A Clinical Study of Heart Disease in Children and Adults who have OI

The OI Foundation would like to bring to your attention a clinical study that is recruiting children and adults who have OI.

Purpose

- The purpose of this study is to gain a better understanding of the distribution and risk of different forms of heart disease in children and adults who have OI.
- To collect data on the types of heart problems experienced by children and adults with OI.
- To develop criteria for heart health screening tests for people with OI.
- To ultimately improve diagnosis and both medical and surgical care for people who have OI and develop heart disease.

Eligibility

Children and Adults, from birth to age 90, are eligible to participate. Participants must have:

- A diagnosis of OI.
- A diagnosis of a heart problem such as heart valve problems, aorta disease, other blood vessel problems, heart disease, hardening of the arteries, previous heart surgery.
- A doctor referral is required for children under age 18.
- Adults with a history of heart disease or prior heart surgery may self-refer.

Center

This study is being offered at one site – the Kennedy Krieger Institute in Baltimore, MD. The Principal Investigator is Dr. Jay Shapiro. Co-Investigators are Dr. Emily Germain-Lee (Kennedy Krieger Institute); Dr. Mary Corretti, Dr. Daniel Judge, Dr. Duke Cameron, and Dr. James Black (Johns Hopkins Hospital).

Study Description

There are two parts to this study. Some participants will only complete the OI Heart Questionnaire and some will complete both the clinical evaluation and the questionnaire.

The Clinical Evaluation

Participants will come to the OI Clinic at the Kennedy Krieger Institute to receive the following assessments:

- Health History and physical examination,
- Echocardiography,
- Lipid profile,
- Brachial artery dilation study
- Carotid artery medial wall thickness measurement
- Collagen gene mutation analysis will be performed where missing.

The OI Heart Survey

- People who participate in this part of the study can receive the questionnaire either electronically or by email.
- It is estimated that it will take 30 minutes to complete the survey.
- No visit to the Kennedy Krieger Institute is necessary for this part of the study.

The study will enroll 30 people in the Clinical Evaluation part and 70 people in only the OI Heart Questionnaire part.

To learn more about this study or to enroll

Please contact Pamela Melvin, RN at the Kennedy Krieger Institute Bone and OI Department
Telephone: 443-923-2707

The official Kennedy Krieger Institute announcement is listed below.
OI AND HEART RESEARCH PROJECT SEEKS PARTICIPANTS

We wish to inform you of a research study about heart problems in OI that we are conducting in the Osteogenesis Imperfecta Program at the Kennedy Krieger Institute (IRB Protocol#NA_00037615). We invite your participation in this study.

The OI Registry, sponsored by the Kennedy Krieger Institute and the OI Foundation, contains the following question: “Have you or your family member had surgery on the aortic or mitral valve?” 127 of a sample of 1798 registered individuals (7.06%) gave a positive response to this question. However, current information about heart disorders in OI children and adults has not been systematically approached: the real occurrence of heart valve or aortic disease or the risk of hardening of the arteries is unknown. Also, information about current cardiac surgical methods or surgical outcomes applicable to OI patients is not available.

In order to better understand possible heart conditions in OI, the Bone and Osteogenesis Imperfecta Department at the Kennedy Krieger Institute plans to evaluate the condition of children and adults with heart problems: heart valve problems, problems with the aorta or other blood vessel problems. Also, we want to learn more about heart surgery in OI patients, and the results after heart surgery. We are hopeful that the information we gain will improve heart diagnosis and care for OI patients.

The research study has two parts:

1) Participants in the OI Registry will be asked to complete a questionnaire about heart problems and prior heart surgery, if applicable.
2) Or, participants may come to the OI Clinic at the Kennedy Krieger Institute to receive the following assessments: history and physical examination, echocardiography to evaluate valvular and myocardial function, and aortic dimensions, lipid profile, brachial artery dilatation study to assess endothelial cell function, and carotid artery medial wall thickness measurement to define atherosclerotic risk. Collagen gene mutation analysis will be performed where missing.

This study will enroll 100 individuals with OI. If you are currently a patient, all procedures can be scheduled during your regular clinic visit.

If you wish additional information, please contact Pamela Melvin, RN at the Kennedy Krieger Institute Bone and OI Department at: 443 923 2707.