

## WHAT ARE NEURAL TUBE DEFECTS?

Two major types of neural tube defects, spina bifida and anencephaly, are birth defects that affect thousands of babies every year in the United States.

- Neural tube defects (NTDs) are birth defects that occur when the embryonic neural tube, which ultimately forms the spinal cord and the brain, does not close properly.

**Anencephaly** — the absence of the brain — is a fatal condition that occurs when the upper end of the neural tube fails to close. Anencephaly often causes miscarriage; infants who are live births usually die in the early weeks of life.

**Spina bifida** is the incomplete development of the back and spinal cord. Spina bifida occurs when the lower end of the neural tube fails to close. This condition is disabling because of the paralysis, lack of bowel and bladder control, and hydrocephalus that frequently occur with it.

## WHAT CAUSES NEURAL TUBE DEFECTS?

NTDs are a result of a combination of factors, not all of which have been identified.

- The causes of NTDs are not completely known, but do include genetic and environmental factors. NTDs are sometimes associated with chromosome abnormalities, single gene disorders such as Meckel syndrome, and maternal risk factors such as hyperthermia or diabetes, amniotic bands, or prenatal exposure to coumadin, valproic acid, or carbamazepine.
- Since 1964 it has been suspected that folic acid deficiency contributes to the occurrence of neural tube defects. Randomized control trials and observational studies have been conducted which show that an adequate intake of folic acid can reduce the incidence of neural tube defects by approximately one-half.<sup>1</sup>
- Women who are obese (have a Body Mass Index [BMI] greater than or equal to 29 kg/m<sup>2</sup>, or a body weight of more than 80 kg /176 pounds) at the beginning of the pregnancy are at a higher risk of having a baby with a neural tube defect.<sup>2</sup> This higher risk is not reduced by adequate or supplemental folic acid intake.

### WHEN DO

NTDs occur in the first 3-4 weeks of pregnancy, before most women know they are pregnant.

- Normally, during fetal development, the neural tube closes by 28 days after conception and before most women know they are pregnant. Therefore, women cannot wait until after they learn of their pregnancy to take action to prevent neural tube defects.

## WHAT IS THE RATE OF NEURAL TUBE DEFECTS?

Approximately 2,500 babies, a rate of **1 out of every 1,000** infants born in the United States each year has a neural tube defect. This figure does not account for pregnancies miscarried or those pregnancies which are electively terminated after prenatal diagnosis.<sup>3</sup>

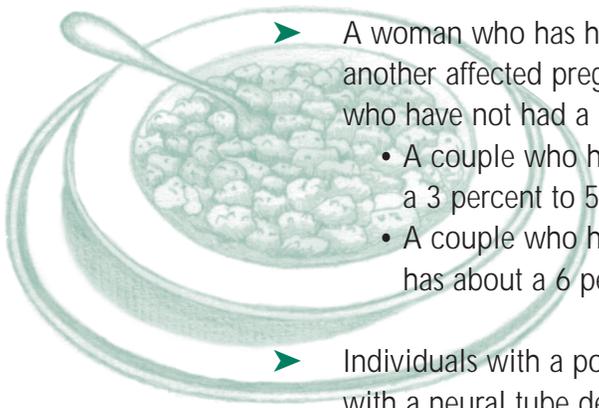
Rates of neural tube defects vary by race and ethnicity. Hispanic whites have higher NTD rates than non-Hispanic whites, and African-Americans and Asians have lower rates than non-Hispanic whites. Births affected with NTDs are more frequent in people with Irish, Scottish, or Welsh background, Sikhs and Egyptians.

- The rate of NTD-affected births in Ohio is about equal to the national average.
- There are approximately **2.5 million women of childbearing age in Ohio**.<sup>4</sup>

## WHO IS AT RISK FOR HAVING A BABY WITH A NEURAL TUBE DEFECT?

Many factors are associated with NTDs, including the following:

- Women with insulin-dependent diabetes mellitus and women with seizure disorders treated with valproic acid or carbamazepine are at increased risk for an NTD-affected pregnancy. The increased risk for either condition is approximately **1 percent**<sup>5</sup>, or 10 times the risk of the general population.
- A woman who has had an NTD-affected pregnancy is at an increased risk to have another affected pregnancy. However, more than 95 percent of NTDs occur in women who have not had a prior affected pregnancy.
  - A couple who has had one pregnancy affected with a neural tube defect has about a 3 percent to 5 percent recurrence risk in subsequent pregnancies.<sup>5</sup>
  - A couple who has had two or more pregnancies affected with a neural tube defect has about a 6 percent to 9 percent recurrence risk.<sup>5</sup>
- Individuals with a positive family history have the following risk for having a child with a neural tube defect:<sup>5</sup>
  - If an individual has spina bifida, his or her risk for having a child affected with a neural tube defect is about 3 to 5 percent.
  - Individuals who have a second degree relative (i.e., a niece, nephew, aunt, uncle, or half sibling) with a neural tube defect have about the same risk level as the general population for having a child with this condition.<sup>5</sup>



## WHAT IS THE DIFFERENCE BETWEEN FOLATE AND FOLIC ACID?

Folic acid is a B vitamin that can reduce the incidence of neural tube defects by as much as 50 percent when taken at least 30 days before and during the first trimester of pregnancy.

- Folate is an essential nutrient found in a variety of foods, and is used by the body to manufacture DNA. DNA is required for rapid cell division and organ/tissue formation in the developing fetus. This vitamin is especially important during periods of increased metabolic activity such as infancy, childhood, pregnancy, and breastfeeding.
- The terms folic acid and folate are often used interchangeably, but they are not the same. Folic acid is the name for the synthetic form of folate used in vitamin supplements or to fortify cereals and other grain foods. Dietary folate is a complex form of the vitamin that occurs naturally in a variety of foods. Synthetic folic acid is more readily used by the body.

## WHAT ARE THE IMPORTANT PUBLIC HEALTH ISSUES CONCERNING NTDs?

There are two important public health issues concerning NTDs that must be addressed. The first relates to **reduction**, and the second relates to **treatment**. The goal of reduction of NTDs is to decrease the number of persons affected by the disabling consequences of spina bifida or the fatal consequences of anencephaly. The goal of treatment of NTDs is to improve the quality of life of people who have spina bifida.

Both reduction and treatment of NTDs are important. Many advances have been made in the treatment of spina bifida which have resulted in increased life expectancy and improved quality of life for individuals with spina bifida. Unfortunately, a treatment has not been developed that will completely eliminate the serious disability or premature mortality associated with spina bifida nor the mortality associated with anencephaly. For these reasons, reduction of NTDs is an important goal.

From a public health perspective, the opportunity for the primary prevention of a common congenital disorder exists by disseminating the following message:

- If all women of childbearing age had adequate folic acid intakes before conception and during the first trimester of pregnancy, the rate of NTD-affected pregnancies could be reduced by as much as half.

However, it is important to recognize that not all NTDs can be prevented by adequate intake of folic acid. The important public health message to be communicated in this instance is that:

- For individuals born with spina bifida, specialized medical interventions can increase life expectancy and improve quality of life.



## WHAT ARE THE COSTS ASSOCIATED WITH NTDs?

The economic costs associated with caring for one child with spina bifida have been estimated as follows:

- In 1991 the total lifetime costs of medical and nonmedical services provided to a child with spina bifida...has been calculated at over \$300,000.<sup>6</sup> This figure does not include costs for nonprescription medications, home modifications, transportation, supplies, or loss of income by the primary caregiver.
- The emotional costs to a family that experiences the loss of an infant, or the birth of an infant with a disability are immeasurable. In the fervor to prevent future births affected by spina bifida, it is paramount that these reduction efforts should not diminish the worth, power or dignity of the individuals who are born and live with this condition each year. It is important to remain sensitive to the family who has a child with spina bifida and their guilt associated with thinking there may have been a preventive action that was not taken by the mother.

## WHEN DO WOMEN NEED FOLIC ACID?

*Women need to take folic acid daily. Folic acid particularly important throughout their reproductive years.*

Half of the pregnancies in the United States are unplanned, and, even in the half that are planned, most women do not know they are pregnant until it is too late to prevent folate related neural tube defects in their babies.

Therefore, the U.S. Public Health Service has issued two specific recommendations:

### RECOMMENDATION #1: FOR WOMEN WITH A PRIOR NTD-AFFECTED PREGNANCY

The recommendation for women who have had a prior pregnancy affected by a neural tube defect and who are planning to start a new pregnancy is a consumption of 4.0 mg daily dose of folic acid from at least one month prior to conception through the first three months of pregnancy. This dosage cannot reasonably be achieved through diet. It requires supplementation. Doses of this level may be associated with other health risks and should be taken only under the supervision of a physician.<sup>7</sup>

Because the effects of high intakes are not well known but include complicating the diagnosis of vitamin B12 deficiency, care should be taken to keep total folic acid consumption at less than 1.0 mg per day, except under the supervision of a physician.<sup>1</sup>

### RECOMMENDATION #2: FOR WOMEN OF CHILDBEARING AGE

All women who are capable of becoming pregnant should consume 0.4 mg (400 mcg) of folic acid per day to reduce their risk of having a pregnancy affected with spina bifida or other neural tube defects.

