

An Introduction to

The Spina Bifida Research Resource

We would like to take this opportunity to introduce ourselves and The Spina Bifida Research Resource (SBRR). We are a group of scientists who share a common goal. ***We want to find the causes of spina bifida.*** However, we can not achieve this goal without your help. The causes of spina bifida can not be identified by scientists alone. Families such as yours must also become involved in this important work. We would like to invite you to join the SBRR and become our partner in the quest to find the causes of spina bifida.

What is the SBRR?

The SBRR is a source of information for scientists and families who are interested in the causes of spina bifida. Families who are members of the SBRR are provided with information about current research relating to the causes of spina bifida, including research conducted by scientists who are members of the SBRR. This information will be included in future issues of the SBRR newsletter. Families may also call the SBRR to ask specific questions and/or to

arrange for a member of the SBRR to give an educational seminar.

Families who are members of the SBRR are asked to provide us with information that will be used in research studies conducted by the SBRR scientists. While we certainly hope that every family will provide this information, you are not required to provide this information to be a member of the SBRR. The SBRR newsletter and other resources are available to all families, whether or not they choose to provide this information.

What information does the SBRR need?

Each family that joins the SBRR is asked to complete an interview. This interview includes questions about your health, and the health of your children and relatives. Some family members are also asked to provide a sample of blood or cheek cells (cheek cells are obtained by rubbing the inside of your cheek with a small brush or swab). These samples are used to obtain a sample of DNA (the genetic material). We hope that every family that joins the SBRR

will provide all of this information. However, you may choose to provide only a portion of this information without affecting the usefulness of the information that you do provide.

How is this information collected?

If you are interested in joining the SBRR, or if you just would like to hear more about it, please contact Katy Hoess at 1-866-275-SBRR. She will provide additional information and answer any questions. In addition, she will arrange for the collection of all necessary information. No extra clinic visits or other travel are required to participate.

Families that join the SBRR are asked to complete an interview and to provide some samples. The interview takes about 30 minutes and will be completed by telephone. The individual with spina bifida, and some other family members, are also asked to provide a sample of blood or cheek cells. Samples of cheek cells, which are easy and painless to obtain, can be collected by mail. Special kits will be sent to all family member who wish to participate. Families that join the SBRR may choose to provide all, some, or none of this information.

Why do we need this information?

It is thought that spina bifida is caused by the interaction of a number of different risk factors. Scientists believe that these risk factors include both genes and environmental agents, and that each individual risk factor only increases the chance that an individual will be born with spina bifida by a small amount. However, very little is known about the specific genes and environmental agents that are risk factors for spina bifida. In addition, since the cause of spina bifida appears to be very complex, information from several hundred families may be needed to identify these risk factors.

The information collected by the SBRR provides a resource for studying specific genes and environmental agents that may be risk factors for spina bifida. The interview that SBRR families complete provides information about environmental agents that may increase the chance that an individual is born with spina bifida. In addition, the DNA from blood or cheek cell samples provided by members of the SBRR families can be used to study genes that may cause spina bifida. When information on both environmental agents and genes are available from the same family, it is also possible to study how these different risk factors interact to cause spina bifida.

How will this information be used?

The information provided by SBRR families will be used by the SBRR scientists to study genes and environmental agents that may influence whether an individual develops spina bifida. Currently, we are studying genes that are involved in the metabolism of folic acid, a vitamin also known as folate. Recent studies have shown that taking folate prior to and during early pregnancy can prevent some, but not all cases of spina bifida. We do not know how folate acts to prevent spina bifida, or why it does not prevent all cases spina bifida.

However, we suspect that the answer to these questions may lie in our genes. Our next newsletter will focus on the connection between spina bifida and folate.

How can I get additional information about the SBRR?

For more information about the SBRR, please contact Katy Hoess at 1-866-275-SBRR or 1-215-573-9319 or by email at khoess@cceb.med.upenn.edu

The SBRR Scientists and Staff

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Dr. Mitchell is an epidemiologist who has been at the University of Pennsylvania since 1996. Her research focuses on the causes of common birth defects, including spina bifida, cleft lip and palate, and heart defects.

Katy Hoess, M.S.

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Ms. Hoess is a graduate of the genetic counseling program at Beaver College, Glenside, Pennsylvania. She is the clinical coordinator for the SBRR.

A. Steven Whitehead, D.Phil

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Dr. Whitehead has recently joined the faculty at the University of Pennsylvania. He comes from Trinity College in Dublin, Ireland where he was actively involved in studies of genes that may act as risk factors for spina bifida.