

SBDS Protein Expression in Peripheral Blood Leukocytes

What is the purpose of this study?

Shwachman–Diamond syndrome (SDS) is a rare condition which is usually diagnosed in infancy or early childhood. It can affect a wide range of organs but most commonly the digestion and the blood system are involved. Bone abnormalities and poor growth are also often seen in SDS patients. Researchers at the Hospital for Sick Children have recently identified the altered gene (SBDS gene) that causes Shwachman-Diamond syndrome. The amount of the SBDS protein detected in the blood may vary depending on the type of SBDS mutation that is inherited. We think it is important to measure the amount of the SBDS protein because it may help us to understand why SDS affects people differently. In addition, we think that measuring the SBDS protein in blood may be helpful to make a diagnosis of SDS (or rule out the diagnosis), because it is not always possible to screen for all the SBDS mutations.

Who will be included?

Patients that have a SBDS gene mutation.

Patients that have one of the following, for which your doctor can find no specific cause

- a problem in the bone marrow and pancreas
- a probl in the pancreas, but not the bone marrow, or in the bone marrow, but not the pancreas.

What samples/information do we need?

- You will be asked to provide a single sample of blood (4 ml/1 teaspoon).
- You will also be asked questions about your health.

What is involved?

If you choose to join this study, a member of the research team will complete a form by asking you questions about your health and, if applicable, will check for documentation of bone marrow, pancreatic, and skeletal abnormalities. You will also be asked to provide a single sample of blood (4 ml/1 teaspoon). Only one blood sample is required.

White blood cells from this sample will be used to analyze for the SBDS protein and a fraction of the sample will be sent to our collaborators in Italy to study SBDS mRNA. We will also provide our Italian collaborators with clinical information collected from you and/or your medical record, but coded so your identity will not be known. The digestive enzymes (trypsinogen and isoamylase) made by the pancreas can also be measured using the serum obtained from the same blood sample.

What will be done with the samples?

Your samples will be sent to the researchers at the Hospital for Sick Children in Toronto Canada.

Collection and Storage of Personal and Medical Information:

Researchers in this study will gather information about your medical history from your chart.

The information from the research study may be published; however, you will not be identified in such publication.

What are the benefits?

If you agree to take part in this research study, there will not be a direct medical benefit for you. The information learned from this research study may benefit other patients with an immune disorder in the future.

Will I get all the facts about the study?

If you are interested in participating in this study, you will meet with a study coordinator who will explain all of the details of the study. The study coordinator will review the consent form and will be sure that all questions are answered. The consent form describes all the procedures, risks, benefits and who to contact with questions or concerns. Study procedures will not begin until the consent has been signed by the patient (if over 18 years) or the parent/guardian of the patient.

What are the risks?

A detailed list of possible risks will be provided to those patients interested in knowing more about the study.

Who should I contact for more information?

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