



# ECD Global Alliance

Supporting Those Affected by Erdheim-Chester Disease

## Organizational Newsletter

December 2012

### SAVE the DATE!!!

#### Physicians –

October 31, 2013

A one-day medical symposium will be held on October 31, 2013, in San Diego. All medical personnel interested in ECD are invited to attend.

#### Patients/Families –

November 1-2, 2013

One of the highly anticipated activities for 2013 is a patient/family meeting to be held in San Diego beginning November 1, 2013. We hope you will mark your calendar and plan to attend. You will be able to meet other patients, their families, and hear from ECD-knowledgeable doctors.

More information is available by emailing - [support@erdheim-chester.org](mailto:support@erdheim-chester.org).

### Future of the ECD Global Alliance

Most people who find the ECD Global Alliance are searching for answers about Erdheim-Chester Disease or looking for support for themselves or someone they know. The organization exists to help all those affected by ECD and all those searching for answers, including patients, family members, friends, and medical professionals.

The organization leaders, volunteers, and staff are working to help find answers. In 2013 there are a number of activities planned to support patients and their loved ones, to help promote research to improve diagnoses and treatments, and to raise awareness of ECD.

The organization is pleased to announce that it will be offering another \$50K research grant in 2013. This grant will be awarded to a researcher who submits a proposal that his/her peers find has the most scientific merit.

Another exciting project for 2013 will be the creation of a short film to help newly diagnosed patients and their local treating doctors better understand the disorder and take appropriate actions for treatment. A secondary hope for this film is that clips might also be used to help raise awareness.

One of the biggest projects the organization will begin in the coming year is the formation of an ECD Patient Registry. Patient registries are databases where patient information, including medical information and family history, is collected and stored in a standardized and secure way. The collected data will be used for medical research. Funding is being sought for this project, but the medical community has made it clear that the development of a registry has a potential to greatly help research efforts. Patients will be contacted as

progress is made on this project.

These and other projects are very aggressive for an organization as small as the ECD Global Alliance. We have confidence that our members, donors, volunteers and medical advisors will provide the help that is needed to be successful.



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### What Members Say about the ECD Global Alliance

- “That’s why we do the chats so we may become our own advocates.” – patient
- “I would like to thank you for all the help you have given our doctor and for the great work you do for the organization.” – patient
- “It helps to read others "Stories" and what they are experiencing in gaining more understanding of what ECD is and how it affects each person.” – patient waiting for diagnosis
- “Your site has been a very valuable source of information, particularly when my partner was first diagnosed, we really appreciate the work that you do. Thanks.” – loved one, registering with the organization
- “My son who is 27 years of age has just been diagnosed with ECD.... How can I help him best get through this? I feel helpless.” – loved one, registering with the organization



## Faces of the ECD Global Alliance & Medical Advisors/Researchers



From left to right: Rachel Mitchell (ECD-GA Board of Directors); Gail Hobson (ECD-GA Volunteer); Filip Janku, MD, PhD (MD Anderson Cancer Center); Juvianee Estrada-Veras, MD (National Institutes of Health); Paul Scheel, MD (Johns Hopkins); Ken McClain, MD, PhD (Texas Children's Cancer Center); Mark Heaney, MD, PhD (Memorial Sloan-Kettering); Ronald Jaffee, MB.BCh (Children's Hospital of Pittsburgh); Kathleen Brewer (ECD-GA Board of Directors); Bridget Gates (ECD-GA Volunteer); Christina Baker (ECD-GA Staff); Glenn Padnick (ECD-GA Board of Directors). Not shown: Carl Allen, MD, PhD (Texas Children's Cancer Center); Razelle Kurzrock, MD (Univ. California San Diego); Ashok Balsaver, MD (The Methodist Hospital); Larry Hobson (ECD-GA Volunteer)

*"Alone we are rare.  
Together we are strong."  
- National Organization  
of Rare Disorders  
(NORD)*



From left to right: Daniela Belloni, PhD; Lorenzo Dagna, MD; Kathy Brewer (ECD-GA); Marina Ferrarini, MD; Elisabetta Ferrero, PhD; Corrado Campochiaro, MD; San Raffaele Scientific Institute, Milan, Italy



Julien Haroche, MD, PhD (left) and Laurent Arnaud, MD, PhD (right)-Hôpital Pitié-Salpêtrière, Paris, France

## 2012 in Review

Thoughts of the past year bring hope for what the future might hold. We have seen an increase of treatment options for Erdheim-Chester Disease (ECD), more scientists are interested in studying ECD, and efforts continue to improve the lives of those suffering. We still have much to learn about this most puzzling disease, but there have been a number of accomplishments this year, each moving us toward our goals of quicker diagnoses, better treatment options, and improved care.

Dr. Juvianee Estrada-Veras, the principal investigator of the natural history study at the National Institutes of Health (NIH), tells us they will see more than 20 ECD patients this year. I think back to four or five years ago and we didn't even know how to contact more than a half-dozen ECD patients! Thank you to everyone who has been a

part of this very thorough study to help learn as much as possible about ECD.

It looks like there will be a record number of publications relative to ECD this year. The ECD Global Alliance is proud that one of those publications, by Dr. Lorenzo Dagna, was the result of the first grant awarded by the organization in 2011! We believe that this year's grant, being awarded to Dr. Julien Haroche, will also bring forth new information that will help ECD patients. These studies and publications help discover better treatment options and raise awareness leading to improved diagnoses rate.

The ECD Global Alliance is itself growing and maturing as an organization. The organization continues to maintain an up-to-date website, which is frequented by patients, family members and physicians. The weekly chat room continues to be a

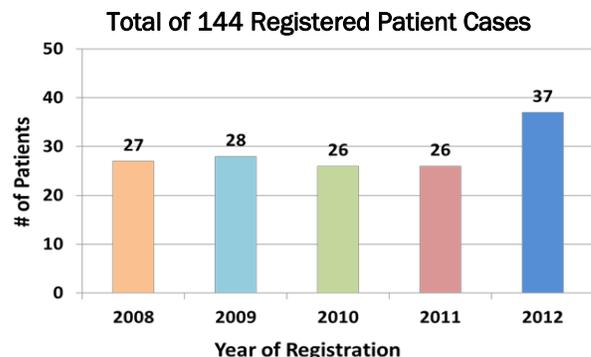
source of information sharing among patients and physicians. More patients have registered with the ECD Global Alliance this year than any year in the past. Although we wish no one ever had a need to find us, we are glad to be here to help.

I am deeply moved by all the people who give freely of their time, talent and treasure to make the ECD Global Alliance successful in its mission. I am

also deeply saddened when I remember those whom we have lost to ECD. It is in their memory that we continue the quest to battle this disease.

To everyone who is involved with the organization, patients, family members, volunteers, donors, medical professionals, and others, a heartfelt, "Thank you". Together we can make a difference. We will not be satisfied until we can find an end to the suffering for everyone.

- Kathy Brewer

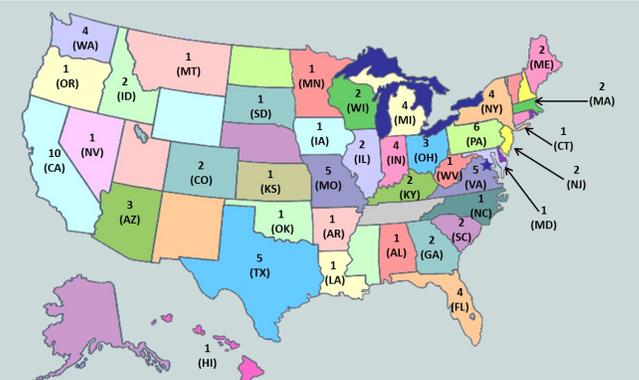


## Known ECD Studies at a Glance

Title / Principal Investigator	Study Objective
<p><u><a href="#">Clinical and Basic Investigations into Erdheim-Chester Disease</a></u>            Juvianee Estrada-Veras, M.D., National Institutes of Health, Bethesda, MD USA</p>	Natural history study designed to understand the disease, identify its cause, and classify it according to presentation and progression. The study does not include any modifications to patient treatment plans although follow on studies may address this.
<p><u><a href="#">Spit for a Cure</a></u> <b>New!!!!</b>            Carl E. Allen, M.D., Ph.D. and Kenneth McClain, M.D., Ph.D., Texas Children's Hospital, Houston, TX USA</p>	Analyze and compare the genetic information of a patient and the parents from several hundred families to identify and uncover the inherited risk factors in Histiocytic Diseases. The goal is to enroll 500 patients and their parents. All patients with a history of any histiocytic disorder and their parents are welcome to participate in this study.
<p><u><a href="#">Erdheim Chester Disease: Clinical Phenotype and Outcome. A Multicentre Survey</a></u>            Augusto Vaglio, M.D., Università degli Studi di Parma, Parma ITALY</p>	Correlate the involvement of different organs with the response to therapy and the outcome. Expand the knowledge of ECD, allowing earlier diagnosis, better classification of the different disease forms, and improved prediction of the patient prognosis.
<p><u><a href="#">Developing a Comprehensive Genomic Database - Histiocytosis Research Laboratory</a></u>            Carl E. Allen, M.D., Ph.D. and Kenneth McClain, M.D., Ph.D., Texas Children's Hospital, Houston, TX USA</p>	Understand the basic causes of histiocytosis which hopefully will lead to more effective treatments.
<p><u><a href="#">Retrospective Review of Patients With ECD Treated With Interferon-alpha or imatinib mesylate</a></u>            Filip Janku, MD., Ph.D., M. D. Anderson Cancer Center, Houston, TX 77030 USA</p>	Understand clinical characteristics, underlying biology, response to therapy and outcome of patients with Erdheim-Chester disease.
<p><u><a href="#">Pilot Study of Sirolimus with Prednisone as Treatment for ECD</a></u>            Augusto Vaglio, M.D., Università degli Studi di Parma, Parma ITALY</p>	Assess whether (and if yes, to what extent) the combination of sirolimus and prednisone may prolong progression-free survival of ECD patients.
<p><u><a href="#">Hypoxia and inflammation in Erdheim-Chester disease microenvironment: insights on the pathogenesis and implications for therapy</a></u>            Lorenzo Dagna, M.D., San Raffaele Scientific Institute, Milano, ITALY (Initial funding by the ECD Global Alliance 2011 Grant)</p>	Better define the pro-inflammatory microenvironment inside ECD lesions. To evaluate the possibility to interfere with the progression of ECD by means of specific inhibitors already available for human usage, in order to find new therapeutic options for the disease
<p><u><a href="#">Pilot Study of Tocilizumab in Patients With Erdheim-Chester Disease</a></u>            Lorenzo Dagna, M.D., San Raffaele Scientific Institute, Milano, ITALY</p>	A clinical trial of tocilizumab (Actemra) in adult patients with extraskeletal Erdheim-Chester disease (ECD). Efficacy will be assessed as the effect of the treatment on the size of the measurable lesions and by the impact of the treatment on symptom control and on patient quality of life.
<p><u><a href="#">Understanding dendritic cell lineages in Erdheim-Chester disease: towards a non-invasive diagnosis</a></u>            Laurent Arnaud, M.D., Ph.D. &amp; Julien Haroche, M.D., Ph.D., Hôpital Pitié-Salpêtrière, Paris FRANCE (To be funded by the ECD Global Alliance)</p>	Understand the primary mechanisms involved in the pathogenic differentiation of histiocytes in ECD, as well as to use this data to derive a screening & diagnostic test for ECD that would rely on blood sampling without the need for a biopsy.

### Patient Locations

Country	# patients	Country	# Patients
Australia	9	Norway	1
Bahrain	1	Pakistan	1
Belgium	1	Romania	1
Canada	4	Russia	1
Germany	1	South Africa	1
Ireland	1	Spain	2
Israel	2	Sweden	1
Italy	2	UK	9
Netherlands	2	United Arab Emirates	1
New Zealand	1	USA	102



## Financial Status of the Organization

The ECD Global Alliance continues to be committed to keeping overhead costs to a minimum and doing all possible to ensure donations are put to use toward the mission of the organization. Volunteers perform most activities of the ECD Global Alliance. However, as the organization matures, it is recognized that a staff is required to ensure the organization is stable and sustainable into the future. This year, a part-time staff member, Christina Baker, has joined the organization to help with planning and implementing projects.

Funds expended by the organization have been focused primarily on promoting medical research to learn more about Erdheim-Chester Disease.

The organization saw the completion of the 2011 \$50K grant to Lorenzo Dagna,

M.D. at San Raffaele Scientific Institute, Milan ITALY. The goal of this study was to better define the pro-inflammatory microenvironment inside ECD lesions in order to find new therapeutic options for the disease.

Another grant for \$47,000 was committed in 2012 to Laurent Arnaud, M.D., Ph.D. and Julien Haroche, M.D., Ph.D. at Hôpital Pitié-Salpêtrière, Paris FRANCE. This study will work toward the derivation of a screening & diagnostic test for ECD that would rely on blood sampling.

In November, 2012, the ECD Global Alliance also hosted a symposium attended by 8 medical researchers, from 7 different medical institutions.

The funding of these studies and the medical symposium were possible due to generous private donations. We hope

you might also feel called to make a personal donation to help ensure the support of ECD research will continue.



Rare Disease Day at the NIH on February 29, 2012. Dr. Juvanee Estrada-Veras, Linda Stowell, Pam Fleming, Kathy Brewer in front of Erdheim-Chester Disease Poster.

## Erdheim-Chester Disease Facts

*An-ultra rare disease of unknown cause, affecting many different organs.*

*Characterized by excessive production of infection fighting cells, called histiocytes, that accumulate within multiple tissues and organs.*

*Involvement may include leg and arm bones, skin, tissues behind the eyes, lungs, brain, pituitary gland, kidney, abdominal cavity, the membrane surrounding the heart, and more rarely other organs. Each patient can have a different combination of organs involved.*

*Not currently categorized as a cancer, infection, autoimmune, or storage disorder.*

*Usually affects adults, but has been found in children.*

*Less than 400 cases have been documented in the world since it was first described in 1930 by Jakob Erdheim (Austrian) and William Chester (American).*

*Unless successful treatment is found, organ failure can result.*

## ECD Global Alliance

*A nonprofit 501(c)(3) Organization*

For more information -

Website: [www.erdheim-chester.org](http://www.erdheim-chester.org)

Phone: +1-337-515-6987

E-mail: [support@erdheim-chester.org](mailto:support@erdheim-chester.org)

### Donations can be mailed to:

ECD Global Alliance  
P.O. Box 775  
DeRidder, LA 70634 USA

### Online Donations made at:

<http://www.razoo.com/story/ECD-Global-Alliance>