., Pai, S.Y, Jordan, M.B., Marsh, R.A., Cowan, M.J., Dvorak, C.C., Craddock, J.A., Prockop, S.E., Chandrakasan, S., Kapoor, N., Buckley, R.H., Parikh, S., Chellapandian, D., Oshrine, B.R., Bednarski, J.J., Cooper, M.A., Shenoy, S., Davila Saldana, B.J., Forbes, L.R., Martinez, C., Haddad, E., Shyr, D.C., Chen, K., Sullivan, K.E., Heimall, J., Wright, N., Bhatia, M., Cuvelier, G.D.E., Goldman, F.D., Meyts, I., Miller, H.K., Seidel, M.G., Vander Lugt, M.T., Bacchetta, R., Weinacht, K.G., Andolina, J.R., Caywood, E., Chong, H., de la Morena, M.T., Aquino, V.M., Shereck,

- E., Walter, J.E., Dorsey, M.J., Seroogy, C.M., Griffith, L.M., Kohn, D.B., Puck, J.M., Pulsipher, M.A., Torgerson, T.R. (2020). Hematopoietic Cell Transplantation in patients with Primary Immune Regulatory Disorders (PIRD): A Primary Immune Deficiency Treatment Consortium (PIDTC) Survey. *Front Immunol.* PMID: 32153572
80. Barmettler, S., Ong, M.S., Farmer, J.R., Yang, N., Cobbold, M., Walter, J.E., Long, A.A., Camargo, C.A. (2020). Gastrointestinal manifestations in common variable immunodeficiency (CVID) are associated with an altered immunophenotype including B- and T-cell dysregulation. *J Allergy Clin Immunol Pract.* PMID: 31704440
81. Cruz, R.J., Dasso, J.F., Duff, C., Krasnopero, D., Long, Z., Ellison, M., Nieves, D., Sriaroon, P., Asante-Koran, A., Walter, J.E. (2020). Hyaluronidase-Facilitated high-dose subcutaneous IgG effectively controls parvovirus B19 infection in a pediatric cardiac transplant patient with severe T-cell lymphopenia. *Open Forum Infec Dis.* PMID: 32391400
82. Sharapova, S.O., Skomska-Pawliszak, M., Rodina, Y.A., Wolska-Kuśnierz, B., Dabrowska-Leonik N, Mikołuc B, Pashchenko OE, Pasic S, Freiberger T, Milota, T., Formánková, R., Szaflarska, A., Siedlar, M., Avčín, T., Markelj, G., Ciznar, P., Kalwak, K., Kołtan, S., Jackowska, T., Drabko, K., Gagro, A., Pac, M., Naumova, E., Kandilarova, S., Babol-Pokora, K., Varabyou, D.S., Barendregt, B.H., Raykina, E.V., Varlamova, T.V., Pavlova, A.V., Grombirikova, H., Debeljak, M., Mersiyanova, I.V., Bondarenko, A.V., Chernyshova, L.I., Kostyuchenko, L.V., Guseva, M.N., Rascon, J., Muleviciene, A., Preiksaitiene, E., Geier, C.B., Leiss-Piller, A., Yamazaki, Y., Kawai, T., Walter, J.E., Kondratenko, I.V., Šedivá, A., van der Burg, M., Kuzmenko, N.B., Notarangelo, L.D., Bernatowska, E., Aleinikova, O.V. (2020). The Clinical and Genetic Spectrum of 82 Patients with RAG Deficiency Including a c.256_257delAA Founder Variant in Slavic Countries. *Front Imm.* PMID: 32655540
83. Barmettler, S., Coffey, K., Smith, M.J., Chong, H.J., Pozos, T.C., Seroogy, C.M., Walter, J.E., Abraham, R.S. (2020). Functional Confirmation of DNA Repair Defect in Ataxia Telangiectasia (AT) Infants Identified by Newborn Screening for Severe Combined Immunodeficiency (NBS SCID) *J Allergy Clin Immunol Pract.* PMID: 32818697
84. Hossny E., Condino-Neto A., Hammarstrom L., Walter J.E. (2020) Editorial: Screening for Primary Immunodeficiency Disorders (PIDDs) in Neonates. *Front Immunol*, 11:633266. PubMed PMID: 33424872; PMCID: PMC7793736.
85. Sharapova S.O., Pashchenko O.E., Bondarenko A.V., Vakhlyarskaya S.S., Prokofjeva T., Fedorova A.S., Savchak I., Mareika Y., Valiev T.T., Popa A., Tuzankina I.A., Vlasova E.V., Sakovich I.S., Polyakova E.A., Rumiantseva N.V., Naumchik I.V., Kulyova S.A., Aleshkevich S.N., Golovataya E.I., Minakovskaya N.V., Belevtsev M.V., Latysheva E.A., Latysheva T.V., Beznoshchenko A.G., Akopyan H., Makukh H., Kozlova O., Varabyou D.S., Ballow M., Ong M.S., Walter J.E., Kondratenko I.V., Kostyuchenko L.V., Aleinikova O.V. (2020) Geographical Distribution, Incidence, Malignancies, and Outcome of 136 Eastern Slavic Patients With Nijmegen Breakage Syndrome and NBN Founder Variant c.657_661del5. *Front Immunol*, 11:602482. PubMed PMID: 33488600; PMCID: PMC7819964.

86. Chitty-Lopez M., Westermann-Clark E., Dawson I., Ujhazi B., Csomos K., Dobbs K., Le K., Yamazaki Y., Sadighi Akha A.A., Chellapandian D., Oshrine B., Notarangelo L.D., Sunkersett G., Leiding J.W., Walter J.E. (2020) Asymptomatic Infant With Atypical SCID and Novel Hypomorphic RAG Variant Identified by Newborn Screening: A Diagnostic and Treatment Dilemma. *Front Immunol*, 11:1954. PubMed PMID: 33117328; PMCID: PMC7552884.
87. Engel E.R., Walter J.E. (2020) Rituximab and eculizumab when treating nonmalignant hematologic disorders: infection risk, immunization recommendations, and antimicrobial prophylaxis needs. *Hematology Am Soc Hematol Educ Program*, 2020(1):312-8. PubMed PMID: 33275746; PMCID: PMC7727502
88. Geier C.B., Farmer J.R., Foldvari Z., Ujhazi B., Steininger J., Sleasman J.W., Parikh S., Dilley M.A., Pai S.Y., Henderson L., Hazen M., Neven B., Moshous D., Sharapova S.O., Mihailova S., Yankova P., Naumova E., Ozen S., Byram K., Fernandez J., Wolf H.M., Eibl M.M., Notarangelo L.D., Calabrese L.H., Walter J.E. (2020) Vasculitis as a Major Morbidity Factor in Patients With Partial RAG Deficiency. *Front Immunol*, 11:574738. PubMed PMID: 33193364; PMCID: PMC7609967.
89. Calabrese, L.H., Winthrop, K., Strand, V., Yazdany, J., Walter, J.E. (2021). Type I interferon, anti-interferon antibodies, and COVID-19. *Lancet Rheumatol*. PMID: 33655222
90. Arkwright, P.D., Walter, J.E. (2021). Introducing a New Epoch in Inborn Errors of Immunity. *J Allergy Clin Immunol Pract*. PMID: 33551040
91. Hale, J.E., Platt, C.D., Bonilla, F.A., Hay, B.N., Sullivan, J.L., Johnston, A.M., Pasternack, M.S., Hesterberg, P.E., Meissner, H.C., Cooper, E.R., Barmettler, S., Farmer, J.R., Fisher, D., Walter, J.E., Yang, N.J., Sahai, I., Eaton, R.B., DeMaria, A., Notarangelo, L.D., Pai, S.Y., Comeau, A.M. (2021). Ten Years of Newborn Screening for Severe Combined Immunodeficiency (SCID) in Massachusetts. *J Allergy Clin Immunol Pract*. PMID: 33607339
92. Gaefke, C.L., Metts, J., Imanirad, D., Nieves, D., Terranova, P., Dell'Orso, G., Gambineri, E., Miano, M., Lockey, R.F., Walter, J.E., Westermann-Clark, E. (2021). Case Report: A Novel Pathogenic Missense Mutation in FAS: A Multi-Generational Case Series of Autoimmune Lymphoproliferative Syndrome. *Frontiers in Pediatrics*. PMID: 33816397
93. Westermann-Clark E., Meehan C.A., Meyer A.K., Dasso J.F., Amre D., Ellison M., Patel B., Betensky M., Hauk C.I., Mayer J., Metts J., Leiding J.W., Sriaroon P., Kumar A., Ayala I., Walter J.E. (2021) Primary Immunodeficiency in Children With Autoimmune Cytopenias: Retrospective 154-Patient Cohort. *Front Immunol*, 12:649182. PubMed PMID: 33968040; PMCID: PMC8100326.
94. Evans M.O., 2nd, Petersen M.M., Khojah A., Jyonouchi S.C., Edwardson G.S., Khan Y.W., Connelly J.A., Morris D., Majumdar S., McDermott D.H., Walter J.E., Murphy P.M. (2021) TREC Screening for WHIM Syndrome. *J Clin Immunol*, 41(3):621-8. PubMed PMID: 33415666; PMCID: PMC7925426.

95. Buchbinder D., Walter J.E., Butte M.J., Chan W.Y., Chitty Lopez M., Dimitriadis V.R., Dorsey M.J., Nugent D.J., Puck J.M., Singh J., Collins C.A. (2021) When Screening for Severe Combined Immunodeficiency (SCID) with T Cell Receptor Excision Circles Is Not SCID: a Case-Based Review. *J Clin Immunol*, 41(2):294-302. PubMed PMID: 33411155; PMCID: PMC8179373.
96. Zeitler K., Walter J., Khan F., Sriaroon C. (2021) Recurrent disseminated *Mycobacterium avium* in a female patient from Thailand with anti-interferon-gamma autoantibodies: dilemma on treatment approach. *BMJ Case Rep*, 14(1). PubMed PMID: 33509870; PMCID: PMC7845683.
97. Labrosse R., Barmettler S., Derfalvi B., Blincoe A., Cros G., Lacombe-Barrios J., Barsalou J., Yang N., Alrumayyan N., Sinclair J., Ong M.S., Camargo C.A., Jr., Walter J., Haddad E. (2021) Rituximab-induced hypogammaglobulinemia and infection risk in pediatric patients. *J Allergy Clin Immunol*. PMID: 33862010.
98. Lanlokun M., Borden A., Nieves D., Walter J.E., Albright D. (2021) X-Linked Agammaglobulinemia Presenting as Neutropenia: Case Report and an Overview of Literature. *Front Pediatr*, 9:633692. PMID: 34262886; PMCID: PMC8273273.
99. Petrov A.A., Adatia A., Jolles S., Nair P., Azar A., Walter J.E. (2021) Antibody deficiency, Chronic Lung Disease, and Comorbid Conditions: A Case Based Approach. *J Allergy Clin Immunol Pract*. PubMed PMID: 34592394.
100. Dey A., Peck J., Wilsey M., Walter J., Nguyen J., Gonzalez-Gomez I., Karjoo S. (2021) The Evolution of Very Early Onset Inflammatory Bowel Disease, Autoimmune Hepatitis, and Primary Sclerosing Cholangitis in a Young Girl. *Pubmed* PMID: 34949979, PMCID: PMC8647121
101. Westermann-Clark, E., Ballow, M., & Walter, J. E. (2022). The new quest in CTLA-4 insufficiency: How to immune modulate effectively? *J Allergy Clin Immunol* PMID: 34915039
102. Sauerwein, K. M. T., Geier, C. B., Stemberger, R. F., Akyaman, H., Illes, P., Fischer, M. B., Eibl, M. M., Walter, J. E., & Wolf, H. M. (2022). Antigen-Specific CD4+ T-Cell Activation in Primary Antibody Deficiency After BNT162b2 mRNA COVID-19 Vaccination. *Frontiers in Immunology*, 13. PMID: 35237272
103. Otani, I. M., Lehman, H. K., Jongco, A. M., Tsao, L. R., Azar, A., Tarrant, T., Engel, E., Walter, J. E., Truong, T. Q., Khan, D. A., Ballow, M., Cunningham-Rundles, C., Lu, H., Kwan, M., & Barmettler, S. (2022). Practical Guidance for the Diagnosis and Management of Secondary Hypogammaglobulinemia: A Work Group Report of the AAAAI Primary Immunodeficiency and Altered Immune Response Committees. *J Allergy Clin Immunol*. PMID: 35176351
104. Durkee-Shock J, Zhang A, Liang H, Wright H, Magnusson J, Garabedian E, Marsh RA, Sullivan KE, Keller MD; Morbidity, Mortality, and Therapeutics in Combined Immunodeficiency: Data From the USIDNET Registry. *J Allergy Clin Immunol Pract*. 2022 Feb 13, PMID: 35172220

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Books, Textbooks, Chapters

105. Walter, J.E., Notarangelo, L.D. Hemophagocytic lymphohistocytosis. In Case studies in Immunology. Chapter 14. 6th eds. R. Geha, L.D. Notarangelo, Garland Science, Taylor and Francis Group, Abingdon, UK. 2011.
106. Farmer, J., Csomos, K., Bacchetta, R., Walter, J.E. Congenital Immunodeficiency Diseases: Crossroad of Infection, Autoimmunity, Hyperinflammation, and Malignancy. In Translational Immunology: Mechanisms and Pharmacologic Approaches, Chapter 2, 1st edition, ed. S.L. Tan, Elsevier, New York, NY. 2015.
107. Walter, J.E., Butte M.J. Primary Immunodeficiency. In Comprehensive Pediatric Hospital Medicine 2nd edition. eds. Chiang V., Zaoutis L.B., Elsevier, New York, NY, 2016.
108. Walter, J.E. Early Diagnosis of Severe Combined Immunodeficiency. In Hematology, Immunology, and Genetics 3rd edition, eds. R.K. Ohls, A. Maheshwari, R.D. Christensen, R.A. Polin, Elsevier, New York, NY, 2018.
109. Walter, J.E. Immunoglobulin genetics. In UpToDate, eds. Puck J., Feldweg A.M., UpToDate, Wolters Kluwer Health, Waltham, MA, 2019.

Recording for teaching purposes

110. Walter J.E., Bonilla F.A., Notarangelo DL. Pediatric Update. Immune Deficiency in Children. (2013)
111. Walter J.E., Congenital Immunodeficiencies, Comprehensive Review of Pediatrics. (2014) (directed by David A. Link, MD, and Ronald E. Kleinman, MD, Harvard Medical School) video recorded Oct 2014
112. Walter J.E., Autoimmunity in CVID and other Primary Immunodeficiencies. (2014, 2016, 2018) BWH Board Review in Allergy and Immunology "Studio"/Distance Learning Course, video recorded Oct 2014, Oct 2016, Nov 2018
113. Stern, J., Walter J.E “Chasing the Rainbow of Immune Dysregulation in CVID”

Broadcasting

1. Walter J.E., Shrearer W. (2014) Autoimmunity due to RAG deficiency and estimated disease incidence in RAG1/2 mutations, Journal of Allergy and Clinical Immunology, online interview for the article entitled <https://www.youtube.com/watch?v=SMq35EqUegc#t=10>
2. Walker M, Sims KB, Walter J.E. (2014) Predisposition to infection and SIRS in primary mitochondrial disorders: 8 years' experience in an academic center. Webinar for Mitoaction. Aug 1, 2014

<http://www.mitoaction.org/blog/immune-function-and-mitochondrial-disease>
<https://itunes.apple.com/podcast/audio-podcast/id290467730?mt=2>

3. Walter J.E. (2015) Clues for early detection, proper immune suppression and support of patients with autoimmune disease and underlying immune deficiency. American College of Rheumatology, Highlights for presentation, San Francisco, CA

4. Walter J.E. (2018) Screening for Severe Combined Immunodeficiencies. Webinar sponsored by Blueprint Genetics. Recorded April 19, 2018.
<https://blueprintgenetics.com/resources/genetic-diagnostics-targeted-therapies-primary-immunodeficiency-diseases-pid/>

5. Kevin Wu. Clinical challenges in Recombination Activating Gene Deficiency and Arthritis. (Moderated by Walter J.E., Milojevic D.) (2018) Clinical Immunology Society Conference Webinar. Recorded October 2018
<https://www.youtube.com/watch?v=pt37q8aUTHo>

6. Walter J.E. Leiding J. (2018) Q&A: What is the “bubble boy” disease? Experts explain severe combined immunodeficiency. Article and video.
<https://www.clickorlando.com/news/qa-what-is-the-bubble-boy-disease>
https://www.clickorlando.com/getting-results-/getting-results-for-your-health/can-new-gene-therapy-save-children-from-bubble-boy-disease?fbclid=IwAR0YY98iDHs7HcFR-bb88D5jKHVEW1_BZDeevXKh2UBnjjHlxKUivJq_7P8

7. Facebook live on COVID-19 for USF

8. ESID WHIM syndrome
https://www.webevents.today/landing_pages/whim_syndrome_panel_discussion/

SCID Variants and the Irish Family, 2021 SCID Compass Summit in April.
<https://www.youtube.com/watch?v=3pD8klR-Hd0&list=PL1Gjgf3508UFBngTPwExUsQF36tMIFA97&index=8>

Other Research and Creative Achievements

Service

Memberships

Member, Hungarian Physicians’ Society

Member, American Society for Virology

Member, Infectious Diseases Society of America (IDSA)

Member, American Academy of Pediatrics (AAP)

Member, American Academy of Asthma, Allergy and Immunology (AAAAI)

Fellow, American Academy of Asthma, Allergy and Immunology (AAAAI)

Member, Robert A. Good Immunology Society (RAGIS)

Member, American Thoracic Society

Member, Clinical Immunology Society (CIS)

Member, Massachusetts SCID Newborn Screening Workgroup
Member, Hungarian Immunology Society
Member, Society of Pediatric Research
Fellow, American Academy of Asthma, Allergy and Immunology (AAAAI)
Member, Robert A. Good Honor Society, Morsani College of Medicine
American Association of Immunologists (AAI)

Service to University and Hospital

Search Committee, Walmart Chair of Nephrology, USF
USF microbiome Initiative Leadership Committee, member
Co-leader of RSC
Reviewer, Robert A. Good Honor Society, Morsani College of Medicine
Reviewer, Scientific Review Committee, Johns Hopkins All Children's Hospital
Co-Chair, Research Council, Department of Medicine, Johns Hopkins All Children's Hospital
Director, Newborn Screening Program for SCID, USF, Tampa Bay Area, FL

Service to Societies

Member, Basic and Clinical Immunology (BCI), Study Section, AAAAI
Member, Division Director's Committee, AAAAI
Diagnostic Interpretation of Genetic Studies in Patients with Primary Immunodeficiency Diseases: A Working Group Report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma, and Immunology (published in JACI)
Member, Steering Committee, SCID Compass Program, Immune Deficiency Foundation
Member, Primary Immune Regulatory Disease (PIRD) Study group, PIDTC
BCI – Genetics initiative study group – Guidelines
Program Organizing Committee, Annual Meeting of Clinical Immunology Society
Expert Review Panel – AAAAI
Steering Committee Member, National NICER group (Hematology-Immunology) Genetics, Molecular Biology & Epidemiology Committee and the Autoimmunity and Altered Immune Response Committee – AAAAI

Reviewer

Publications

Ad hoc reviewer, Journal of Allergy and Clinical Immunology, Human Immunology
Ad hoc reviewer, Clinical Immunology
Ad hoc reviewer, Journal of Clinical Immunology
Ad hoc reviewer, American Journal of Respiratory and Critical Care Medicine
Ad hoc reviewer, Frontiers of Immunology, Immunology Letters
Ad hoc reviewer, British Journal of Nutrition, Clinical Case Reports and Reviews
Ad hoc reviewer, Pediatric Blood and Cancer
Ad hoc reviewer, Journal of Allergy and Clinical Immunology: In Practice
Ad hoc reviewer, Journal of Clinical Investigations

Meeting abstracts

Ad hoc reviewer, abstracts for Annual Meeting for Clinical Immunology Society
Ad hoc reviewer, abstracts for Annual Meeting of AAAAI

Study Sections and Funding Agencies

Ad hoc reviewer, Wellcome Trust (UK)

Ad hoc reviewer, grants Jeffrey Modell Foundation, Translational Research Program

Ad hoc reviewer, grants for European Joint Programme on Rare Diseases

Scientific Review Member, Jeffrey Modell Foundation, Translational Research Grants Program 2019 (Cycle 7), 2020 (Cycle 8), 2021 (Cycle 9)

Ad hoc reviewer, NIH Center for Scientific Review:

- 2019 ZRG IMM-K (52), Primary Immunodeficiency special emphasis review panel

- 2020 ZRG1 IMM-N (52), Primary Immunodeficiency special emphasis review panel

- 2022 NIH Cellular and Molecular Immunology – A Study Section

Ad hoc reviewer, Austrian Science Fund

Editorial work:

1. *Special Issue Editor, Journal of Immunology Research*

“Advances in Primary Immunodeficiencies: Challenges in Biology, Diagnosis Management and Treatment”

2. *Chapter Editor, Encyclopedia of Medical Immunology* by Ian R. Mackay, Noel R. Rose “Congenital Immunodeficiency Syndromes”

Special Issue Editor, *Frontiers of Immunology*:

3. *Topic Editor in Frontiers of Immunology*:

- Screening for Primary Immunodeficiency Disorders (PIDDs) in Neonates (2019)

- Lessons Inborn Errors of Immunity Hold for Common Autoimmune and Autoinflammatory Disorders (2022)

- The Interplay Between the Immune System and Bone Marrow Function in Inborn Errors of Immunity and Marrow Failure (2022)

- Mechanism of Dysregulated Antibody Responses in Inborn Errors of Immunity, *Frontiers in Immunology* (2022)

4. *Member of the Editorial Board, JACI: In Practice (since 2020)*

- Editor for the February theme issue on immunodeficiencies (Feb 2021)

- Editor for July theme issue on genetics of immunodeficiencies (Jul 2022)

5. *Editor, Genetics 101 for the Practicing Immunologist* (Elsevier, by invitation) (2021)