Condition: Neural tube defect (includes anencephaly, myelodysplasia, spina bifida)

Inheritance:

Multifactorial

Genetic etiology:

Unknown

Frequency:

Population-specific.

Clinical features:

The brain and spine form in early embryogenesis by folding of a neural plate into a tube. Incomplete closure results in a malformation in the developing neural tube. If the anomaly is anterior, anencephaly or encephalocele results. Posterior defects result in meningocele or myelomeningocele. Anencephaly is lethal. Posterior defects can cause loss of neurological function below the level of the lesion, including paralysis of the legs and loss of bowel/bladder function. Infants born with open defects are at risk of infection.

Management:

For non-lethal defects, the lesion is closed by surgery. Supportive care is then provided for neurological deficits and orthopedic problems.

Genetic counseling:

Recurrence risk is based on empirical data. Folic acid intake can prevent approximately 50% of neural tube defects. Prenatal diagnosis is possible by ultrasound and maternal serum alpha fetoprotein testing in the second trimester.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.