

## Risk Factors for Spina Bifida

*When a couple has a baby with spina bifida, they often ask Why did this happen? . There is usually no simple answer to this question. In fact, there are only a few things that are **known** to increase the risk of having a child with spina bifida.*

*In this newsletter we review the known risk factors for spina bifida, and discuss other factors that are suspected (but not proven) to increase the risk of having a child with spina bifida.*

### What is a risk factor?

In the United States, about 1 in every 2,000 pregnancies is affected by spina bifida. This means that for an average couple, the risk of having a child with spina bifida is about 1 in 2,000 or 0.05%. A risk factor is anything that increases, or makes this risk higher. For example, if something increases the risk of having a child with spina bifida by 2, the risk of having a child with spina bifida would increase to about 2 in 2,000 or 0.1%.

### Known Risk Factors for Spina Bifida

#### Diabetes

Women who have diabetes before they become pregnant are at higher risk of having a child with spina bifida, and other types of birth defects, than are women who do not have diabetes. The risk of having a child with spina bifida, for a woman who has diabetes before she becomes pregnant, is 2 — 10 times higher than the risk for a woman who does not have diabetes. To help reduce the risk of having a child with a birth defect, women who are diabetic should obtain medical care prior to becoming pregnant and

maintain careful control of their glucose (sugar) levels before and during their pregnancies.

Some women develop diabetes during their pregnancy. This type of diabetes is called **gestational diabetes**. Women who develop mild gestational diabetes do not seem to have a higher risk of having a child with spina bifida or other type of birth defect. However, a recent study suggests that the risk of having a child with a birth defect may be higher in women who develop

gestational diabetes with hyperglycemia (high blood sugar levels). More work is needed to determine whether gestational diabetes with hyperglycemia is truly a risk factor for spina bifida and other birth defects.

Men who have diabetes do not appear to be at higher risk of having a child with spina bifida or other birth defects.



### Prescription Drugs

Many of the drugs that are used to treat epilepsy (seizures) can increase the risk of having a child with spina bifida and other birth defects, if they are taken by a woman during the first three months of pregnancy. These drugs include **valproic acid** and **carbamazepine**. The risk of having a child with a birth defect is 2 — 4 times higher in women who use these drugs than in women who do not. In addition, the risk is higher in women who use two or more of these drugs than in women who use only one.

In addition to being used to treat epilepsy, valproic acid may also be used in the treatment of bipolar disorder and to prevent

migraine headaches. Carbamazepine may also be used to treat bipolar disorder and for pain relief. Women who use these drugs for any reason should discuss their treatment options with their physician before they become pregnant.

Valproic acid and carbamazepine belong to a group of drugs called folic acid antagonists. Drugs in this group interfere with the body's use of folic acid, and may increase the risk of having a child with spina bifida by decreasing the amount of folic acid that is available for the developing baby. Recent studies suggest that other drugs that are folic acid antagonists may also increase the risk of having a child with spina bifida. These drugs include: **phenobarbital, phenytoin, primidone, sulfasalazine, triamterene and trimethoprim**. Women who use these, or any drug, should discuss their treatment options with their physician before they become pregnant.

Men who take seizure medications and other folic acid antagonists do not appear to be at increased risk of having a child with spina bifida or other birth defects.

### Family History

Couples who have had a child with spina bifida or anencephaly are at increased risk of having another child with a one of these conditions. For couples who have had one affected child, the risk of having a second child with spina bifida or anencephaly is approximately 3% (3 in 100) or 30 times the risk for a couple that has never had an affected child. However, the risk for any one couple will depend on whether they have other relatives who have spina bifida/anencephaly, and whether there are other known risk factors (for example, diabetes).

Individuals who have had a child with spina bifida/anencephaly are at increased risk of having another affected child, even if they change partners (for example, due to a divorce). In addition, individuals who have a one or more relatives (sibs, cousins, etc.) who are/were affected with spina bifida/anencephaly may also be at increased risk of having an affected child.

Individuals who have had a child with spina bifida/anencephaly, or who have one or more affected relatives should seek genetic counseling prior to becoming pregnant. A genetic counselor can provide estimates of

risk for specific situations, provide information about ways to reduce risk, and discuss options for prenatal diagnosis of these conditions.



### Folic Acid

Women who do not take folic acid prior to and during early pregnancy have a higher risk of having a child with spina bifida than women who take folic acid. This has been shown to be true both for women who have never had an affected child and for women who have previously had an affected child. Past issues of the SBRR Newsletter have covered the topic of folic acid in detail. If you would like copies of these newsletters please visit our website ([www.sbrr.info](http://www.sbrr.info)) or call our toll free number and request the Winter 1998, and Winter 2001-2002 newsletters.

## Suspected Risk Factors for Spina Bifida

### Body Temperature

A person's normal body temperature is about 98.6 degrees. There is some evidence that increased body temperature or hyperthermia, during early pregnancy may increase the risk of having a child with spina bifida. Studies suggest that if a woman's body temperature goes up to 101 — 102 degrees or higher, during early pregnancy, her risk of having a child with spina bifida may double.

A common cause of hyperthermia is fever. Women who develop a fever during their pregnancy should contact their health care provider.

Hyperthermia may also result from the use of hot tubs, very hot baths, saunas and possibly tanning beds. It is also possible that intense exercise, or exercising in extremely hot, humid weather, may cause hyperthermia. Pregnant women, and women who are planning to become pregnant, should avoid activities that could raise their body temperature.



### Obesity

Several studies suggest that women who are obese (very overweight) before they become pregnant are at an increased risk for having a child with spina bifida. Many of these studies measured obesity using the body mass index or BMI. (There are several internet sites that provide BMI calculators, if you are interested in calculating your BMI.) In these studies the risk of having a child with spina bifida was about 2 times higher in women who had a high or very high BMI.

We do not know why women who are very overweight may be at increased risk for having a child with spina bifida. In addition, we do not know whether losing weight will reduce their risk. It is possible that the factors which cause someone to be overweight may also increase their risk of having a child with spina bifida. If this is true, then weight loss alone may not reduce the risk of having a child with spina bifida.

Women who are concerned about their weight should consult with their physician before they begin a weight loss program. This is very important for women who are pregnant or are trying to become pregnant.



### Absorption

**Gastric bypass** is a surgical procedure that is used to help extremely overweight people lose weight. After a gastric bypass, the body's ability to absorb vitamins (including folic acid) and other nutrients is reduced. There is evidence that women who have had a gastric bypass are at increased risk of having a child with spina bifida.

Other conditions, such as **celiac disease and inflammatory bowel diseases (Crohn's disease, ulcerative colitis)** also reduce the body's ability to absorb vitamins and other nutrients. Whether or not these conditions increase a woman's risk of having a child with spina bifida has not been adequately studied.

Women who have a condition that may interfere with their ability to absorb vitamins and other nutrients should consult with their physician or other health care provider prior to becoming pregnant. Such women may have special nutritional requirements during pregnancy.

### Genes

Because the risk of having a child with spina bifida is higher for individuals who have had an affected child, or who have an affected relative, it is thought that the risk of having a child with spina bifida is partly determined by genes (inherited factors). The Winter 2000 SBRR newsletter covered the topic of genes in detail. If you would like a copy of this newsletter please visit our website ([www.sbr.info](http://www.sbr.info)) or call our toll free number.

*The SBRR scientists are working to find the factors that increase the risk of spina bifida. Through your participation in the SBRR you are helping to answer the question Why did this happen? .*

If you would like more information on any of the topics covered in this newsletter, please contact the SBRR staff, or visit the websites listed below.

**Diabetes**

<http://www.modimes.org/HealthLibrary/>

click on "Quick Reference and Fact Sheets" and choose "Diabetes in Pregnancy"

**Epilepsy**

[http://www.ninds.nih.gov/health\\_and\\_medical/pubs/seizures\\_and\\_epilepsy\\_htr.htm#Pregnancy](http://www.ninds.nih.gov/health_and_medical/pubs/seizures_and_epilepsy_htr.htm#Pregnancy)

**Hyperthermia**

[www.otispregnancy.org](http://www.otispregnancy.org)

click on Fact Sheets and choose Hyperthermia

**Body Mass Index (BMI)**

<http://www.cdc.gov/nccdphp/dnpa/bmi/bmi-adult.htm>

Copies of previous SBRR newsletters can be obtained at our website ([www.sbrr.info](http://www.sbrr.info)) or by contacting Katy Hoess at: [khoess@cceb.med.upenn.edu](mailto:khoess@cceb.med.upenn.edu) or 1-866-275-SBRR

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**References**

Hernandez-Dias S, Werler MM, Walker AM, Mitchell AA. Neural tube defects in relation to use of folic acid antagonists during pregnancy. *American Journal of Epidemiology* 2001;153:961-968.

Sheffield JS, Butler-Koster EL, Casey BM, McIntire DD, Leveno KJ. Maternal diabetes mellitus and infant malformations. *Obstetrics and Gynecology* 2002;100:925-930.



## Go Texans!

Dr. Laura Mitchell has moved to Houston, Texas to begin a new position at the Texas A&M University System Health Science Center. This move has not affected Dr. Mitchell's research interests, and she remains the Principal Investigator for the SBRR.

Sarah Guerra has joined the SBRR team as our new project coordinator. Ms. Guerra, who is located in Houston, is bilingual (English/Spanish) and will be responsible for expanding the SBRR to include families from Texas and surrounding states. Sarah can be reached (toll-free) at **1-866-521-SBTX**, or by email at: [sguerra@ibt.tamu.edu](mailto:sguerra@ibt.tamu.edu)

Katy Hoess, MS, the SBRR Genetic Counselor, continues to work at The University of Pennsylvania, and can be reached (toll-free) at **1-866-275-SBRR**, or by email at: [khoess@cceb.med.upenn.edu](mailto:khoess@cceb.med.upenn.edu)

### Reminder:

Katy Hoess no longer attends the weekly spina bifida clinic at A.I. duPont Hospital for Children. She can be reached at **1-866-275-SBRR** to schedule an in-person or telephone appointment.

### Thank You!

We appreciate the recent involvement of the SBAA, the SBA of Nassau County, Albany, Greater PA, Iowa, and Minnesota, the Spina Bifida Global Village, and the spina bifida on-line support groups.

### BLOOD

We are able to get DNA from cheek swabs, but we get more DNA from blood samples.

If you would like to provide a blood sample, please call Katy Hoess at **1-866-275-SBRR**.

### Visit Us in San Antonio

The SBRR is excited to be a part of the SBAA 30<sup>th</sup>  
Annual Conference

San Antonio, Texas  
June 22 — 25, 2003

Please visit our booth in the exhibit hall.

### Mail in Your Cheek Swabs!!

If you received a cheek swab kit in the mail and you wish to provide a sample, please follow the instructions included in the kit and return your swabs as soon as possible - before the summer heat.