Patient Information Leaflet

PATIENT INFORMATION SHEET IN ENGLISH

Title: ***"*Multicentric Collaborative Study Of The Clinical Biochemical and Molecular Characterization Of Lysosomal Storage Disorders In India: The initiative For Research in Lysosomal Storage Disorders Task Force Study'**

Introduction

You are invited to take part in this research study. The information in the document is meant to help you to decide whether or not to take part. Please fill free to ask if you have any queries or concerns. You are being asked to participate in this study being conducted in AIIMS New Delhi because you satisfy our eligibility criteria which are:

- Diagnosis of LSD's
- Age between 0 days (at birth) to 18 years

Purpose of research

LSD's are rare disorders characterized by coarse facial features, neurodevelopmental delays and regression, joint contractures, organomegaly, stiffness, progressive respiratory insufficiency, cardiac vascular disease, skeletal and opthalmological changes and cervical vertebrae subluxation.The disease is usually caused by deficiency of a Lysosomal enzyme which breaks down macromolecules either forms the cell itself or acquired outside the cell leading to storage of undegraded molecules.If not treated the condition is known to lead to significant morbidity and early mortality from respiratory insufficiency.

We would like to investigate whether you have one of the diseases, what is your disease burden and if this is confirmed what mutation you carry in your genes which has caused this clinical presentation.

Study Design

Seven ml blood will be withdrawn from a peripheral vein for analysis of enzyme levels and biomarkers levels and for mutation analysis as indicated. Detailed clinical evaluation, ophthalmic examination and MRI if required. You may also be asked to give 10 ml of early morning urine sample for analysis. This procedure may require 2-3 visits to the genetic clinic.

Confidentiality of the information obtain from you

You will have the right to confidentiality regarding the privacy of your medical information (personal detail, results of physical examination, investigation and your medical history). By signing this document, you will be allowing the research team investigator and institutional ethic committee to review your information if required.

The results of clinical tests performed as a part of this research will be included in your medical record. The information from this study, if published in scientific journals or presented in scientific meeting will not reveal your identity.

How will your decision not to participate in the study affect you?

Your decision not to participate in the research study will not affect your medical care or your relationship with the investigator or the institution. Your doctor will still take care of you and you will not lose any benefit to which you are entitled.

Contact Persons

For further information/queries you can contact us at the following address.

Principal investigator Dr Madhulika Kabra/ Dr Neerja Gupta

Division of Genetics
Department of Pediatrics
AIIMS, New Delhi
Phone : 011-26594585

In case of conflict regarding your rights in the study you can contact the chair person of the institutional ethics committee at the following address

Prof T.P. Singh All India Institute of Medical Sciences, New Delhi, 110029

By signing this document it is clear that you have understood or have been told verbally all issues pertaining to the research and you are giving consent voluntarily.

Signature of Patient……. Date:……..

Signature of Witness…….. Date:…….

INFORMED CONSENT FORM

Title of Project : **Title: "Multicentric Collaborative Study Of The Clinical Biochemical and Molecular Characterization Of Lysosomal Storage Disorders In India: The initiative For Research in Lysosomal Storage Disorders Task Force Study'**

Principal investigators:

Principal investigator Dr Madhulika Kabra & Dr Neerja Gupta

Division of Genetics
Department of Pediatrics
AIIMS, New Delhi
Phone : 011-26594585

Name of Communicator:

1. I confirm that I have read and understand the information sheet dated 10/4/2010. (1st version) for the above study and have had the opportunity to ask questions.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason, without my medical care or legal rights being affected.

3. I understand that sections of any of my medical notes may be looked at by responsible individuals from AIIMS regulatory authorities where it is relevant to my taking part in research. I give permission for these individuals to have access to my records.

4. I agree to take part in the above study.

_____ _____ _____

Name of Participant Son / Daughter of (Name) Signature

Date

Researcher/ Investigator