



bridges

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A publication of the
International Spinal Muscular
Atrophy Patient Registry



Welcome to the very first edition of Bridges, a newsletter publication of the International Spinal Muscular Atrophy (SMA) Patient Registry. The name Bridges was chosen for our newsletter because it reflects the main purpose of the Registry which is to function as a bridge or connection between families wishing to participate in SMA research and researchers who wish to study SMA. Bridges will be your opportunity to learn about all of the exciting things going on here at the Registry. We'll keep you informed about your opportunities to participate in research and keep you connected to the world of SMA research. We'll let you know how the Registry is growing and what improvements you can expect. We'll introduce you to the people who make the Registry run and the members of the SMA community that help keep us connected. Join us as we work together to find a treatment and cure for SMA.



This project is supported by the Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials (ICC) which includes: Families of SMA, FightSMA, the Muscular Dystrophy Association, the Spinal Muscular Atrophy Foundation, and other SMA advocacy groups.

Contact Information

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What is the International SMA Patient Registry?

The Registry, which was established in 1986, is located at Indiana University School of Medicine. For over 25 years, researchers in the Department of Medical and Molecular Genetics (Division of Hereditary Genomics) have focused on the collection of extensive data in a number of diseases including Huntington disease, Charcot-Marie-Tooth disease, Alzheimer disease, Parkinson disease, Familial Intracranial Aneurysm and others.

Since its inception, the Registry has functioned as a “middle-man” between families interested in participating in research and researchers interested in studying SMA. With information from over 2,000 families and over 1,900 individuals affected with SMA, the Registry has helped recruit participants for clinical trials and has provided data for important SMA research studies. The Registry helps centralize information on this rare genetic disease, provides families a way to learn about research studies and connects researchers to research participants.

The Registry computerizes the names of families, including information about the person with SMA, the history of SMA in the family and other related data. Researchers interested in studying SMA can submit a request to use Registry data. All research requests are reviewed by a scientific advisory committee. Once a study has been approved, the researcher can submit one of two types of requests for information. They can request anonymous information or identifiable information. Anonymous information is de-identified, meaning that names and personal identifiers are removed and can be given to researchers without having to contact Registry families. Identifiable information includes data such as names, dates of birth, and family structure. Identifiable information can be used to help researchers find volunteers who are willing to participate in SMA research projects. In any instance where identifiable information may be given to a researcher, the Registry will contact participants in advance to ask if they are willing to share their identifiable information before any information is ever released. Participation in the Registry is voluntary. All information received by the Registry remains completely confidential.

We would like to thank the many families who have participated in the Registry by providing clinical and family history information. We believe the Registry is a valuable scientific

resource which may one day assist in the discovery of treatment and a cure for SMA. Individuals and families affected by SMA are invited to join the Registry. Visit the Registry website, <https://smaregistry.iu.edu> or call us at 1-866-482-0248 or 317-274-5745 to learn more about how to join!



The History of the Registry

In 1986 Families of SMA (FSMA) approached Indiana University School of Medicine about creating a unique registry consisting of families and individuals with Spinal Muscular Atrophy. FSMA recognized the need for patients and families to be able to contact researchers and for researchers to have access to patients and families willing to participate in research studies. The International Spinal

Muscular Atrophy Patient Registry (the Registry) was created to meet this need and to be an important tool in SMA research.

As the Registry grew, and many new SMA organizations were founded, it was soon realized that the Registry should be coordinated with input from all SMA lay organizations, researchers, scientists, physicians and family members. In September of 2004, the National Institute of Neurological Disorders and Stroke sponsored an International Spinal Muscular Atrophy Conference in Bethesda, Maryland. The goal of this meeting was to bring everyone together in a collaborative effort aimed at mapping out the future of clinical trials in SMA. The idea of everyone working together to provide oversight for the Registry was discussed and as a result of this meeting the International Coordinating Committee (ICC) for Spinal Muscular Atrophy was organized. The purpose of the ICC is to provide guidance to the SMA community at large by coordinating the efforts and resources of a number of groups, and to foster SMA research through international collaboration of researchers, physicians and families. This project is supported by the Patient Advisory Group of the International Coordinating Committee for SMA Clinical Trials (ICC) which includes: Families of SMA, FightSMA, the Muscular Dystrophy Association, the Spinal Muscular Atrophy Foundation, and other SMA advocacy groups.

Today, exciting changes are underway! The Registry is deeply involved in SMA research efforts, including helping with recruitment for several clinical trials. The Registry is also joining forces with international groups in order to further global research efforts. A new Registry website has recently been unveiled. This website allows participants to join the Registry and fill forms out online. These improvements will help carry the Registry forward, making it an even more valuable tool in SMA research!



The Registry and Research

Since the Registry was first formed, it has answered many requests for de-identified statistical data and helped researchers recruit participants for clinical trials. Below, you will find information about research studies that we are currently assisting with recruitment. For more information or to participate in these studies, please visit <https://smaregistry.iu.edu>, or call us at 1-866-482-0248 or 317-274-5745.

The SMA Registry is currently helping to recruit for several studies, including:

- **The Role of Motor Unit Number Estimation (MUNE) in Adults with Spinal Muscular Atrophy:**
MUNE is a research study that follows the course of motor neuron loss in adults with SMA using the electrophysiological technique of motor unit number estimation (MUNE).
- **Prospective Controlled Trial of Valproic Acid in Ambulant Adults with Spinal Muscular Atrophy (VALIANT SMA) Study:**
VALIANT SMA is a research study to assess the efficacy of oral Valproic Acid (VPA) in adult patients with SMA.
- **Outcome and Effect of Pregnancy in Spinal Muscular Atrophy:**
This is a research study of the effects of pregnancy and delivery on adult women with SMA. To participate in this study you do not have to have ever been pregnant.
- **Phase I/II Trial of Valproic Acid and Carnitine in Infants with Spinal Muscular Atrophy Type I (CARNI-VAL Type I):**
Carni-Val Type I is a research study to evaluate the combination of Valproic acid (VPA) and L-Carnitine for the treatment of SMA in infants with SMA type I. These clinical trials are being conducted at several universities across the United States and one location in Canada.
- **A Phase I/IIa Clinical Trial of Sodium Phenylbutyrate in Pediatric Subjects with Type I Spinal Muscular Atrophy (SMA):**
The purpose of this research is to determine the maximum tolerated dose (the highest dose) of sodium phenylbutyrate that can be safely given to children with SMA I. These clinical trials are being conducted at six universities across the United States.

- **Isolation and Characterization of Human Embryonic Stem Cells Carrying Disease Genes Obtained from IVF Clinics:**
The purpose of this study is to derive disease specific human embryonic stem cell lines that can be used to study human genetic diseases. These cell lines are obtained by using embryos that have undergone a procedure called pre-implantation genetic diagnosis (PGD).

Construction Zone - Improved Registry Website Unveiled

Recently, the Registry has seen numerous improvements and upgrades. In order to be more useful and accessible to families and researchers, we have created a new website with more information and tools. To see our new online look, visit us at <https://smaregistry.iu.edu>.

Our most exciting new feature is the Participant Portal – a feature that allows you to participate in the Registry via a secure web based connection and keeps you up to date on all the studies the Registry is recruiting for. You can continue to join and participate in the Registry by requesting forms be sent to you through the mail or by downloading them off the internet site. Now you will also be able to join and participate by filling your forms out online! Even if you have already joined and completed paper forms in the past, we encourage you to log in to the Participant Portal and complete new forms online.



Global Connections: TREAT-NMD

The global SMA community is becoming more and more connected everyday. The International Spinal Muscular Atrophy Patient Registry has joined in this global effort by teaming up with the group, Translational Research in Europe for the Assessment and Treatment of Neuromuscular Diseases (TREAT-NMD).

What is Treat-NMD?

TREAT-NMD is a global database that compiles de-identified data (data that has no names, dates of birth or other information that can identify a person) from a number of different neuromuscular diseases. All participating registries from around the world submit standardized data (everyone uses the same questions/answers) which will ensure that data can be shared, compared and combined across studies and countries. The main objective of the TREAT-NMD database is to help with clinical trials. This international database will also be used to answer questions about how common particular neuromuscular disorders are and to support other activities such as setting standards of care and diagnosis.

How do I participate?

The International SMA Patient Registry is a participating registry in Treat-NMD. As a participant in our registry, we ask your permission to forward de-identified data to Treat-NMD. This will allow you the opportunity to find out about multi-national clinical trials. To give us permission to include your de-identified data in the Treat-NMD database, we will need you to visit <https://smaregistry.iu.edu> and fill out a new informed consent document if you have not filled a new one out since October of 2008.

Global collaboration is an important key in finding a cure for SMA. With more researchers, more resources and more participants joining together, we are sure to find a way to beat SMA!



Sixty participants from twenty countries attend the 2nd Annual TREAT-NMD 2nd Registry Curator's Meeting – Fall, 2008



Coordinator's Corner - Meet the Staff

In 1984, Connie Garland began working at Indiana University School of Medicine in the Cytogenetics Division of the Department of Medical and Molecular Genetics. In 2003, she started her position as a Genetic Assistant for the International Spinal Muscular Atrophy Patient Registry. Connie answers calls and corresponds with families, enters SMA data, participates in monthly SMA conference calls and helps researchers who wish to use SMA data to complete the process of having their study approved. Connie has attended four Families of SMA (FSMA) conferences and has met many registry families in person. She is looking forward to the upcoming FSMA conference and hopes to meet new families who have recently joined. Look for her and other Registry staff at the upcoming conference.