



Advances in the Care of Children with Spina Bifida

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Keywords

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EPIDEMIOLOGY AND RISK FACTORS

Key points

- The risk of a mother with 1 neural tube defect (NTD)-affected pregnancy having another NTD-affected pregnancy triples.
- Adequate consumption of folic acid preconceptionally can prevent 50% to 70% of NTDs.
- Ultrasonography is a reliable method to identify NTDs by the end of the first trimester of gestation.

Introduction

Neural tube defects (NTDs) are a multifactorial disorder that results from a complex combination of genetic and environmental interactions. The worldwide incidence of NTDs ranges from 1.0 to 10.0 per 1000 births in specific geographic locations, with almost equal frequencies between 2 major categories: anencephaly and spina bifida [1,2]. The birth prevalence of NTDs in specific populations is influenced by the availability of prenatal diagnosis and elective pregnancy termination; higher frequencies occur in miscarriage material [3]. Each year, 300,000 to 400,000 infants worldwide are born with 1 of these 2 forms of NTDs.

Risk factors

Current identified risk factors for NTDs include a mother who previously had an NTD-affected pregnancy, maternal pregestational insulin-dependent diabetes, maternal pregestational obesity, hyperthermia, other environmental exposures, specific anticonvulsant drugs, including valproic acid, genetic variants, race/ethnicity, and nutrition (particularly folic acid [FA] insufficiency) [4]. The teratogenic potential of maternal pregestational diabetes is well established and includes a 2-fold to 10-fold increase in the risk of central nervous system malformations (including NTDs) among the offspring of affected women, relative to the general population. Women in the highest body mass index (BMI, calculated as weight in kilograms divided by the square of height in meters) categories (usually defined as a prepregnancy body mass index $>29 \text{ kg/m}^2$) have a 1.5-fold to 3.5-fold higher risk than women with lower indices. There is evidence that maternal hyperthermia increases the risk of having a child with an NTD by up to 2-fold. The fungal product fumonisin caused a doubling of NTD incidence along the Texas-Mexico border in the early 1990s. Among women taking valproic acid or carbamazepine, the risk of having a pregnancy affected with spina bifida may be as high as 1% to 2%. In a few cases, anencephaly and spina bifida occur as part of malformation syndromes that result from teratogenic exposures [5]. Of particular clinical significance is valproic acid, an anticonvulsant that increases the risk of spinal NTDs by roughly 10 times when taken early in pregnancy. Although the teratogenic mechanisms are hypothesized to involve antifolate effects, particularly for

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