Abstract. As the diagnosis of Spina Bifida (SB) is often made prenatally, SB-specific prenatal counseling is needed. It is essential to provide information about medical care and lifelong impact of this diagnosis, treatment options available to women carrying fetuses affected, and resources that will assist in the care of individuals with SB. This article outlines the SB Prenatal Counseling Guidelines from the 2018 Spina Bifida Association’s Fourth Edition of the Guidelines for the Care of People with Spina Bifida and acknowledges that further research in SB prenatal counseling is warranted.

Keywords: Spina bifida, myelomeningocele, fetal therapies, prenatal care, counseling, neural tube defects

1. Introduction

The prenatal diagnosis of Spina Bifida (SB), or a neural tube defect (NTD), is a pivotal moment for families. Information is shared that will forever alter the family, regardless of decisions made. As such, it is essential that information shared in prenatal counseling about NTDs be timely, evidence-based and objective.

Critical to counselling is clarity regarding the type of NTD that has been identified as they can be open or closed. Open refers to NTDs with neural elements directly exposed, whereas closed NTDs have skin covering the defect and as a result do not pose an immediate risk to the infant’s wellbeing. Identification of the type of finding determines important immediate counseling considerations, particularly as a child with open NTDs may be eligible for closure in utero which requires expedient referrals to centers that offer this opportunity. In addition, the anticipated medical, surgical, and developmental course varies with the type of NTD [1,2].

The manner in which information about NTDs is conveyed is critical to the family’s future and provides the basis for important family decisions [3,4]. Families may experience intense emotions such as uncertainty, confusion, grief, anxiety and anger. At the same time, families need to absorb a great deal of information about their options and to understand the risks and benefits of those options. Differing cultures have disparate perceptions of disability, and this may impact
families’ expectations about treatment options and/or their understanding of the condition [5]. There is a time constraint as well, with one of the potential options, fetal surgery, available only for a short window [2,6]. Therefore, it is essential amidst this background of intense emotions, time pressure, and cultural disparity that parents and caregivers have timely access to information. The information provided should detail the anticipated clinical course across a lifespan as well as the strengths and challenges associated with SB [2]. Ideally, a team of providers with experience providing care to individuals with NTDs should be involved. This information should be individualized and provided in a neutral and collaborative manner that meets the needs, values, and beliefs of each family [5,7].

2. Guideline goals and outcomes

The goals of this prenatal counseling guideline are both practical and aspirational.

Primary outcome

- Convey information about medical care and lifelong impact of SB in a value-neutral, collaborative manner while seeking an understanding of families’ needs, values and beliefs.

Secondary outcome

- Provide education regarding all treatment options available to women carrying fetuses affected by SB, including fetal surgery, postnatal closure, adoption, and termination.

Tertiary outcome

- Provide families with information about providers and resources that will assist in the care of their child with SB.

3. Methods

The methodology for writing these guidelines has been published by Dicianno et al. [8]. The process included collaboration of numerous SB experts from around the world, as well as input from those with SB and reviewed readings regarding the perception of prenatal testing amongst the disability community. This topic of prenatal counseling was a new subject area since the last and 3rd version of Spina Bifida Guidelines, Guidelines for Spina Bifida Health Care Services Throughout the Lifespan, 2006 [9].

3.1. Clinical questions framing the guidelines

These guidelines were framed by the following clinical questions:

1. What are the essential components of prenatal consultation for any child affected by a potentially disabling condition?
2. What are the essential treatment options that are to be discussed as part of the prenatal consultation for a family with a fetus affected by an NTD?
3. Are there critical language elements to be considered?
4. Are there critical and specific medical providers that should participate in the prenatal consultation for a family affected by a fetus with an NTD?

4. Results

The guidelines are presented in Table 1. The first column is the recommendation, and the second column is the evidence supporting that recommendation.

These guidelines were drawn from evidence around SB and its impact on lifelong functional outcomes as well as antenatal counselling for disabling conditions. We recommend that all evidence-based treatment options be offered, including antenatal or fetal closure, closure after birth, termination of the pregnancy, and adoption [2,4,10–14]. In order for all of these options to be available, timely consultation should occur as soon as possible following the antenatal identification, between 18 and 20 weeks gestation [12,13]. The fetal closure is an option until the end of the 25th week gestation and should be offered at treatment centers with expertise in the surgical and obstetrical management of NTDs [11,12,14]. The surgery itself and the obstetrical implications should be reviewed by those surgeons and obstetricians with experience managing high risk pregnancies and/or providing care to infants with NTDs [12]. In addition, the option of delivery at term and closure following birth should be offered for those with open NTDs, explaining that cesarean delivery at 37 weeks gestation and closure within 24 hours of delivery is to be expected [15]. Parents or caregivers should be informed of the details of the surgery and the postoperative course, as well as the different likelihoods for specific maternal and fetal outcomes, particularly related to hydrocephalus in the infant, between the fetal and postnatal closures [13,15]. For those parents not able to raise a child with medical complexity but not comfortable with termination, the option of adoption
Table 1
Clinical questions that informed the family functioning guidelines

| Guideline                                                                 | Guideline details                                                                                                                                                                                                 | Evidence                  |
|---------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------|
| Convey information about the medical care and lifelong functional impact  | Ideally, consultations with the parents will take place shortly after identification of the NTD is made at the 18th week and before the 24th week of gestation.                             | [12, 13]                  |
| of SB in a value-neutral, collaborative manner while seeking from families an | Efficient consultation is essential and should happen quickly, soon after the identification of the NTD, to allow parents the broadest array of options and to provide enough time to consider the option for fetal surgery.      | [11–14]                  |
| needs, values and beliefs                                                 | Base consultation with families on a collaborative, shared decision-making model that includes the medical team and parents.                                                                                       | [5, 7]                    |
|                                                                           | Avoid using words that assign value or bias, such as “risk,” “bad news,” and “bad outcomes”. Use words that impart the importance of the decision, such as “important news,” “significant outcome,” and “potential challenges.” This allows parents to assign their own values to the news provided. | [5, 7]                    |
|                                                                           | Offer a review of prenatal testing and results to clarify any misunderstandings or confusion that may exist. Typically the diagnosis is made by a high resolution ultrasound examination that is performed during the second trimester at a maternal-fetal medicine unit. Ultrasound can define the location and size of the lesion, whether it is open or closed (in most instances), and secondary findings such as hydrocephalus. Given the increased risk of other abnormalities, fetal echocardiography should be considered. Genetic evaluation by amniocentesis for chromosomal microarray should be recommended because the identification of a genetic abnormality in a fetus with an NTD has important implications for counseling regarding pregnancy management, and determining whether the patient is a candidate for in utero NTD repair. Measurement of amniotic fluid acetylcholinesterase helps to differentiate between open and closed NTDs and is a component of many preoperative evaluations for fetal repair. Fetal MRI also may be considered for assessment of unclear findings on ultrasonography. | [3, 13, 16–19]           |
|                                                                           | Expect to provide critical information about the likelihood of survival and the spectrum of outcomes (i.e., neurosurgical, cognitive, developmental, urologic, orthopedic, and dermatologic conditions) for children with NTDs. | [1, 2, 4, 12]            |
|                                                                           | Discuss disability. Provide information on outcomes with a lifespan approach.                                                                                                                                     | [7]                       |
|                                                                           | Review general principles associated with lesion levels, as well as the difficulty with providing specific predictions based on lesion level.                                                                       | [15]                      |
|                                                                           | Review treatment options for conditions associated with NTDs with an emphasis on functional outcomes.                                                                                                               | [20]                      |
| Review evidence-based treatment options with the family, including fetal  | Treatment options should include prenatal closure for open NTDs offered at treatment centers with expertise in the surgical and obstetrical management of NTD. Fetal surgery and the details of the surgical and obstetrical impacts should be reviewed by surgeons/obstetricians with experience managing high-risk pregnancies and/or providing care to infants with NTDs. | [10–12]                  |
| surgery                                                                   | Also present the option for term delivery and postnatal closure for open NTDs, explaining to parents and caregivers that cesarean delivery at 37 weeks and closure within 24 hours of delivery is generally recommended when the decision is made for postnatal closure. Ensure that the parents are aware of what to expect at birth and after the surgery. Closed NTDs usually do not require surgical intervention in the newborn period but should have the same monitoring and investigations in the newborn period. Present adoption as an option for parents who are not open to termination but are not able to raise a child with a disability. Review termination of the pregnancy as another option for the family. | [15] Clinical consensus    |
|                                                                           | Specialists in fetal medicine/obstetrics familiar with management of a pregnancy complicated by a prenatal diagnosis of NTD are key to review management options and initiate referrals for additional evaluation. Neurosurgeons offer insight into the details of surgical options as well as perspective on lifelong functional and surgical outcomes. Urologists can assist with social continence which is a key functional outcome. Clinical genetics can provide clarity around genetic testing and recurrence risks. Orthopedic surgeons assist with the monitoring of skeletal growth and mobility. Developmental pediatricians, physiatry, and advanced practice nurses who focus on developmental disabilities can offer functional programming to optimize function, resource identification, advocacy, and access to care. | [10–12] Clinical consensus |
| Offer families the opportunity to meet with key members of the SB team     |                                                                                                                                                    | [11, 12]                  |

Clinical questions that informed the family functioning guidelines:

- Convey information about the medical care and lifelong functional impact of SB in a value-neutral, collaborative manner while seeking from families an understanding of their needs, values and beliefs.

- Base consultation with families on a collaborative, shared decision-making model that includes the medical team and parents.

- Avoid using words that assign value or bias, such as “risk,” “bad news,” and “bad outcomes.” Use words that impart the importance of the decision, such as “important news,” “significant outcome,” and “potential challenges.” This allows parents to assign their own values to the news provided.

- Offer a review of prenatal testing and results to clarify any misunderstandings or confusion that may exist. Typically the diagnosis is made by a high resolution ultrasound examination that is performed during the second trimester at a maternal-fetal medicine unit. Ultrasound can define the location and size of the lesion, whether it is open or closed (in most instances), and secondary findings such as hydrocephalus. Given the increased risk of other abnormalities, fetal echocardiography should be considered. Genetic evaluation by amniocentesis for chromosomal microarray should be recommended because the identification of a genetic abnormality in a fetus with an NTD has important implications for counseling regarding pregnancy management, and determining whether the patient is a candidate for in utero NTD repair. Measurement of amniotic fluid acetylcholinesterase helps to differentiate between open and closed NTDs and is a component of many preoperative evaluations for fetal repair. Fetal MRI also may be considered for assessment of unclear findings on ultrasonography.

- Expect to provide critical information about the likelihood of survival and the spectrum of outcomes (i.e., neurosurgical, cognitive, developmental, urologic, orthopedic, and dermatologic conditions) for children with NTDs.

- Discuss disability. Provide information on outcomes with a lifespan approach.

- Review general principles associated with lesion levels, as well as the difficulty with providing specific predictions based on lesion level.

- Review treatment options for conditions associated with NTDs with an emphasis on functional outcomes.

- Treatment options should include prenatal closure for open NTDs offered at treatment centers with expertise in the surgical and obstetrical management of NTD. Fetal surgery and the details of the surgical and obstetrical impacts should be reviewed by surgeons/obstetricians with experience managing high-risk pregnancies and/or providing care to infants with NTDs.

- Also present the option for term delivery and postnatal closure for open NTDs, explaining to parents and caregivers that cesarean delivery at 37 weeks and closure within 24 hours of delivery is generally recommended when the decision is made for postnatal closure.

- Ensure that the parents are aware of what to expect at birth and after the surgery.

- Closed NTDs usually do not require surgical intervention in the newborn period but should have the same monitoring and investigations in the newborn period.

- Present adoption as an option for parents who are not open to termination but are not able to raise a child with a disability.

- Review termination of the pregnancy as another option for the family.

- Specialists in fetal medicine/obstetrics familiar with management of a pregnancy complicated by a prenatal diagnosis of NTD are key to review management options and initiate referrals for additional evaluation.

- Neurosurgeons offer insight into the details of surgical options as well as perspective on lifelong functional and surgical outcomes.

- Urologists can assist with social continence which is a key functional outcome.

- Clinical genetics can provide clarity around genetic testing and recurrence risks.

- Orthopedic surgeons assist with the monitoring of skeletal growth and mobility.

- Developmental pediatricians, physiatry, and advanced practice nurses who focus on developmental disabilities can offer functional programming to optimize function, resource identification, advocacy, and access to care.
should be available. Lastly, termination of the pregnancy should be offered to the family [11].

Upon identification of an open NTD, the first step is to review the prenatal testing available to date and clarify any misunderstanding or confusion that exists [3]. The ultrasound, if at all unclear, should be repeated with a high resolution exam in a maternal fetal unit to clearly define the location and size of the NTD and confirm if it is open or closed [16]. In addition, the examination for secondary findings such as hydrocephalus should be done [16]. Given the association with other abnormalities, fetal echocardiography should be considered [13]. Genetic evaluation with amniocentesis should also be recommended, particularly for those considering fetal closure. The identification of genetic abnormality in the fetus has important implications for counseling regarding prognosis, pregnancy management, and whether in utero NTD closure is an option [17,18]. Measurement of amniotic fluid acetylcholinesterase helps to differentiate between open and closed NTDs and is a component of many perioperative evaluations for fetal closure [17,18]. Fetal MRI also may be considered, particularly if unclear findings on ultrasonography exist [19].

In order to make decisions, counseling should provide information that describes the likelihood of survival and the spectrum of medical conditions associated with NTDs [4,12]. These include neurosurgical (hydrocephalus, Chiari II malformation, tethered cord syndrome), urological (neurogenic bladder, incontinence, social continence), gastrointestinal (constipation, neurogenic bowel, incontinence, social continence), orthopedic (ambulation probability, assistive devices, musculoskeletal complications), and dermatological (de-cubitus ulcers, sensory loss) conditions [1,2]. Treatment options for these conditions should be reviewed with an emphasis on optimizing function, rather than ‘fixing’ [20].

The ideal team to provide this information includes specialists in fetal medicine/obstetrics familiar with the management of a pregnancy complicated by a prenatal diagnosis of an NTD [10–12]. These providers are often the first to encounter the family and introduce this life changing information (see below). In addition, key members of the team include neurosurgeons familiar with NTDs as they can provide detailed information about surgical approaches (both antenatal and postnatal) and provide both short-term and realistic long-term expectations for the child and family [11,12]. Experts in clinical genetics clarify testing results, provide information about folic acid, and discuss recurrence risk as well as potential implications for future pregnancies [3,7,17,18]. Developmental pediatricians, physiatrists, and advanced practice nurses provide a focus on developmental disabilities and focus on the optimization of function. These specialists can provide insight into potential medical needs as well as functional goals across a lifespan using evidence-based information [3,20]. They also assist with resource identification, access to care, and utilization as well as caregiver support and mental health support. Social workers are another key member of the team as they provide emotional support and mental health screening. They can also facilitate links to local, national, and international financial resources and sources of emotional support [12,14]. Urologists provide a key aspect of the care team as they can discuss options to optimize function, particularly as it relates to various degrees of urinary continence and social continence [12]. Orthopedic surgeons provide the management of skeletal growth, mobility, and function with participation and inclusion [12]. Lastly, it is beneficial for families to meet with the neonatal team, to understand what to expect at delivery, the likelihood of admission to the neonatal intensive care unit, as well as normal newborn care such as breastfeeding, support for breastfeeding and skin-to-skin care [11,12,14,21,22]. They can review what potential outcomes to expect should the child have additional support needs as a result of premature gestational age.

It is important that families understand that the etiology of NTDs is not completely understood and that the NTD was not caused by their actions. Counseling should include the importance of folic acid in diminishing the chance of a pregnancy being affected by an NTD, although it is not entirely preventative [23,24]. The recurrence risk should be reviewed along with the 1991 US Public Health Service guideline for daily consumption of 4 milligrams (4000 micrograms) of folic acid beginning prior to conception (one month) and

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**Table 1, continued**

| Guideline | Guideline details | Evidence |
|-----------|-------------------|----------|
| - The neonatal care team is important as they will often be involved in the initial resuscitation and providing initial links to subspecialty care. They can also help with establishing important newborn care that is universal to all children, including those with NTDs. | [11, 12, 14, 21, 22] |
| - Social work support is critical to provide mental health support to the family and individual as well as to facilitate links to resources (educational and financial). | [12, 14] |
continuing through the first three months of the pregnancy (see Women’s Health Guidelines).

General principles around lesion level and outcomes should be reviewed, with an emphasis that specific predictions of function are challenging based on lesion level [15]. In addition, the consultation should include a discussion of what to expect cognitively, with acknowledgment that exact predictions of cognitive function are challenging. Rather, a description of the variety of challenges, such as specific aspects of learning, need for special education support, and learning skills being impacted should be reviewed [4,12].

In addition to the discussion of medical care and outcomes, much of the counseling should focus on functioning and independence, including a discussion of disability [7]. These recommendations recognize that perceptions around disability are highly variable. It is essential, therefore, that the consultation be done in a value-neutral manner, avoiding words such as ‘risk’ or ‘bad news’, and instead using words that impart the importance of the decision, such as ‘significant outcome’ and ‘potential challenges’ [7]. We recommend using these value-neutral words as the condition is described in both medical and functional terms. This approach allows the family to assign their own values to the important news provided, rather than adopt the medical provider’s values or biases. It is recommended that the approach be collaborative, seeking to understand from the family their values and cultural beliefs with the goal of a shared decision making process [5,7].

5. Discussion

This guideline was developed to help providers who care for pregnancies affected by SB, as well as those who care for SB postnatally, to conceptualize key components involved in prenatal counseling. Limited studies exist describing parental experiences and the variables that influence decision making. While short-term outcome data for prenatal surgery is promising, long-term outcome data are not yet available. In addition, there is limited data on trauma-informed care and this approach to prenatal counseling.

There is also limited data on intervention strategies to facilitate coping for parents and families as they navigate the health care system.

While there is some guidance to best practice approaches regarding counseling of families affected by SB prenatally, this is a rapidly changing field due to the intense interest and advances being made in the field of prenatal repair for this diagnosis. Further research is needed to understand what variables and data are helpful for decision making and research delineating parent experience with this process will assist in improving the counseling and support provided. Key components, however, will remain as outlined in this guideline: parental choice, collaborative shared decision making, and data provided in a value-neutral manner.

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The Spina Bifida Association has already embarked on a systematic process for reviewing and updating the guidelines. Future guidelines updates will be made available as they are completed.

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Conflict of interest

The authors have no conflicts of interest to report.

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