

ECD PATIENT GATHERING

LEARN SHARE UNITE

JULY 2019



7TH ANNUAL

These annual events have proven to be a valuable contribution to this community. It is our hope that this event will enlighten and encourage all who attend. Thank you to all presenters and participants for your dedication, time, and hope!

Table of Contents

Schedule of Events July 11 – 12, 2019	1
Map of Meeting Locations San Raffaele Hospital and Hotel Rafael.....	1
Presentation Agenda Friday, 12 July - 0930 to 1630.....	2
Presenter Biographies	3
STÉPHANE BARETE, MD, PHD	3
MATTHEW COLLIN, MD, PHD.....	3
CARINE COURTILOT, MD	4
PROF. LORENZO DAGNA, MD, FACP	4
ELI L. DIAMOND, MD.....	5
JUVIANEE ESTRADA-VERAS, MD	6
RONALD S. GO, MD	7
PROF. JULIEN HAROCHE, MD, PHD	7
MARK HEANEY, MD, PHD	8
PROF. FILIP JANKU, MD, PHD	9
AUGUSTO VAGLIO, MD, PHD	9
Thank You	11
Notes	12

Schedule of Events | July 11 – 12, 2019

DATE	TIME	EVENT	LOCATION
Wednesday, 10 July	1500 - 1700	Meet & Greet Enjoy introductions before the evening social!	Hotel Rafael
Thursday, 11 July	1800 to 2000	Welcome Reception	San Raffaele Hospital: EAT Canteen
Friday, 12 July	0700 to 0900	Breakfast	Hotel Rafael
	0900 to 0930	Registration and Sign-in	San Raffaele Hospital: San Raffaele Hall
	0930 to 1630	ECD Patient & Family Gathering	San Raffaele Hospital: San Raffaele Hall

Map of Meeting Locations | San Raffaele Hospital and Hotel Rafael



Presentation Agenda | Friday, 12 July - 0930 to 1630

<i>Time</i>	<i>Subject</i>	<i>Presenter</i>
9:00	Registration/Check-In	
9:30	Welcome & Overview of ECD	Lorenzo Dagna, MD, PhD
9:40	Drug Management	Lorenzo Dagna, MD, PhD
10:00	The Drug Trial Process	Filip Janku, MD, PhD
10:20	Organ Involvement in ECD	Julien Haroche , MD, PhD
10:40	Break	
11:10	ECD and the Brain	Eli L. Diamond, MD
11:30	Question & Answer with the Experts	<i>Moderator:</i> Ronald Go, MD
12:45	Group Photo	
1:00	Lunch	
2:00	Skin Involvement	Stéphane Barete, MD, PhD
2:20	Mutations in ECD	Mark Heaney, MD, PhD
2:40	Endocrinology & Hormones	Carine Courtillot, MD
3:00	Break	
3:15	Renal & Ureteral Involvement	Augusto Vaglio, MD, PhD
3:35	Daily Care	Juvianee Estrada-Veras, MD
3:55	ECD Registry Update	Eli L. Diamond, MD Matthew Collin, MD, PhD
4:15	ECDGA Close & Evaluations	ECDGA Team

Presenter Biographies

Stéphane Barete, MD, PhD

Dermatology, Sorbonne Universités, Upmc Univ Paris Vi, Assistance Publique-Hôpitaux De Paris, Département De Dermatologie Et Allergologie, Hôpital Tenon, Centre De Référence Des Mastocytoses, Paris, France
Presentation: Skin Involvement

Contact Information

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Matthew Collin, MD, PhD

Hematology, Newcastle University, Human Dendritic Cell Laboratory, Institute of Cellular Medicine, Newcastle upon Tyne, UK
Presentations: Expert Panel, Patient Registry Update

Matthew Collin is Professor of Hematology at Newcastle University and Director of Bone Marrow Transplantation at Newcastle upon Tyne Hospitals. He received a PhD in 1992 and graduated with an MD in 1995 from Oxford University. His research focuses on the in vivo analysis of human dendritic cells and macrophages. In addition to work on histiocytosis, his studies include the genetic control of dendritic cell hematopoiesis (GATA2, IRF8 and IKZF1) and dendritic cell/macrophage homeostasis in graft versus host disease.



Dr. Collin is also a co-founder of the global ECD Patient Registry that is supported by the ECD Global Alliance.

Contact information

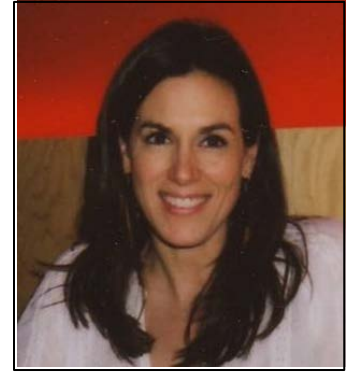
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Carine Courtillot, MD

Endocrinology, Metabolism and Reproductive Medicine, Hôpital Pitié-Salpêtrière, Paris, France

Presentation: Endocrinology & Hormones

Carine Courtillot, MD personally sees, for the endocrine evaluation and the endocrine follow-up, every patient with ECD admitted in the department of Internal Medicine at IE3M, Pitié-Salpêtrière. To date she has seen around 110 patients with ECD, having conducted a study to evaluate the endocrine consequences in ECD patients. In January 2016, Dr. Courtillot published her experience on 64 patients in the Journal of Clinical Endocrinology and Metabolism.



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Prof. Lorenzo Dagna, MD, FACP

Associate Professor of Medicine, Vita-Salute San Raffaele University, IRCCS San Raffaele Scientific Institute, Milano, Italy

Presentations: Host, Overview of ECD, Drug Management, Expert Panel

Lorenzo Dagna graduated in Medicine in 1999 and is Board Certified in Internal Medicine. He is currently Associate Professor of Medicine and Vice-Coordinator of the San Raffaele International MD Program at Vita-Salute San Raffaele University, in Milano, Italy. Prof. Dagna is also Coordinator for Clinical Immunology and Immunopathology at the Department of Internal Medicine at San Raffaele Scientific Institute, Milano.



Besides being a general internist, the clinical activity of Prof. Dagna is focused on the care of patients with immunological and rheumatic diseases, in particular those with rare immune-mediated disorders. He is also involved in both basic and clinical research on the immunopathogenesis and immunotherapy of those diseases. Lorenzo Dagna is a Fellow of the American College of Physician (ACP), Honorary Fellow of the European Federation of Internal Medicine (EFIM), and member of the Italian Society for Internal Medicine (SIMI).

Dr. Dagna serves as the Editor for Immunology and Rheumatology for the European Journal of Internal Medicine. Prof. Dagna is Chair of the Rare Disease Working Group of the European Federation of Internal Medicine, member of the Commission Expert Group on Rare Diseases, European Commission (Luxembourg, L), member of the Healthcare Professionals Working Party of the European Medicine Agency (EMA, London,

UK), member of the Medical Advisory Board of the Erdheim-Chester Global Alliance (DeRidder, LA), member of the International Foundation of Medicine Oversight Committee of the National Board of Medical Examiners (NBME, Philadelphia, PA).

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Eli L. Diamond, MD

*Neuro-Oncology, Neurology; Memorial Sloan Kettering Cancer Center;
New York, NY, USA*

Presentations: Expert Panel, ECD and the Brain, ECD Registry Update

Eli L. Diamond, MD, is a neuro-oncologist who specializes in the care of patients with ECD and other histiocytosis, as well as brain tumors and neurologic complications of cancer. He did his neurology residency at the Massachusetts General Hospital and Brigham and Women's Hospital and his neuro-oncology training at Memorial Sloan Kettering Cancer Center (MSK), where is currently a staff neuro-oncologist. He conducts and has conducted several research studies for patients with ECD including the vemurafenib trial, the cobimetinib trial, a study looking for gene mutations in ECD tumors, a study to improve ECD biopsies, and a study about ECD and the brain. He is also the co-founder of the global ECD Patient Registry that has been supported by the ECD Global Alliance. At MSK, he works with a large team across many disciplines to offer the highest quality medical care and supportive care for patients with ECD, as well as to better our understanding and treatment of ECD with collaborative research.



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Juvianee Estrada-Veras, MD

Genetics, National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland, USA
Presentation: Expert Panel Moderator



Juvianee Estrada-Veras, MD, is a Clinical Investigator at the National Human Genome Research Institute (NHGRI) at the National Institutes of Health (NIH) in Bethesda, Maryland, USA. Dr. Estrada-Veras received his MD magna cum laude from the Instituto Tecnológico de Santo Domingo (INTEC), Dominican Republic in 2003. He completed his residency at the State University of New York Health Science Center at Brooklyn and Downstate Medical Center with the Department of Pediatrics in 2009, and a fellowship at the National Institutes of Health/National Human Genome Research Institute, Bethesda, Maryland in Clinical Genetics and Medical Biochemical Genetics in 2011.

Dr. Estrada-Veras is Associate Investigator of the “Erdheim Chester Disease Natural History Protocol” and the “Dabrafenib and Trametinib in People with BRAF V600E Mutation Positive Lesions in Erdheim Chester Disease” studies at the NIH, as well as with Dr. William Gahl and colleagues in the study for diagnosis and treatment of inborn errors of metabolism at the NHGRI.

Recently, he joined the department of Medical Genetics at Walter Reed National Military Medical Center where he provides clinical and research support to The Children’s Center at Walter Reed Bethesda and The John P. Murtha Cancer Center, the only designated cancer center for the US Department of Defense (DoD). Additionally, he is Assistant Professor of Pediatrics at The Uniformed Services University of the Health Sciences.

Dr. Estrada-Veras is a member of the ECD Global Alliance Board of Directors.

Contact information

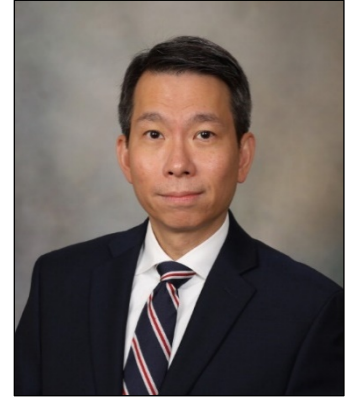
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Ronald S. Go, MD

Chair, Core/Consultative Hematology, Mayo Clinic, Rochester, MN, USA

Presentation: Expert Panel Moderator

Ronald S. Go, M.D. investigates the pathogenesis, diagnosis and treatment of histiocytic disorders such as Erdheim-Chester disease, Rosai-Dorfman disease and Langerhans cell histiocytosis as well as complement-mediated hematologic conditions such as paroxysmal nocturnal hemoglobinuria, atypical hemolytic uremic syndrome.



Dr. Go's clinical and research interests include histiocytic disorders (Erdheim-Chester disease, Langerhans cell histiocytosis, Rosai-Dorfman disease), thrombotic microangiopathies (thrombotic thrombocytopenic purpura, atypical hemolytic uremic syndrome), and monoclonal gammopathy of undetermined significance (MGUS). He also performs health outcomes research in hematologic malignancies.

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Prof. Julien Haroche, MD, PhD

Service de Médecine Interne, Hôpital Pitié-Salpêtrière, Paris, France

Presentations: Organ Involvement, Expert Panel

Julien Haroche is a professor in internal medicine, at Pitié-Salpêtrière, Paris, France. Since 2003, his main research field is Erdheim-Chester disease upon which he has acquired a world-renowned expertise. To date, he has seen more than 200 patients at his institution. His other research fields are the other histiocytoses, such as Langerhans cell histiocytosis, mixed histiocytosis and Rosai-Dorfman disease. He is also interested in vasculitis, lupus and antiphospholipid syndrome. Dr. Haroche serves on the ECD Global Alliance Medical Advisory Board.



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Mark Heaney, MD, PhD

Associate Professor of Medicine at CUMC, Columbia University Medical Center, New York, New York, USA

Presentation: Expert Panel, Mutations in ECD



Dr. Mark Heaney received his undergraduate education at Harvard and graduate education at the University of Virginia where he received an M.D. and a Ph.D. in microbiology. After Internal Medicine residency at SUNY Stony Brook, he had fellowship training in Hematology and Medical Oncology at Memorial Sloan-Kettering Cancer Center. Dr. Heaney subsequently joined the faculty at Memorial Sloan-Kettering Cancer Center. As a member of the Leukemia Service for 20 years, Dr. Heaney continued laboratory research, initially studying the cell surface receptors that govern white blood cell development and leukemia cell growth. More recently, he has focused on metabolic differences between leukemia cells and normal cells that can provide insight into how leukemia behave in the individual patient and how they respond to treatment. His finding that vitamin C has the potential to blunt the beneficial effects of chemotherapy has been widely cited.

Dr. Heaney has also been an active clinical investigator. He has been the Principal Investigator of many clinical trials that have aimed at developing new treatments for leukemia. In particular, he has focused on new treatments of the myeloproliferative neoplasms, primary myelofibrosis, polycythemia vera and essential thrombocythemia, but has also studied new treatments for chronic myeloid leukemia, mastocytosis and Langerhans cell histiocytosis in keeping with his clinical expertise in rare blood cancers.

Dr. Heaney joined the faculty at CUMC in January 2013. He has continued his commitment to caring for patients with blood cancers, developing new treatments including cutting-edge clinical trials, and laboratory research that concentrates on translating discoveries from the bench to the bedside. He also renewed his investment in medical education, becoming Director of the Hematology and Medical Oncology Fellowship Program in 2014.

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Prof. Filip Janku, MD, PhD

Assistant Professor, Oncology, the University of Texas MD Anderson Cancer Center, Houston, TX, USA
Presentation: ECD Trials & Studies



Filip Janku, MD, PhD is Assistant Professor, Department of Investigational Cancer Therapeutics, Division of Cancer Medicine at The University of Texas MD Anderson Cancer Center, Houston, TX. Dr. Janku received his MD and PhD degrees from the Charles University, Prague, Czech Republic. In addition to his expertise in Erdheim-Chester disease, Dr. Janku is principal investigator for numerous phase I trials, most of which involve novel, targeted agents, some of those being first-in-human studies. He also runs a laboratory program focused on genomic analysis of liquid biopsies. Dr. Janku received multiple awards for his research efforts, including four Merit Awards from ASCO as well as an American Association for Cancer Research Scholar-in-Training Award and highly prestigious Kimmel Award.

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Augusto Vaglio, MD, PhD

Associate Professor of Nephrology, University of Firenze and Meyer Children's Hospital, Firenze Italy
Presentation: Expert Panel, Renal & Ureteral Involvement



Dr. Vaglio graduated at the University of Parma (Italy) in 2000, and at the same institution completed his Nephrology post-graduate course (2005) and his PhD (2009). In 2005-2006, he was post-doctoral fellow at the Emory University under the supervision of Dr C. Weyand.

Dr. Vaglio's research interests include systemic vasculitis, lupus nephritis, retroperitoneal fibrosis and IgG4 related disease, and Erdheim-Chester disease. He is a member of the European Vasculitis Society (EUVAS) and is one of the coordinators the European EGPA study group. He published 148 papers in international, peer-reviewed journals, and was an invited speaker at 30 international scientific meetings.

Dr Vaglio currently serves as Nephrology Consultant and Associate Professor of Nephrology at the University of Firenze, Italy.

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THANK YOU to all attendees, the San Raffaele hosting team, speakers, and volunteers for contributing to this event and the work of the ECD Global Alliance!

SPECIAL THANKS TO THIS YEAR'S SPONSOR

Reverend Charles Balnaves is a man of faith, love, devotion, and compassion. Reverend Balnaves lost his beloved wife, Tanya, to Erdheim-Chester Disease in 2011. Rather than leaving a distant gap between himself and the disease that took his wife, Rev. Balnaves turned hurt into hope and compassion.

Since 2017, Rev. Balnaves has made the Patient & Family Gathering possible by funding the event in its entirety. This has been a major contribution to the ECDGA, allowing its limited funding to be spent on other initiatives that serve the community into the future.

Over the days, months, and years, since the loss of his wife, Rev. Balnaves has spent countless time with other ECD families all around the world.

Rev. Balnaves, the ECDGA thanks you! We are truly grateful for all the kindness that you have shown to the ECDGA and its members.



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