



NIH “Clinical and Basic Investigations into Erdheim-Chester Disease”

Study Principal Investigator: Kevin O’Brien, CRNP

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<http://clinicaltrials.gov/ct2/show/NCT01417520?term=erdheim+chester&rank=1>

Basic Information about the Study

1. What is the purpose of the “Clinical and Basic Investigations into Erdheim-Chester Disease” study being conducted at the NIH?

The objectives of the ECD natural history study at the NIH include:

- a. Understand the disease, identify its cause, and classify it according to presentation and progression
- b. Pursue the basic defect in ECD by examining candidate genes and performing metabolic investigations

The study does NOT include changing or providing treatments to patients, although findings might lead to future therapeutic approaches.

2. Why would a patient want to participate in this study?

Patients participating in this research study will have the benefit of improved accuracy of diagnosis and a complete, free evaluation. Involvement in the study will help grow experience and knowledge of Erdheim-Chester Disease that could potentially help all patients in the future.

3. How many patients are needed for the study?

The study is currently designed for a maximum of 100 patients.

4. Would medical records of deceased patients be of help to the study?

Yes, medical records of deceased patients would be helpful. Any biopsy samples that might be available would be most helpful.

5. Who are the investigators involved in the study?

The Principal Investigator is Kevin O’Brien, CRNP (obrienke@mail.nih.gov). Associate Investigators are William Gahl, M.D., PhD, Clinical Director, NHGRI and Bernadette Gochuico, M.D., MGB, NHGRI.

Study Eligibility

6. Who is eligible to participate in the study?

To be eligible for the study, patients must:

- Have been diagnosed with Erdheim-Chester Disease based upon a pathological evaluation
- Be between 2 and 80 years of age
- Be able to travel to the NIH hospital in Bethesda, Maryland

7. Am I required to travel to Bethesda to participate in the study?

Yes, patients will need to travel to the NIH hospital in Bethesda, Maryland for 4 to 5 days in order to participate in the study.

8. Are non-US citizens able to participate in the study?

Yes, non-US citizens are able to participate. As with all patients, they will need to be accepted into the study and be able to reach the NIH campus. They will be required to bring proper identification.

What Participants Should Expect

9. How long will a patient need to stay at the NIH?

Each patient will be scheduled for 4-5 days of testing at the NIH. Patients will be admitted to the hospital as an inpatient. Depending on the patient's testing schedule, it may be possible for a patient to receive a 'pass' to leave the hospital for a short time during the week.

10. Will ECD patients be evaluated at the NIH at the same time?

No, one ECD patient a week will be scheduled to visit the NIH.

11. What tests will the patient undergo while at the NIH?

Patients will be thoroughly examined during their stay at the NIH for signs and symptoms of ECD. This examination may include, but not be limited to: (1) medical history; (2) general physical examination; (3) blood and urine samples; (4) CT, MRI, bone and/or PET scans; (5) ultrasounds; (6) skin biopsy; (7) pulmonary function test; (8) echocardiogram/ electrocardiogram/ 6-minute walk test; (9) bronchoscopy; (10) EMG and nerve conduction tests; and (11) electroencephalogram. The patient evaluation will include examinations from an ophthalmologist, pulmonologist, hematologist/oncologist, and rheumatologist. When warranted, patients may also be seen by a gastroenterologist, nephrologist, dentist, cardiologist, neurologist, endocrinologist, gynecologist, nutritionist, rehab specialist, and/or a pain consultant. In addition, photographs of patient's face and body, with underwear on, will be taken.

12. Will patients be required to use their home medications while in the NIH hospital, or will the hospital pharmacy be providing their medications, including their ECD treatments?

Patients should bring their home medications with them. Patients will be asked to send a list of their medications to the NIH in advance of their travel. The NIH will check to determine what medications can be provided by the NIH pharmacy. Patients taking medications not kept in the NIH pharmacy will be required to use their home medications while an inpatient. Refrigeration for medication will be available.

13. Will follow up visits to the NIH be required?

Plans allow for patient follow-up at the NIH every two years, with more frequent visits for selected patients who have an unusual presentation of ECD.

14. Will patients be required to stop taking medications or supplements while a participant in the study?

No, the study is not designed to alter a patient's medication regime. However, while in the hospital, it may be required for a medication to be temporarily withheld for 24 hours if a test procedure calls for this. An example could be a nuclear scan requiring contrast might require certain medications to be temporarily withheld to reduce the impact to the patient's kidney function.

15. Will a patient receive the results of their tests?

All clinical data will be given directly to the patient at a debriefing session just prior to discharge. Study participants will be encouraged to stay in contact with the principal investigator who will be able to communicate advances made. Study participants may also be invited to enroll in pertinent future studies.

Registering for the Study

16. How do patients enter the study?

Patients must provide their medical records to be accepted into the study. Scans, biopsy results, a medical summary provided by a treating or diagnosing doctor, a current medication list, and a completed medical history questionnaire are the minimum amount of records needed, although all pertinent records will be helpful.

17. How is patient participation scheduled?

Patients can contact the principal investigator, Kevin O'Brien, CRNP (obrienke@mail.nih.gov) or Kathy Brewer (support@erdheim-chester.org) to begin the process. Once the patient provides the needed medical records, s/he will work with personnel at the NIH to schedule their travel and their stay at the NIH.

Costs Related to the Study

18. Will patients be charged any medical costs for participating in the study?

No, the NIH will cover all medical costs. This includes the cost of the patient's hospital stay, tests performed and evaluations by consulting physicians.

19. Who pays for travel costs incurred by the patient?

The NIH hopes to pay travel costs for all patients who need assistance. However, the budget of the NIH may be affected by federal budget cuts that are being discussed. If this should happen, the NIH will be required to re-evaluate their ability to cover travel costs.

20. Will reimbursement of travel costs for non-US citizens be considered?

Currently the NIH will pay for travel costs incurred with the USA. This means a patient would need to cover their transportation costs from their home to US soil. Once on US soil, the NIH could then pick up the cost of travel. However, the budget of the NIH may be affected by federal budget cuts that are being discussed. If this should happen, the NIH will be required to re-evaluate their ability to cover travel costs.

21. Will travel costs be paid for a family member/caregiver traveling with a patient?

The NIH hopes to pay travel costs for a family member or caregiver traveling with the patient. However, the budget of the NIH may be affected by federal budget cuts that are being discussed. If this should happen, the NIH will be required to re-evaluate their ability to cover travel costs of family members.

22. Can the NIH cover the travel costs of multiple family members traveling with a patient?

No, the NIH is able to cover the travel costs of only a single person traveling with the patient.

Rights of the Study Participants

- 23. Will participating patients have the right to refuse a single test in the evaluation if they choose to do so?**
Yes, patients can refuse a test if they so desire. However, the goal of the study is to have a complete suite of tests on all patients for the best data and analysis. If patients refuse a procedure, it is acceptable. If a patient refuses more than one test, then a determination will need to be made as to whether that patient's participation will be warranted. Certainly, patients are encouraged to consider their participation in the study and what each test will mean to them individually. The study is a multi-year study so time is available for deliberation.
- 24. Can a patient bring a family member/care giver?**
Yes, family members are encouraged to participate in the study.
- 25. How will patient privacy be protected?**
Patient samples, cell lines and data files will be coded with numbers, not names. The code to patient identities, as well as other patient data, will be kept in a password protected database. Access to the code will be restricted to the principal investigator and an associate investigator, Dr. William Gahl. Paper records will be kept in a locked file cabinet.