

**ERDHEIM CHESTER DISEASE: CLINICAL PHENOTYPE AND OUTCOME.  
A MULTICENTRE SURVEY  
ECD001**

Consent form.

**1) Invitation paragraph**

You are being invited to take part in a research study. Before you decide it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information fully and discuss it with others if you wish.

Thank you for reading this.

**2) What is the purpose of the study?**

You have been diagnosed with a condition called Erdheim Chester Disease (ECD) which is a rare non-Langerhans cell histiocytosis. ECD is characterised by heterogeneous manifestations which largely depend on the involved sites. ECD typically affects the long bones, causing pain mainly involving the knees and the ankles; other frequently involved sites include the brain, retro-orbital tissues, the lungs, the heart, the retroperitoneum, the endocrine system and the skin. Some patients may also present with subcutaneous nodular masses.

The severity of the disease varies widely, as it may range from a localised form limited to the bone with few symptoms to an extended life-threatening disease which could be lethal. There are no established predictors of a poor outcome, although it is believed that patients with severe brain, cardiovascular or lung involvement are at higher risk of death.

The patients involved in this study are people diagnosed with ECD. We are going to study about 120 patients with the same condition.

**3) Aim of the study.**

The objective of the study is to get a "picture" of the characteristics of the patient at the diagnosis and to correlate the involvement of different organs with the response to therapy and the outcome. This study should expand the knowledge on this rare disease, thus allowing earlier diagnosis, better classification of the different disease forms, and improved prediction of the patient prognosis.

**4) What are you invited to do?**

We need to have a complete overview of the course of your disease, from diagnosis to the last examinations. For this purpose, we ask you to provide a detailed report written by the treating physician (either primary care physician or a specialist) summarising your medical history. If you desire to participate you will also have to send us hard copies or electronic copies (CD-ROM, DVD) of your most relevant imaging examinations (Bone Scintigram, CT scans of the orbit, Brain MRI, Chest CT scan, Abdominal CT scan, Echocardiography, Cardiac MRI, MRI scans of the orbit, X-rays of the long bones), blood tests and consultations (dermatological examination in case of skin masses, endocrinological evaluation in case of suspicion of diabetes insipidus or hypogonadism, etc.).

If your illness lasted less than 6 months you will not be able to participate because we need data from patients with a minimum follow-up of 6 months; if your diagnosis was made at least 6 months we will need your medical history at the moment of the diagnosis and at the sixth month; additional documentation regarding the status of your disease as well as radiological and laboratory examinations are needed at the twelfth month and the latest available visit. Anyway if you are not sure you can send us all the medical history concerning your disease.

**5) What are the possible benefits of taking part?**

If you decide to participate you will not have an immediate benefit but we hope that the information we will get from this study may help to have an earlier diagnosis and a better classification of ECD.

**6) Will your personal data be kept confidential?**

Data obtained will be used to create an electronic database and your name will be replaced by a code. All records in which your name appears will be kept strictly confidential.

Patient Consent Form

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**Chief Investigator:** Dr Augusto Vaglio

**PARTICIPANT STUDY NUMBER (please do not fill):**

**Please tick the boxes if you agree.**

1. I confirm that I have read and understood the information sheet.	
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, and without my medical care or my legal rights being affected.	
3. I agree for my clinical details (excluding personally identifiable information) to be collected and recorded on an electronic database.	
4. I agree for my personal information to be stored confidentially by Dr. Augusto Vaglio's research team so that they can contact me in the future to invite me to participate in any future related research studies. I understand that my participation in any future related study will be entirely voluntary and I can decide not to participate.	
5. I agree that my non identifiable data can be stored on a password encrypted data base for the purpose of this study and undefined future related studies. Any future study must be ethically approved.	
6. I understand that if I change my mind and withdraw consent from this study at a later date, any clinical information obtained until the time that I withdraw from the study will continue to be used for the study.	

Signed.....

Full name.....Date.....