INFORMED CONSENT FOR THE COLLECTION OF GENETIC MATERIAL

Background and Objectives

Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis is a rare hereditary disease caused by a renal tubular disorder. It has a high risk of progression to ESRD and definitive diagnosis is made by the identification of its genetic basis. Mutations in genes CLDN16 or CLDN19 are linked to the disease.

The aim of this website is to advance in current knowledge that exists on familial hypomagnesemia with hypercalciuria and nephrocalcinosis. To do so, the collection of epidemiological data, clinical and laboratory of patients with this disease, will be included in a database for later analysis. At the same time, genetic tests are made in order to identify mutations in genes involved in the disease.

Benefits of the study

This website aims to expand scientific knowledge currently available on familial hypomagnesemia with hypercalciuria and nephrocalcinosis. This will make possible an earlier diagnosis, more appropriate treatment guidelines and closer monitoring protocol, decreasing complications and improving the prognosis of this disease. Genetic studies in patients may offer an accurate diagnosis of the disease and allow us to assess a possible correlation between specific mutations and prognosis. Mutational analysis in the patient's family will also provide additional information. There are no additional medical risks associated with participation in this study, except those indicated later in relation with the collection of blood sample.

Enrolment, confidentiality and data protection

Participation in the study is completely voluntary. The information provided by your physician in this website will enable the inclusion of patient medical data in a database, and any other clinical information arising in the course of monitoring. The patient is free to present his defiance to participate in this study at any time, without any impact on his health care. To do this, you should contact first with your doctor who will notify the website the withdrawal of approval.
Participation in this survey is anonymous and confidential. It will ensure the preservation of personal identity and confidentiality at any time, according to Real Decreto 1720/2007 of December 21, Ley Orgánica 15/1999 of December 13, Ley de Protección de Datos de Carácter personal and Ley Básica 41/2002 of November 14, which regulate patient´s autonomy and the rights and duties of clinical documentation. Each patient will be assigned with an identification code and all data included will be confidential through this encrypted code. The physician can update the information in the database as new data appear in the course of the disease, but always through the identification code. The identity of persons will not be disclosed in any future publications. For additional information please consult the Privacy Policy of this website.

Blood sample for DNA testing

The purpose of obtaining and studying your DNA is the search for mutations or differences in a specific gene or genes that may be related to the disease. Since this disease has a genetic basis, the definitive diagnosis is done through genetic testing. The DNA is obtained from a blood sample from the patient, a similar procedure to that performed for a routine analysis. The possible side effects are secondary to the puncture like dizziness, fainting, pain, bleeding, bruising and, rarely, infection.

INFORMED CONSENT TO THE COLLECTION OF GENETIC MATERIAL

Hereby authorize the isolation of DNA from a blood sample in order to study only from the standpoint of Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis diagnosis.

Your DNA sample may also be conserved and used for future studies, if you show your approval (check the box of your choice)

- o Yes, I wish that my sample is retained for further study.
- o No, I prefer that my sample is removed after the study.

I have been informed of the risks of extraction a routine blood sample: possibility of pain and local hematoma. Whenever possible, this extraction can be accomplished if puncture is needed for another reason. In that case, a greater amount of blood will be collected.

I have been informed that, in relation to this study, confidentiality will be maintained at all times and identity of all individuals tested will not be disclosed in case of publications, according to Royal Decree 1720/2007 of 21 December, which approves the Regulation implementing Law 15/1999 of 13 December on the Protection of Personal Data and the Basic Law 41/2002 of November 14, Regulatory Autonomy Patient Rights and Duties of information materials and clinical documentation. If the individual study concludes in a diagnostic tool helpful for the patient and/or his family, this information will be transmitted immediately to the doctors responsible for the clinical management of the family. The researchers responsible will be available to answer questions related to the ongoing study.

I / We.……

I / We have been informed (s) and I / we understand correctly the details of the research study in which I / we, my child I / we / will be included / s, the potential benefits and risks. I / we could perform the necessary clarifying questions.

I / We have that participation is voluntary and i/we may withdraw from the study at any time, without explanation and without any impact on medical health care.

In ... ..., ..., 2.0 ... .

Patient's signature	In case of incapacity	Signature of physician / researcher
IDENTIFICATION:	Legal Representative's signature	IDENTIFICATION:
	IDENTIFICATION:	