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Review

Health-related quality of life is compromised in individuals with spina bifida: results from qualitative and quantitative studies

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ABSTRACT

Spina bifida (SB) accounts for approximately 90% of total neural tube defects. According to the degree of SB severity, the range of associated symptoms and complications varies greatly. Given the complexity and diversity of these complications, individuals with SB might suffer from livelong impairment. This review presents an overview of the impact of SB on patients' health-related quality of life (HRQoL) and explores results from published quantitative and qualitative studies regarding the HRQoL impact of SB on patients, as well as comparing results of existing studies to national norms.

A literature search using three electronic databases PUBMED, PsycINFO, and Embase was performed to identify relevant studies dating from January 1976 to November 2010. To satisfy the initial inclusion criteria, articles had to contain studies that were specific to HRQoL in patients with SB.

Findings highlight that HRQoL is significantly impaired in patients with SB. Making sure that the public is aware of the disease and how to minimise the risk of NTD, such as SB (e.g., through adequate folate levels at time of conception, etc.) is essential to ensure that fewer individuals face the burden of NTDs in the future.

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Introduction

Neural tube defects (NTDs) are the second most common birth defect after cardiac abnormalities [1–3]. They are caused when the embryonic neural tube does not close properly, usually during the first six weeks of embryonic development, and often before a woman knows she is pregnant [3–8].

There are various types of NTDs including anencephaly (where babies are either stillborn or die shortly after they are delivered) and spina bifida (SB). Children with SB have a higher probability of lifelong physical and mental handicap compared to those with no chronic conditions and only a minority of these children are able to go on to function independently as adults [9].

In the US one in every 1000 newborns (or about 8000 babies a year) are still born with SB and overall prevalence rates are similar in the EU [10]. Total prevalence rate across Europe is estimated at 9.8 per 10,000 births, the Ukraine has the highest rate of NTDs with 20.5 per 10,000, with Croatia and Italy reported to have lower rates around 4.6 NTD births per l0,000 [11]. With advances in treatment life expectancy of SB patients is rising, with 85% of children born with SB surviving into adulthood [12,13].

There are three forms of SB, with the least severe of the three being SB occulta; characterised by a small defect or gap in one or more of the vertebrae of the spine. The spinal cord and nerves are usually normal as the spinal cord does not protrude through a gap and is covered with skin; most affected individuals are asymptomatic and have no problems. As such, SB occulta is not typically considered to be a true form of SB and is often referred to as incomplete posterior fusion. In SB's rarest form, Meningocele, the vertebrae develop normally but the protective membranes (i.e. the meninges) surrounding the spinal cord are forced into the gaps between the vertebrae and form a meningeal cyst. The cyst can vary in size, and can be removed by surgery, allowing for normal development. The most serious form of SB is called myelomeningocele. When SB is open the spinal cord as well as the meninges and fatty tissues may protrude through a gap in the spine. This is what is known as a myelomeningocele. Myelomeningoceles are often intact, with a meningeal sac enclosing spinal fluid [14,15]. However, in many cases the sac is disrupted causing spinal fluid to leak out. When this occurs, babies are at high risk of infection until their back is closed surgically, although antibiotic treatment may offer temporary protection [16].

Many factors can cause a birth defect. Through multifactorial inheritance these factors are usually a combination of genetics from both parents and environmental factors such as diabetes or certain prescribed medications. In addition, spina bifida can sometimes occur more frequently as part of a syndrome or chromosomal disorder along with other birth defects [17]. Folate is seen as a key preventative measure for those with and without a multifactorial inheritance pre-disposition and has the potential to reduce the frequency of other possible syndromes. Indeed, the US Food and Drug Association (FDA) Advisory Committee for Reproductive Health Drugs now recommend that women (pre-pregnancy and early pregnancy) should increase their intake of folic acid to 400 mcg a day [18]. In addition, there are screening procedures (such as ultra sound and Maternal Serum Alpha-Fetoprotein (MSAFP)) that aid in the diagnosis of SB while the child is still in utero. While we recognise the significant impact a positive diagnosis may have on a parent, as they either wait for further results or make a decision about whether or not to terminate, it is the purpose of this study to focus on the HRQoL of SB after the child is born.

Dependent on the lesion size, location, surgical intervention, and co-morbidities, the SB symptoms, severity levels and HRQoL outcomes can be unpredictable. Individuals' lower limbs may be paralysed and they may experience lack of sensation. They may be unable to control bowel and bladder functions. Some cases exhibit Chiari II malformation, a condition in which the lower part of the brain is crowded and may be forced into the upper part of the spinal cavity. Hydrocephalus can result from this malformation as the displaced cerebellum interferes with the flow of cerebrospinal fluid, causing an excessive accumulation of cerebrospinal fluid in the brain. Further, although most individuals have a normal level of intellectual functioning, some have learning disabilities or mental retardation [19]. Executive deficits are also common in individuals with SB [20].

Given the complexity and diversity of lifelong complications, individuals with SB must be closely monitored and often receive wide-ranging and time-consuming treatments. Initial surgery often occurs in an infant's first days and subsequent surgeries may be carried out thereafter. In hydrocephaly, a mechanical shunt is installed to decrease the pressure and amount of cerebrospinal fluid in the cavities/ventricles of the brain. Most children with SB require physical therapy, bracing, and other orthopaedic assistance in order to walk. Further, a variety of approaches, including periodic bladder catheterization, surgical diversion of urine, and antibiotics are used to protect urinary function. An early start to renal care may help protect the kidneys and promote social continence [21]. Also, patients with SB commonly present with bowel incontinence which is typically managed by diet and mild laxatives combined with enemas or digital manoeuvres, with surgery required in patients who do not respond to said measures. Educational intervention, physical therapy, and occupational therapy may be useful for those with learning disabilities. Counselling can help with self-image and lessen barriers to socialisation [22].

Several studies have explored individuals' health-related quality of life (HRQoL) in SB [9,13,19,23–28], but the extent of individuals' experiences with regard to HRQoL requires further exploration. Often, studies focus either on qualitative (therefore overlooking the statistical and clinically meaningful impact of SB on patients compared to the general population) or quantitative aspects (missing the detailed descriptions that can be achieved by closely examining what patients actually say about their experiences). In addition, to our knowledge, no studies have attempted to synthesise the data available in the literature to demonstrate the overall impact of SB on patients' HRQoL.

Assessing the psychosocial factors in SB (for example, the impact it has on school, socialising and self-image) in addition to clinical and physiological information, provides a more complete picture of the burden people with SB are facing in their daily life. The prevalence of SB in the US has seen a large decrease following a combination of health education programmes, recommendations and mandatory food fortification with folic acid [29]. However, despite raised awareness that increased and regular folic acid consumption around the time of conception can help prevent the occurrence of folate dependent SB, declines in prevalence have not been as dramatic as expected with the vast majority of European women still not taking folic acid supplements peri-conceptionally, thus NTD incidence remains unnecessarily high [30,31]. The burden caused by NTDs to individuals, families and carers is incalculable, while direct costs of managing NTDs currently run to many thousands of Euros per patient per year [32]. It is therefore imperative that this burden is fully understood, in order to comprehend the level of support that individuals with SB may require from society and to help overcome the daily challenges they face.

This review explores results from published quantitative and qualitative studies regarding the HRQoL impact of SB on patients, as well as comparing results of existing studies to national norms.

Methods

This review was a sub-study of a larger study investigating the burden of NTDs (especially SB) and the associated level of unmet needs.