

ERDHEIM CHESTER DISEASE

PATIENT REGISTRY PROGRESS REPORT 2020

Thank you to the Memorial Sloan Kettering Cancer Center Registry Team for this update!



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WHAT IS THE ECD PATIENT REGISTRY?

The Erdheim-Chester Disease Registry, led by Memorial Sloan Kettering Cancer Center (MSK), unites leading researchers with people like you who are interested in taking part in building an informational database. The focus of this registry is to help scientists advance our knowledge of ECD, how it affects the lives of patients, and how ECD treatments work.

The ultimate goal – a cure!

The focus is to understand more about what kinds of health problems are caused by ECD, what happens as a result of different treatments for ECD, and how ECD affects people's feelings and attitudes. We also want to learn how these things change over time for people with ECD. To try to figure this out, doctors would like to collect information about people who have Erdheim-Chester disease and how they are treated for the disease.

YOUR IMPACT

Patient registries help doctors and scientists make key advancements in the way we treat and care for people with rare diseases like Erdheim-Chester disease (ECD). By securely collecting a defined set of health and demographic information from patients with a specific condition, registries can give physicians insights into how diseases affect patients, what treatments are beneficial, and a disease's impact on patients' quality of life. Registries can also help doctors identify previously unknown symptoms or characteristics of a condition. The ECD Registry at Memorial Sloan Kettering Cancer Center (MSK) aims to improve the lives of patients with ECD worldwide. Through the generosity of the Erdheim-Chester Disease Global Alliance community, MSK has built a robust database that is transforming treatment recommendations and symptom monitoring. We are pleased to share a summary of our findings and future directions.

HOW IT WORKS

A specialized team at MSK first secures consent for obtaining and reviewing medical information, as well as obtaining ECD samples for research. The team comprehensively reviews ECD Registry participants' medical records, biopsies, and scans to track and identify the characteristics of ECD. Individual privacy and identifiers are always protected. All information is password protected and securely stored on MSK servers, with study members accessing the information to answer important research questions. The team also collects information about patients' symptoms and general well-being that collectively helps paint a picture of what living with ECD really means.

STUDY PROGRESS AND FINDINGS

Currently, 161 eligible participants have enrolled in the study from various countries – the United States, Romania, Wales, Australia, the United Kingdom, the Philippines, Israel – and institutions, including MSK, MD Anderson, Columbia, Dana-Farber, Duke, Johns Hopkins, Mayo Clinic, Orlando Health, Stanford, Yale, Rush University, Cleveland Clinic, New York University, Broward Health, and the University of California in San Diego, Los Angeles, and San Francisco. Patients on the registry range in age from 18 years to 79 years. The average patient is 55 years old.

Gender of Registry Participants



91 (57%)



70 (43%)

Race of Registry Participants

146 (90%)

WHITE

8 (5%)

ASIAN

3 (2%)

BLACK OR AFRICAN AMERICAN

1 (<1%)

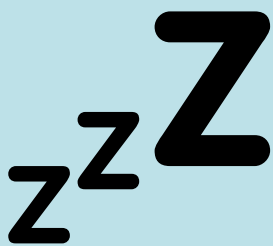
MORE THAN ONE RACE

4 (2%)

UNKNOWN

Of the participants, 161 have completed survey assessments of their symptoms and quality of life, and a detailed review of medical histories and records has been completed for 111. Additionally, expert ECD radiologists have reviewed over 100 scans (such as PET and MRI).

Our understanding of the burden of ECD symptoms has grown exponentially as a result of this registry. We've learned a great deal about commonly experienced ECD symptoms and side effects of treatment. The most frequent symptoms and side effects of ECD treatment are depicted in the tables below. Of note, we observed a tremendous delay in ECD diagnosis. On average, participants had ECD symptoms for 739 days until receiving a proper diagnosis.



MOST COMMON ECD SYMPTOMS

Symptom	% of Registry Patients Who Experience This Symptom
Fatigue or sleepiness	62%
Aching bones or joints	51%
Stress or anxiety	49%
Trouble with balance or walking	45%
Memory problems	39%

MOST COMMON SIDE EFFECTS OF ECD TREATMENT

Symptom	% of Registry Patients on Treatment Who Experience This Symptom at Least Occasionally
Headache	100%
Pain	92%
Sad feelings	88%
Swelling of the hands and feet	81%
Nausea	67%



READ MORE IN THIS STUDY ARTICLE

[A scale for patient-reported symptom assessment for patients with Erdheim-Chester disease](#)

Published March 2019

In our assessment of overall quality of life, we learned that people with ECD report a diminished quality of life, even in comparison to individuals with cancer or other chronic diseases. One important contributing factor is very severe fatigue, which on average was rated as greater than 5 on a scale from 0 to 10, with 10 indicating greatest severity. We also identified several needs for supportive care, which are listed in the table below.

SUPPORTIVE CARE

Supportive Care Need	% of Registry Patients Who Need Help With This
Lack of energy or tiredness	64%
Not being able to do things you used to do	61%
Uncertainty about the future	58%
Concerns about the worries of those close to you	53%
Learning to feel in control of your situation	51%

We've also learned about the current landscape of ECD treatments: 79% of participants are taking targeted therapies (such as BRAF or MEK inhibitors) and 21% are taking conventional treatments (like interferon, immune medications, and chemotherapy). We have not identified any differences in symptoms or quality of life for patients on these two kinds of treatment.

Thus far, we have written three articles based on ECD Registry findings: the [first-ever study of ECD symptoms](#), a detailed study of the ways that ECD affects the brain and the most effective treatments for this kind of ECD, and a study about the importance of PET scans in identifying areas of the body affected by ECD.

MSK researchers are also conducting a study to understand the experiences and needs for information and support among [ECD CAREGIVERS](#). To learn more or join this study, please email neuhistiores@mskcc.org.

COLLABORATIONS WITH OTHER RESEARCHERS

The ECD Registry has led to groundbreaking new collaborations with other clinical and research groups worldwide. In an ongoing collaboration with Assuta Medical Center in Israel, we are examining new markers in the blood (called micro RNA) as indicators of response to ECD treatment. Another collaboration with Leiden University in the Netherlands is investigating when in the blood cell development process that gene mutations (such as BRAF) occur and lead to ECD. The ECD Registry provides a rich and unparalleled source of data to allow us to partner with leading institutions to expand ECD research.

FUTURE DIRECTIONS

There are many directions for this research. Future studies will examine the best treatments for ECD's effects on other organ systems. We will also be able to analyze what types of ECD and treatments have the greatest impact on symptoms and quality of life. In addition, we are beginning a detailed study of the different blood disorders that people with ECD develop. We believe that it is critical to take the knowledge gained from this study to help achieve earlier diagnosis and treatment for people with ECD and to understand the medical complications that can accompany ECD. The advancements in our understanding of ECD and the development of new, safe, effective therapies to treat it would not be possible without the participation of patients with ECD.

If you have **questions** about the ECD Registry or would like to become a participant, please call 212-610-0720 or email neuECDRegistry@mskcc.org to speak to a study member.



LEARN MORE:

[HTTPS://ERDHEIM-CHESTER.ORG/PATIENT-REGISTRY/](https://erdheim-chester.org/patient-registry/)