

Thank You

The SBRR began enrolling families at the Children's Hospital of Philadelphia in 1997. Since that time, the SBRR has grown in many ways. In 2000, we received our first grant from the NIH and we began to recruit families from other institutions. In 2002, Dr. Mitchell moved to The Institute of Biosciences and Technology in Houston, Texas, a bilingual (English/Spanish) project coordinator was added to the staff, and we began to recruit families across the United States. Our goals through all of this were to recruit 700 families that were affected by spina bifida, and to work with these families to increase our understanding of the causes of spina bifida.

We are pleased to report that we have reached our initial goals. Six hundred and seventy-seven families have enrolled in the SBRR, and the information provided by these families has been used to study several genes that may be related to the risk of spina bifida. Two of these genes are discussed in this newsletter, and information about the other genes can be found in past newsletters, which are available on our website (www.sbrr.info) or by contacting the SBRR, toll free at: 1-866-521-SBTR (Barbara Weyland) or 1-866-275-SBRR (Katy Hoess).

We could not have done it without you - the individuals and families who have contributed their time (and DNA!) to the SBRR.



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Because of the progress that we have made, the SBRR has been awarded an additional five years of funding by the National Institutes of Health. Future issues of this newsletter will include details of our plans for the next five years. Be assured that we will continue to enroll families in the SBRR, and that there will be new activities for you to participate in if you choose. So, be on the lookout for future newsletters and other mailings from the SBRR.

In addition to the families that have joined the SBRR we would also like to thank the groups that have made financial contributions to the SBRR.

Aspen East Health and Fitness Center

For the second year in row, Aspen East Health and Fitness Center in Montclair, New Jersey sponsored a fund raising event for the SBRR. In order to top last year, this year's event kept participants cycling outside of the fitness center for 101 continuous hours. We truly appreciate the generosity and endurance of all of the event organizers and participants.

Spina Bifida Association of Houston Gulf Coast

The Spina Bifida Association of Houston Gulf Coast has plenty of worthwhile activities to spend money on, not the least of which is sending their kids to camp. We are honored by their support of the SBRR.

Research Update

The SBRR scientists continue to analyze the data that has been provided by families enrolled in Phase I. This work has led to the publication of two recent scientific publications.

Reminder: Each gene that we study has more than one form. The different forms of a gene are called alleles. Different alleles may function differently. Every individual has two alleles of each gene (one inherited from their mother and one from their father). The combination of alleles for each gene is called a genotype.

Loss of functions polymorphisms in *NAT1* protect against spina bifida. Jensen LE, Hoess K, Mitchell LE, Whitehead AS. Human Genetics 2006; 120:52-57.

NAT1 is a gene that is involved in controlling the amount of folate (produced from folic acid) in cells, and modifying chemicals (e.g. cigarette smoke) in the body. *NAT1* has many different alleles. Analysis of the SBRR data provided evidence that the *NAT1* genotypes of both the mother and the baby are related to the risk of spina bifida. Certain rare alleles of *NAT1* have little or no activity. Having one or more of these alleles appears to protect against spina bifida. Having less *NAT1* activity could protect against spina bifida by increasing the amount of folate available to the cells or by decreasing the amount of a harmful chemical. The SBRR

For each gene that we study, we are trying to answer three questions:

Is the gene associated with spina bifida?

Is the risk of spina bifida associated with the genotype of the mother, the genotype of the baby or both?

Does the effect of the genotype depend on other genes or environmental factors?

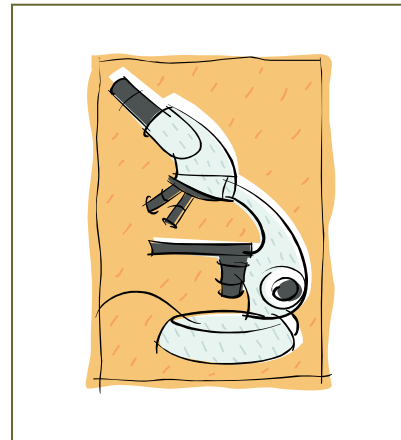
data suggest that individuals with two copies of the common (active) allele are 2.7 times more likely to be born with spina bifida than individuals with at least one rare (inactive) allele, and that mothers who have two copies of the common allele are 2.4 times more likely to have a child with spina bifida. These results need to be confirmed in larger studies and by other groups.

*“..SBRR data provided evidence that the *NAT1* genotypes of both the mother and baby are related to the risk of spina bifida”*



Maternal Genotype for the Monocyte Chemoattractant Protein 1 A(-2518)G Promotor Polymorphism Is Associated With the Risk of Spina Bifida in Offspring. Jensen LE, Etheredge AJ, Brown KS, Mitchell LE, Whitehead AS. American Journal of Medical Genetics 2006; 140A:1111-1118.

The monocyte chemoattractant protein 1 (MCP1) gene is involved in controlling the body's response to factors such as infection or fever. MCP1 is also active very early in the development of the fetus. Analysis of the SBRR data indicates that the mother's (but not the baby's) genotype for MCP1 may be associated with the risk that her child will have spina bifida. This is the first report of an association between spina bifida and this gene, so these results should be considered preliminary until confirmed in larger studies and by other groups.



SBRR Bulletin Board

Dr. Mitchell was named an "Outstanding Contributor" by the Spina Bifida Association of Houston Gulf Coast for her work on the SBRR.

Welcome to our new families from...
...SBAs of Maine and Central Ohio
and those who have found us
through the internet.

Thank you to...
...the SBAs that have added a link to
the SBRR on their websites.

¿HABLA ESPANOL?

Ahora tenemos información sobre el estudio de espina bifida y formas de participación en español. Si usted habla español y necesita literatura en su lengua, llame a Barbara Weyland, gratis, a 1-866-521-7289. ¡Gracias!

We are starting Phase II of enrollment!

The SBRR welcomes the participation of any individual with spina bifida, regardless of geographical location in the US.

Please let your friends know about us!

Have you moved? Planning to move? Changed your name?

Send your new information to us so that you don't miss out on future issues of the SBRR Newsletter.

The Institute of Biosciences and Technology took second place in the company team section for their contribution to the Spina Bifida Association of Houston Gulf Coast's Second Annual Walk-A-Thon.

the
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research
resource**

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causes of spina bifida**

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