After you sign consent, there will be an initial clinic visit to determine whether your child is eligible to participate in the MLD Natural History Study. If your child is eligible to participate, your child will be enrolled into the study.

Enrolled patients will be asked to come to the clinic for study visits over about 2 years. Patients will come to the clinic approximately every 12 to 14 weeks during the study. Approximately four weeks after the End of Study Visit, the study doctor or nurse will contact you for a Follow-Up Visit or Telephone Call to ask about how your child is feeling and any changes in medications or new medications your child is taking.

If your child participates in this study, your child will be asked to undergo physical examinations, testing of motor functions, testing of the ability to swallow, testing of nerve conduction, testing of the ability to adjust to the environment, a questionnaire of health status, testing of cognitive behavior, collection of blood, urine, and spinal fluid, and imaging procedures to take pictures of the brain. During each clinic visit you will be asked about your child's general health and well-being in addition to any new medications or changes in medications.

Your child's study procedures and study-related follow-up care costs will be paid for by the study sponsor. The study sponsor, Shire, is paying the Institution to run the study.

What are the next steps?

For more information regarding the MLD Natural History Study, please ask your child's doctor about your child's possible participation in the study.

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MLD™
NATURAL HISTORY STUDY
of Children With Metachromatic Leukodystrophy

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Has Your Child Been Diagnosed With Metachromatic Leukodystrophy?

Your child may be a candidate for the Metachromatic Leukodystrophy Natural History Study

Has your child been diagnosed with Metachromatic Leukodystrophy?

Metachromatic Leukodystrophy (MLD) is a genetic disorder that affects the nerves, muscles, organs, and behavior. MLD is a rare disease that occurs in most parts of the world. MLD occurs in about 1 in 100,000 live births in the western world. There are three types of MLD, which include late infantile, juvenile, and adult. The late infantile form of MLD is the most common type and is usually diagnosed around 2 years of age.

What causes MLD?

MLD is a genetic disorder, specifically an inherited autosomal recessive disorder. This means that both males and females carry the gene and both parents must carry the affected gene in order to have an affected child. MLD is caused by a lack of an enzyme called arylsulfatase A or ASA. Because the ASA enzyme is missing, chemicals called sulfatides build up in the body. The buildup of the sulfatides causes damage to the nervous system, kidney, gall bladder, and other organs. Specifically in the nervous system, the sulfatides build up in the white matter of the brain and central nervous system and cause destruction of the myelin sheath or nerve coating. The myelin sheath insulates the nerve and assists communication between the nerves and the brain and through the body. Your child's doctor is the most appropriate person to answer any questions that you have related to the cause and symptoms of MLD.

How is MLD treated?

There are no approved treatments for patients with MLD. The focus of current care is to reduce symptoms and provide support to preserve the patient's quality of life through physical and occupational therapy. Your child's doctor is the most appropriate person to answer any questions that you have related to the care of a patient with MLD.

What is the MLD Natural History Study?

Our clinic is now involved in a research study to understand the natural course of MLD. This type of study is called a Natural History Study. The purpose of the MLD Natural History Study is to better understand how MLD affects your child. This information could help future patients by giving important insights into the natural course of MLD.

Who can participate?

If your child first had symptoms of MLD at or before 30 months of age, is under 12 years of age, has a minimum motor function score (as assessed by the GMFM-88 tool), and you agree for your child to undergo the study-related procedures, your child may be a candidate for the MLD Natural History Study. If your child is less than 30 months old and has experienced symptoms of MLD but is not able to walk yet, your child may be a candidate for the MLD Natural History Study.

Your child's physician is best qualified to advise you on your child's participation in this clinical study. Your child's physician will review your child's medical history and consider whether your child can be considered for this study. If your child's doctor determines that your child should be considered for this study, then your child will need to attend an initial clinic visit to determine whether or not your child is eligible for this study.

Who is not a candidate for the MLD Natural History Study?

Patients with a history of hematopoietic stem cell transplantation, use of any investigational products within 30 days prior to study enrollment or at any time during the study, or any medical condition that, in the opinion of your child's physician or the study physician, would prevent your child's participation in this trial.

How does the study work?

Your doctor will talk to you about what will be required of you and your child during the study. You will then have the chance to ask questions about any of the study procedures or requirements. If you would like for your child to participate, you will then need to sign a consent form agreeing to your child's participation in the study. This process is called informed consent.