PRIMEVIEW

SPINA BIFIDA

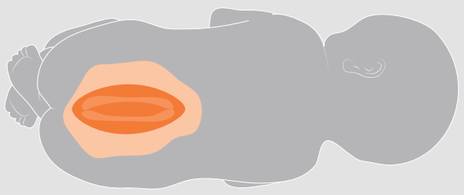
nature REVIEWS DISEASE PRIMERS

For the Primer, visit doi:10.1038/nrdp.2015.7

Spina bifida is a congenital disorder characterized by failed closure (primary neurulation) or formation (secondary neurulation) of the neural tube, which results in abnormalities of the vertebral column and/or spinal cord. Myelomeningocele (also termed open spina bifida) is the most common and disabling form, and is associated with an open spine and neural tissue degeneration.

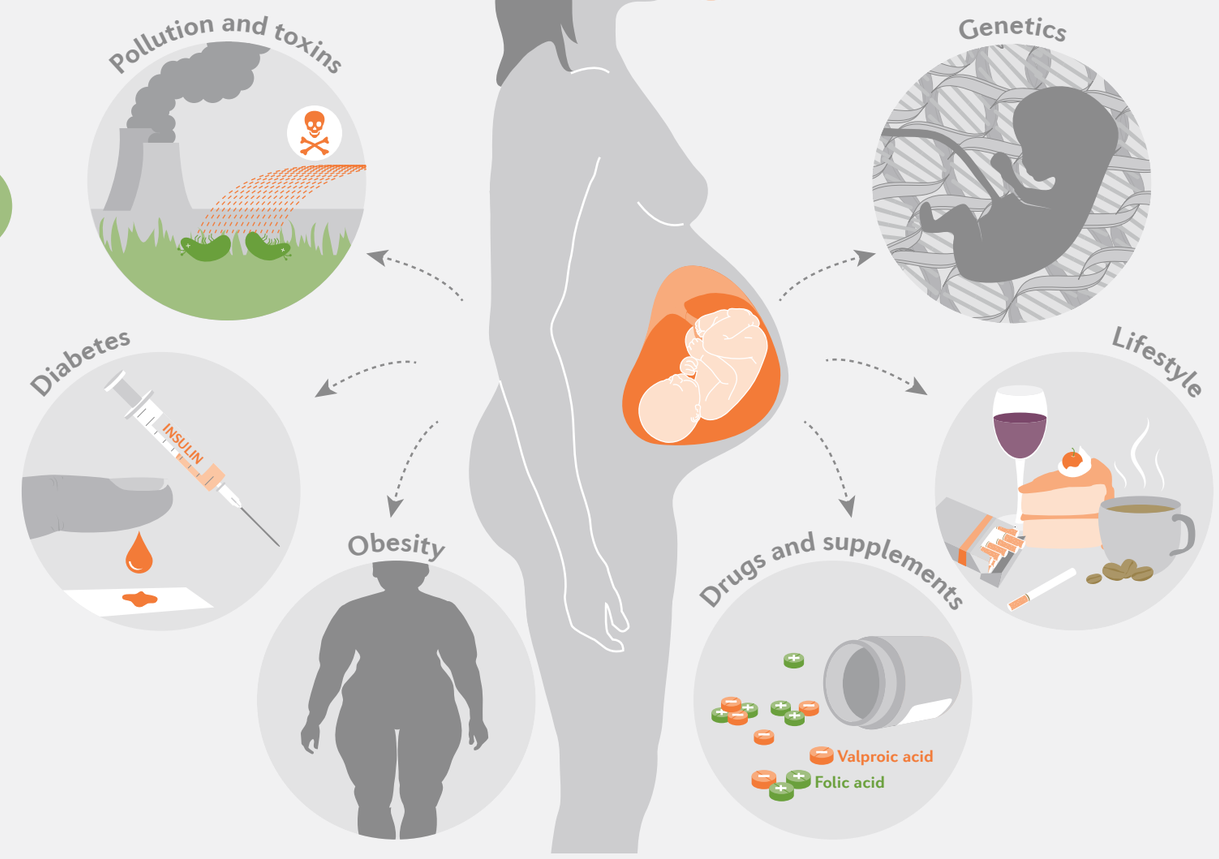
EPIDEMIOLOGY

The prevalence of spina bifida and other neural tube defects is approximately one per 1,000 births in the United States and Europe. Worldwide, this extrapolates to 140,000 neural tube defects per year. The prevalence rates have varied substantially over time and between geographical locations, a phenomenon that is largely attributed to variations in preventive actions, including folate supplementation. In addition, ethnic and environmental factors have a role.



MECHANISMS

Genetic predisposition is estimated to contribute 60–70% to disease development. Recurrence risk for siblings is increased up to 50-fold; after an affected pregnancy, the risk of spina bifida rises by 3–10%.



QUALITY OF LIFE

Myelomeningocele often results in neurological deficits below the level of the lesion, typically affecting motor and sensory function. A major complication is lower-limb weakness that hampers or prevents walking.The pathology is frequently accompanied by brain abnormalities such as the Chiari II malformation and hydrocephalus, which can lead to neurocognitive anomalies. The psychosocial impact on patients includes low self-esteem, lack of autonomy and depressive symptoms.

Survival rate depends on the location of the spinal lesion: 20% of patients with lesions above T11 survive to the age of 40 years, whereas this is 60% for lesions below L3

OUTLOOK

Spina bifida affects not only the patients, but also their families and society. A number of factors that could eventually improve outcomes for patients remain under investigation. First, mechanistic insight into the preventive action of folic acid is currently lacking but is essential if we are to understand and bypass cases of toxicity and resistance. Second, the use of stem cells during *in utero* surgery might help to halt progression or even restore lost function, but the translation to clinical practice is still a long way off. Finally, patients will benefit from the development of standardized psychosocial interventions, such as goal management training.

Lifetime costs are estimated at €500,000, more than 35% of which is directly related to medical interventions

PREVENTION

Folic acid supplementation is the cornerstone of spina bifida prevention. Concerns over a lack of adherence to voluntary supplementation recommendations and the need to supplement before pregnancy led to food fortification in many countries, but not in Europe. Together, these actions have reduced the occurrence of spina bifida substantially.

DIAGNOSIS

Prenatal diagnosis depends on measurements of α-fetoprotein in the maternal blood and ultrasonographic evaluation of the shape of the fetal skull and spine.

MANAGEMENT

Following diagnosis, three options are available: termination of the pregnancy or repair (either postnatally or *in utero*). Prenatal surgery has considerably improved the outcomes of patients with spina bifida, reducing the occurrence of hydrocephalus and improving neuromotor function. Premature delivery remains a drawback.

Designed by Laura Marshall

Article number: 15051; doi:10.1038/nrdp.2015.51; published online 30 April 2015