



2022 ANNUAL NEWSLETTER

ECD GLOBAL ALLIANCE

Supporting those affected by Erdheim-Chester Disease worldwide

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The ECD Global Alliance is dedicated to the awareness, support, education, and research related to Erdheim-Chester Disease (ECD). The organization works with the global community to accomplish its mission in supporting those living with ECD and other rare adult histiocytic disorders.

ECD Global Alliance is a 501c3 patient advocacy group supporting patients living with Erdheim-Chester Disease.

BOARD OF DIRECTORS

Since its inception in 2008, countless donors, volunteers, medical professionals, and supporters have made the ECDGA what it is today. Thanks to the leadership of our boards, patient support, and dedicated physicians, efforts have created a community filled with hope. The ECDGA is very fortunate to have the Board of Directors and Medical Advisory Board volunteer their time and energy to help lead the ECDGA into the future.



A TRIBUTE TO Mark Heaney, MD, PhD

This year we lost our longtime friend and advocate Dr. Mark Heaney. Dr. Heaney served as Medical Advisory Board Chairman, he consistently shared creative solutions and ideas that were practical and effective for the community.

He made a difference in so many lives and the impact of his contributions will live on forever. His humble and kind nature made him an absolute joy to work with and we considered him a friend. He will be greatly missed.

Kathy Brewer
President



Diane Schriener
Vice President



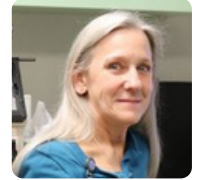
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MEDICAL ADVISORY BOARD

The medical professionals who make up the MAB are dedicated clinicians and researchers who provide medical and scientific input and direction to the organization.

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Paul Scheel, Jr., MD, F.A.S.N.D

Razelle Kurzrock, MD, FACP

Welcome from our Founder, Kathy Brewer:

Welcome to our annual newsletter! As we look back on the year, I hope you share my genuine sense of pride and gratitude for everything we've accomplished together for patients and families. Behind every story and statistic in this newsletter is the hard work and dedication of volunteers, patients and families, medical professionals, and staff. I'm honored by your continued support and look forward to what we will accomplish in 2023.

A Look Back at 2022

SECOND FDA DRUG APPROVAL FOR ECD: COBIMETINIB

In November of 2022, the Food and Drug Administration approved cobimetinib (Cotellic) for the treatment of ECD, for both BRAF V600E positive and negative. Cobimetinib is an anti-cancer medication that was first approved for the treatment of melanomas.

The ECDGA was honored to have worked with Dr. Eli L. Diamond at Memorial Sloan Kettering Cancer Center in promoting the Cobimetinib study and is grateful for all those courageous ECD patients who participated. Dr. Diamond and the staff at MSK have done a remarkable job with this outstanding work!



"For a rare disease like ECD, having two FDA-approved treatments is truly remarkable. We have made tremendous progress in understanding and treating ECD over the last several years, which is demonstrated by this significant achievement with the FDA approval. A huge shoutout to the community; I am sure this news brightened the day of many ECD warriors like myself." - Mohammad Chowdhury, ECDGA Treasurer & ECD Patient.



*"I'm extremely excited about the opportunity for better health that this treatment offers ECD patients and would like to thank our collaborators who supported us throughout the process and the patients who entrusted us with their participation in the study."
— Dr. Eli L. Diamond, Principal Investigator*

NEW CARE CENTERS ADDED TO REACH MORE PATIENTS

ECDGA welcomed three new ECDGA Care Centers in 2022, including two centers in Florida and our first center in Spain!

Hospital Universitari de Bellvitge became ECDGA's first ECD Care Center in Spain! Dr. Xavier Solanich, who specializes in internal medicine, will be the leading physician at the new center and has become one of Spain's foremost experts in treating and studying ECD!

"Our group has gained experience over the last decades caring for ECD patients and from now on we would like also to do teaching and research in this field," said Dr. Solanich. "Being part of the ECD Global Alliance allows us to improve the management of our patients and support patients cared for in smaller centers in Spain."



LIGHT IS BROUGHT TO THE CHALLENGES OF CAREGIVING FOR ECD AND RARE DISEASE PATIENTS

[A new paper](#) published in Lancet eClinicalMedicine, details both the meaningful benefits and the challenges of caregiving for patients with rare cancers, such as Erdheim-Chester Disease (ECD).

The paper was created through a collaboration of physicians and health organizations, including the Erdheim-Chester Disease Global Alliance (ECDGA), and was authored by Dr. Eli L. Diamond, Hannah-Rose Mitchell, Ph.D., MPH, and Dr. Allison J. Applebaum, to name a few, all with Memorial Sloan Kettering Cancer Center. [Click here to view the full article.](#)

REGIONAL CONNECTIONS

This year the ECDGA held a series of three Regional Patient and Family Gatherings at ECD Care Centers in the USA. Patients and medical professionals met at the University of Alabama at Birmingham, University of Cincinnati, and the University of California San Diego. It was an opportunity for patients/families to learn more about ECD and to share time with medical professionals and others affected by ECD.



**VIEW THE
PRESENTATION
RECORDINGS!**

WEBINARS

Our expert led webinars allow patients, families, and caregivers to expand their knowledge of ECD conveniently from any location. We are grateful to everyone who volunteered their time, talents, and knowledge to our webinars in 2022. [Watch Now!](#)

PATIENT NAVIGATOR PROGRAM

As a new ECDGA program, the Patient Navigator, Belinda Cobb serves as a point of contact for patients and caregivers to assist and help with information about medical processes.



CONTACT: support@erdheim-chester.org



EDUCATION & SUPPORT
FOR ECD PATIENTS LIVING
IN CHINA



DR. XINXIN CAO
Peking Union Medical
College Hospital

GLOBAL ADVOCACY EFFORTS

The ECDGA is dedicated to serving patients and families around the world living with ECD. 2022 saw important milestones in these efforts as we expanded our care networks and created new partnerships with international organizations.

[Check out our blog post](#) about the International Histo Advocacy Coalition Meeting in Sweden!



Research and Grant Updates

The ECDGA is the leading organization supporting ECD research and grants. We have invested nearly a million dollars to fund medical research that drives new treatments and resources for ECD patients and their families with more in the pipeline.



\$450,000 IN UPCOMING GRANTS

ECDGA has announced that in 2023 they will award up to \$450,000 in new research grant funding. This will be the largest investment made in grants and research in one year since ECDGA began in 2009!

FEATURED GRANTS FUNDED IN 2022

\$50,000 EARLY CAREER GRANT FOR DR. FRANCESCO PEGORARO

The ECD Global Alliance (ECDGA) has awarded the 2021 ECD Research Grant to Dr. Francesco Pegoraro, with Meyer University Children's Hospital, Florence, Italy. His winning proposal, Exploring the genetic landscape of Erdheim-Chester disease by integrating GWAS and -omic data, was selected for funding for \$50,000. The ECDGA is honored to be funding this groundbreaking work and believes it will result in a brighter future for ECD patients and a better understanding of histiocytosis.

"The aim of our project is to perform the first analysis of the monocyte methylome and transcriptome in a large cohort of patients with ECD and controls, to detect molecular pathways involved in ECD pathogenesis. The results of this study may help clarify the genetic predisposition to ECD and provide insights into how histiocytes infiltrate and damage target organs, and possibly, help discover molecules targetable by specific treatments." -Dr. Francesco Pegoraro



\$10,000 TOWARDS THE UPLIFTING ATHLETES PARTNER GRANT FOR DR. GAURAV GOYAL

Dr. Gaurav Goyal is an Assistant Professor of Hematology-Oncology and ECD Care Center lead physician at the University of Alabama at Birmingham. In collaboration with Uplifting Athletes, the ECDGA has awarded Dr. Goyal a \$20,000 grant for a Histiocytic Disorders Survivor Study (HDSS). Results from the study will be used to help devise guidelines for the follow-up care of ECD and other histiocytosis patients and develop intervention strategies in the future to mitigate long-term complications.

"Despite advances in the diagnosis and treatments of ECD, the long-term outcomes are still unknown. The motivation to conduct this study comes from our clinical practice where we routinely see patients who are in remission from ECD, yet have a high burden of symptoms and health problems that cannot be explained by their laboratory and radiographic studies (scans). The results from our study will help define the long-term healthcare needs and devise follow-up guidelines for people affected with ECD." -Dr. Gaurav Goyal



Event Highlights

PATIENT GATHERING

We are excited to announce our Annual International Patient & Family Gathering will be taking place April 27-28 in Rochester, Minnesota near the Mayo Clinic. Our annual family gathering provides two days of family and patient focused ECD education, fellowship, and opportunities to speak directly to the world's leading ECD physicians. Clinicians and medical professionals are invited to attend as well!

For more information and to register click here.



2023 CHAT SESSIONS

Patients & Community:

Monthly meetings will continue for face-to-face peer social interaction.

Caregivers:

Monthly to bi-monthly meetings for those caring for an ECD patient.

Grieving:

This support group is available to all family members that have lost someone to ECD.

International:

Spanish and French sessions will be available.

CHAT SCHEDULE

MEDICAL SYMPOSIUM

On April 26-27, 2023, the Medical Symposium will be held at the Mayo Clinic in Rochester, MN. Presentations are technical in nature and best suited for medical professionals and researchers.

To view details, register, or submit an abstract follow [this link](#).



ECD AWARENESS WEEK September 11-16, 2023

Become an ECD Community Awareness Champion this year for our 9th Annual ECD Awareness Week! Bringing awareness of ECD is critical because early detection is a key component to better treatment and health outcomes! Educating physicians is a great way to start and we hope you can help this year.

RARE DISEASE DAY February 28, 2023

We at ECDGA support Rare Disease Day! Rare Disease Day is an observance held on the last day of February to raise awareness for rare diseases and improve access to treatment and medical representation for individuals with rare diseases and their families.



BECCA BOONE FINDS SUPPORT AND FELLOWSHIP WITH ECDGA

Like many people who suffer from a rare disease, Becca Boone has spent a lot of time in doctor's offices. Becca, who is now 57, can speak in detail about a decade of tests, multiple misdiagnoses, more tests, and hours spent in waiting rooms and medical offices across the country.

It all began ten years ago when Becca found herself drinking far more water than usual and feeling pain and discomfort in her stomach. This led to her internist sending her to the hospital for a water deprivation test which resulted in him diagnosing her with Diabetes Insipidus.

As time passed, more severe symptoms crept into Becca's daily life. She began losing her balance and yellow bumps began to form around her eyes. She was frequently feeling tired and knew something was simply not right with her health.

Her doctors worked diligently to find what was causing the symptoms, testing her for and diagnosing her with everything ranging from Multiple Sclerosis to Vertigo but there always seemed to be one piece of the puzzle that was missing.

Then, in August of 2022, while sitting with her Irish Setters on her back patio she received a call from her neurologist.

She had just been diagnosed with Erdheim-Chester Disease (ECD). "I couldn't believe it," said Becca. "I was just diagnosed with this ultra-rare blood disease that I had never even heard of." And at that point, neither had most of her doctors. But her neurologist, Dr. Jill Conway, had a suspicion that it could be ECD having had another patient diagnosed with it just two years earlier by the Mayo Clinic.

Becca didn't know it at the time, but her story is common among ECD patients. Because the disease is so rare, it is often the last possible option doctors test for and often not diagnosed at all.

Doug Boone, Becca's husband recalls learning of the diagnosis both with shock and an odd sense of relief. "At least now we know what we're fighting against, and we can get to work on treatment." Doug and their two kids quickly began researching ECD online looking for more information, which led them to the ECDGA website.

At the beginning, it was very scary," said Becca. "It seemed like most of the information on the internet made you feel hopeless. Once we found the ECDGA website though and the information it provided, we stopped using google and began referring to ECDGA for information."

Dr. Conway referred Becca to a hematologist oncologist, Dr. Alan Skarbnik, for treatment. Amazingly, he too was familiar with ECD from a prior patient but admitted that his knowledge was before recent advancements in treatment developed by many of the doctors associated with the ECDGA. Through her network of physicians and the ECDGA, Becca was referred to the Memorial Sloan Kettering Cancer Center In New York City, where she is now meeting with Dr. Eli Diamond, one of the country's lead ECD physicians, to provide consultative support to her local doctors back home.

Listening to the many videos available on the ECDGA website, participating in caregiver and peer Zoom sessions, and talking with the ECDGA Patient Navigator and other team members have provided Becca and Doug hope that while finding the right treatment may be rocky, she is on a long journey of recovery and a fulfilling life with family and friends.

She even made a point to send ECD Brochures to all her previous doctors this year during ECD awareness week.

When Becca talks about her future and wanting to get back into skiing (she still struggles with her balance) and attending the next ECDGA event, you get a sense of how optimistic and hopeful she is. And, how different this feeling must be from her state of utter disbelief that afternoon in August on the back porch with her dogs.



Becca Boone enjoys spending a day with her children and dogs.

“ECDGA was one of the first groups that made me realize you can live with this disease. You can have a high quality of life and work through this. I didn't really feel that way until I discovered ECDGA and it has changed my life.” -Becca Boone

ECD PATIENT REGISTRY

FUNDED BY THE ECD GLOBAL ALLIANCE

The Erdheim-Chester Disease Registry, led by Memorial Sloan Kettering Cancer Center (MSK), unites leading researchers in building a comprehensive database with ECD patient information. Investigations into health problems caused by ECD and the result of different treatments can be performed using this database. All ECD patients with medical records written in English are encouraged to participate.

For more information [click here](#).



The Principal Investigator for the ECD Registry is Dr. Eli Diamond, who can be reached at diamone1@mskcc.org or call: 1-212-610-0243.

Studies & Trials

Participating in ECD studies and trials is one of the most important and impactful ways patients, and sometimes families, to contribute to the future of ECD treatments, monitoring, and quality of life. Scientific trials are at the heart of the medical breakthroughs that have led to so many of our current ECD treatments.

FEATURED STUDIES & TRIALS CURRENTLY RECRUITING



For complete list of patient studies and trials:
<https://erdheim-chester.org/studies-trials>

INTERNATIONAL RARE HISTIOCYTIC DISORDERS REGISTRY

The International Rare Histiocytic Disorders Registry (IRHDR) is designed to facilitate uniform diagnosis of rare histiocytic disorders. It will allow scientists to learn more about ECD and other rare histiocytic diseases. This is an observational study that does not require travel or additional testing. All ECD patients can participate who were diagnosed before 1995. For more information or to sign up click [here](#).

HISTIOCYTIC DISORDER FOLLOW-UP STUDY

You can earn \$25 for participating in this study!

This study will help us better understand possible health problems that people with histiocytic disorders live with. Dr. Gaurav Goyal, Principal Investigator, also sets out to identify people at risk for these complications to help personalize treatment in the future. Findings from these studies will also help identify those that need to be followed more closely or those that need preventive measures to ensure a healthy life free of complications. If you or your loved one have ever been diagnosed with a histiocytic disorder and speak English, you are eligible for this study. For more information or to sign up for the study click [here](#).

ECD CARE CENTERS

The International ECD Care Center program continues to grow! ECD Care Centers serve as vital medical clinics that focus on treating ECD patients and in some cases performing research to learn more about ECD. Currently, we recognize 36 ECD Care Centers internationally with 3 new centers opening in 2022.

For a full list of ECD Care Centers visit:
<https://erdheim-chester.org/care-centers/>

PHYSICIAN DIRECTORY

Medical Professionals with Erdheim-Chester Disease Knowledge/Experience

NEW!



See how
you can help
in 2023!

Supporter Spotlight

At the ECDGA, we are blessed to have the support of an amazing group of volunteers and advocates. Their dedication drives the community forward and inspires all of us to give 100% of ourselves every day to make them proud.



RuthAnn
Batchelder

"I support the ECDGA because it does so much for patients and caregivers around the world. This life can be very isolating, but with the support of the ECDGA we are connected to others who are dealing with similar issues. I am proud to be a member of the ECD community."

"Over a decade ago, on the worst day of our lives, our family was thrown a life line by Kathy Brewer and the ECD Global Alliance. Because of their dedication, perseverance, and passion my daughter has been afforded medical treatments, physician lectures, testing, coordination by the NIH and most of all hope. That's why we give and ask that you do too."



Sherry Gold



Father Charles
Balnaves

"My wife Tanya (d. 2012) lived with ECD for at least 13 years. I support ECDGA on an ongoing basis in honor of a beautiful, saintly, woman. Equally I support ECDGA because it can facilitate communication, discussion, shoulders to cry on for people with this oh-so-rare disease. Research/treatments are essential! Having someone to talk to who shares something of the same experience: a grace beyond measure. If I can help ensure those happen I have helped bring grace into the lives of others."

"I volunteer with web technology because it lets me apply my background in software development to help people, and as an ECD patient I've got a vested interest in facilitating awareness." -



Andy Skaggs



CHAT MODERATORS



Ruthann Batchelder



Fr. Charles Balnaves



Amy Teare



Julie Jones



Elaine Sanchez



Pascale Baugnet



Mariangela Martinelli



YOUR DOCTORS VOLUNTEER, TOO!

In between patient care and research, your doctors are serving the ECD community even further by creating educational materials, providing educational presentations, collaborating with other researchers and clinicians, consulting on complex cases, co-hosting events and webinars, managing the grant and care center programs and so much more. These are just a few of your supporters. Please thank your care team for going above and beyond!

Eli L. Diamond
Gaurav Goyal
Ronald Go
Paul Hendrie
Juvianee Estrada-Veras
Nishant Gupta

Ashish Kumar
Robert Franklin
Aaron Goodman
Nicole Coufal
Ila Saunders
Ginny-Tyler Meadows

A Message From Our Staff

We are grateful for your loyalty and dedication to those fighting Erdheim-Chester disease every single day. As we embark on another year together, we hope that you find inspiration, support, and hope in our services and within your community of peers.



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THE ECD GLOBAL ALLIANCE IS HERE TO HELP



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www.FundECD.org

