Best Practice & Research Clinical Obstetrics and Gynaecology 28 (2014) 367–377

Contents lists available at ScienceDirect

Best Practice & Research Clinical Obstetrics and Gynaecology

journal homepage: www.elsevier.com/locate/bpobgyn

3

Diagnosis of spina bifida on ultrasound: Always termination?

Amanda S. Trudell, DO, Maternal Fetal Medicine Fellow, Department of Obstetrics and Gynecology Washington University School of Medicine *, Anthony O. Odibo, MD, MSCE, Associate Professor, Department of Obstetrics and Gynecology, Director of Ultrasound Services, Division of Maternal-Fetal Medicine and Ultrasound

Obstetrics and Gynecology, Washington University in St. Louis, 4911 Barnes Jewish Hospital Plaza, Saint Louis, MO 63117, USA

Keywords:
open spina bifida
open neural tube defect
myelomeningocele
ultrasound
termination of pregnancy

Open spina bifida is a non-lethal fetal anomaly. Significant advances in the prevention, diagnosis and treatment of open spina bifida have been made over the past 75 years. The most significant strategy for the prevention of open spina bifida has been with folic acid supplementation; however, further investigation into the complicated role that genetics and the environment play in metabolism are coming to light. Ultrasound is the gold standard diagnostic tool for spina bifida. Three-dimensional ultrasound and magnetic resonance imaging are also beginning to play a role in the characterisation of the open spina bifida spinal lesion. Lesion level has been closely correlated to short and long-term outcomes, and prenatal characterisation of lesion level on ultrasound is important for patient counselling. Long-term outcomes of people living with spina bifida are available and should be used for non-directive patient counselling about pregnancy choices for women with open spina bifida.

Introduction and historical perspectives

Open spina bifida (myelomeningocele) is a specific type of neural tube defect (NTD) resulting from a failure of closure of the caudal region of the neural tube early in embryogenesis distinct from

* Corresponding author. Tel.: +1 314 362 895; Fax: +1 314 747 1720.
E-mail address: trudella@wudosis.wustl.edu (A.S. Trudell).

1521-6934/$ – see front matter
http://dx.doi.org/10.1016/j.bpobgyn.2013.10.006

anencephaly, which results when the cranial portion of the neural tube fails to close. The diagnosis and management of open spina bifida has evolved dramatically over the past century. The development of shunts for the treatment of hydrocephalus in the 1950s was a revolutionary breakthrough. Improved survival meant that children were living long enough to develop urinary complications; hence, progress was made in the 1960s with the introduction of urinary diversion procedures. Before the 1960s, the favoured approach for postnatal treatment was postponement of surgery until 2 years of life, with the belief that the strongest would survive and would therefore be the best candidates for surgical intervention [1]. With the advancements of the 1950s and 1960s, this Darwinian approach came to be challenged as evidence of benefit of immediate neonatal intervention within 12–48 h of life. [2] The development of Lorber's criteria in the 1970s for triaging treatment generated a great ethical debate of triage versus immediate intervention for all and selective intervention [3]. The 1980s and 1990s saw improvements in shunt technology and management, clean intermittent catheterisation protocols to reduce renal complications, and the development of spina bifida clinics to manage the medical, physical, and social needs of children, families, and adults with spina bifida. Over the past 25 years, improvements in prenatal diagnosis and advancements have been made in fetal therapy, with the publication of a randomised-controlled trial demonstrating benefit of fetal surgical repair of myelomeningocele [4].

Although progress in the field of myelomeningocele diagnosis and treatment has revolutionised the medical treatment of open spina bifida, the ethical debate surrounding the postnatal treatment of myelomeningocele that emerged in the 1970s has evolved significantly and is now complicated by issues surrounding prenatal diagnosis, including availability, economic feasibility, and selection for invasive fetal surgery and termination of pregnancy.

Epidemiology

The prevalence of spina bifida is known to have geographic, temporal, race, and ethnic variation [5,6]. An estimated 3.5 cases per 10,000 live births occurred in the USA between 2004 and 2006, so the actual prevalence of myelomeningocele has been difficult to ascertain because of the availability of prenatal diagnosis and elective termination of pregnancy [7]. Johnson et al. [8] recently conducted a meta-analysis of prenatally diagnosed cases of anencephaly and spina bifida. It included retrospective or prospective cohort studies and studies using birth defects registry data conducted during or after 1990. Significant heterogeneity was found between studies [8]. The investigators identified 15 studies limited to spina bifida and estimated the overall rate of termination of pregnancy to be 63%, with an individual study range of 31–97% [8]. Rates of termination varied based on geographic location with termination of pregnancy being more frequent in Europe than the USA (66% *v* 50%). Five of the studies provided information on isolated myelomeningocele and reported a 56% termination of pregnancy rate for isolated spina bifida [8].

Risk factors

Several risk factors have been studied in association with spina bifida; however, only 28% of cases of spina bifida have attributable risk factors, and most of these associations are weak and have not been replicated in subsequent studies [9]. The strongest established risk factor for an NTD is a family history. Several studies have estimated that women with a history of a child with an NTD carry a 3–8% risk of and NTD, including spina bifida, in a subsequent pregnancy, a risk that is consistent with most multifactorial inherited conditions [5].

Folic acid and genes related to folate metabolism have been identified to play a strong role in neural tube development. Folic acid deficiency has been associated with a two- to eight-fold increased risk of NTDs [5]. Although women with pre-gestational diabetes have been traditionally considered to be at risk for diabetic embryopathy, including an NTD, a report by Correa et al. [10] using the National Birth Defects Prevention study, a population-based cohort study of 10 birth defects surveillance programmes in the USA, found no association between type I or type II diabetes with increased risk of isolated spina bifida [10]. The investigators showed the rarity of isolated congenital defects in women with pre-existing diabetes, and the study was likely underpowered to detect an association between pre-