Prenatal Counseling

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Introduction

The prenatal diagnosis of Spina Bifida or a neural tube defect (NTD) is a pivotal moment for families. NTDs can be open or closed, and the type of finding determines important counselling considerations, particularly as closed NTDs often do not need surgical intervention in the neonatal period. The manner in which information about NTDs is conveyed is critical to the family's future and provides the basis for important family decisions (e.g. possible fetal surgery versus postnatal closure for open NTDs, adoption and termination).[1] 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 around disability, and this may impact families' expectations about treatment options and/or their understanding of the condition. It is essential that parents and caregivers are told about the clinical course and the anticipated strengths and challenges associated with Spina Bifida. This information should be individualized and provided in a neutral and collaborative manner that meets the needs, values, and beliefs of each family.[2-3] Families should be offered the opportunity to meet with key members of the Spina Bifida team.[1,4-5] Neurosurgeons experienced with and dedicated to caring for patients with NTDs are uniquely qualified to discuss both short term and realistic long-term expectations and challenges facing a child born with Spina Bifida.

Outcomes

Primary

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

Secondary

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

Tertiary

1. Provide families with information about providers and resources that will assist in the caring for their child with Spina Bifida.

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 a 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 a NTD?

Guidelines

1. Convey information about the medical care and lifelong functional impact of Spina Bifida in a value-neutral, collaborative manner while seeking from families an understanding of their needs, values, and beliefs.
    - 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.[5-6]
    - 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. (clinical consensus)
    - Base consultation with families on a collaborative, shared decision-making model that includes the medical team and parents. (clinical consensus)
    - 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.[3-4]
    - Offer a review of prenatal testing and results to clarify any misunderstandings or confusion that may exist.[1] Typically, the diagnosis is made by a high-resolution ultrasound examination that is performed during the second trimester at a maternal–fetal medicine unit. An 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.[7] 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 prognosis, pregnancy management, and determining whether the patient is a candidate for in-utero NTD repair.[8-9] 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.[10]
    - Expect to provide critical information about the likelihood of survival and the spectrum of outcomes (i.e. neurosurgical, cognitive, developmental, urologic, orthopedic, dermatologic) for children with NTDs.[5,11]
    - Discuss disability. Provide information on outcomes with a lifespan approach.[3]
    - Review general principles associated with lesion levels, as well as the difficulty with providing specific predictions based on lesion level.[12]
    - Review treatment options for conditions associated with NTDs with an emphasis on functional outcomes.[11]
2. Review evidence-based treatment options with the family, including fetal surgery.
    - Treatment options should include prenatal closure for open NTDs offered at treatment centers with expertise in the surgical and obstetrical management of NTDs.[4-6,13] It is recommended that 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.[4-6,11,13]
    - In addition to the option for prenatal closure, also present the option for term delivery and postnatal closure for open NTDs. Explain to parents and caregivers that caesarian 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.[12]

- Although closed NTDs usually do not require surgical intervention in the newborn period, it is recommended that they have the same monitoring and investigations in the newborn period as open NTDs. (clinical consensus)
- Present adoption as an option for parents who are not open to termination but are not able to raise a child with a disability. (clinical consensus)
- Review termination of the pregnancy as another option for the family.[4]

3. Offer families the opportunity to meet with key members of the Spina Bifida care team:
    - Specialists in fetal medicine and/or obstetrical medicine are familiar with managing pregnancies complicated by a prenatal diagnosis of NTD. These providers are first to share the results of the testing. The prenatal diagnosis of Spina Bifida should be made in a value-neutral manner.[4,13]
    - Neurosurgeons provide information about management approaches such as fetal surgery and postnatal closure.[4-5] Neurosurgeons experienced with and dedicated to caring for patients with neural tube defects (NTDs) are uniquely qualified to discuss both short term and realistic long-term expectations and challenges facing a child born with Spina Bifida.
    - Experts in clinical genetics can clarify test results, discuss the genetics of NTDs, provide information about folic acid, and discuss recurrence risk and potential impact on future pregnancies.[1,3]
    - Developmental pediatricians, advanced practice nurses, and physiatrists focus on childhood disabilities and how to optimize function. These specialists can provide essential insight into potential medical needs and functional goals across the lifespan. They provide parents with evidence-based, up-to-date information. They can also assist with resource identification, access to care, and utilization, including caregiver support and mental health resources.[1,14] (Family Functioning Guidelines, Mental Health Guidelines)
    - Social workers provide critical emotional support and screening for parental mental health and are recommended to be included in all consultations. They provide families with links to important financial resources and sources of emotional support, including caregiver support and mental health resources.[5,13] They can also provide information to families about local, national, and international sources.[1] (Family Functioning Guidelines, Mental Health Guidelines)
    - Urologists and orthopedists can provide more detailed discussions on interventions available for optimizing functional outcomes.[5] (Orthopedic Guidelines, Urology Guidelines)
    - Neonatologists can provide information and resources in advance about the child's immediate care needs such as breastfeeding, skin-to-skin care, and tours of the neonatal intensive care unit (NICU).[5,13]
4. Offer information about what to expect at birth.
    - Review that the child may need to be admitted to a special care or intensive care nursery (all with open NTDs, some closed NTDs may be cared for collaboratively with the nursery and community teams) and that psychosocial support is available to them in that setting.[4] (Family Functioning Guidelines, Mental Health Guidelines)
    - Help families to anticipate that specialists in Spina Bifida will need to be present at the delivery to examine the child, that there exists the possibility that intravenous fluids/antibiotics will be necessary, and of the possibility that the child will need additional support.[4]

    - If fetal surgery is not an option, review the timing of delivery by 37 weeks[12] and post-natal management. The first step should be the closure of the spinal defect within 24 hours[12] followed by attention to hydrocephalus as indicated.[15]
    - Emphasize typical aspects of newborn care, including breastfeeding and skin-to-skin care.[16-17]
    - Counsel families who choose to continue the pregnancy that there are many normal aspects of pregnancy, caregiving for the newborn and parenting across the lifespan. (clinical consensus)
5. Discuss folic acid and recurrence risk.
    - Reassure the parents that while NTDs are not completely understood, this birth defect was not something that was caused by their actions.[1]
    - Reassure the parents that while folic acid can help diminish the chance of a pregnancy being affected by NTD, it is not entirely preventative.[18-19]
    - Counsel women about their recurrence risk and review the 1991 U.S. Public Health Service guideline for daily consumption of 4 milligrams (4000 micrograms) of folic acid beginning at least one month (but preferably 3 months) before they start trying to get pregnant and continuing through the first three months of pregnancy. (Women's Health Guidelines)

Research Gaps

There exist limited studies describing parental experiences and the variables that influence decision-making. While the 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 counselling. There is also limited data on intervention strategies to facilitate coping by parents and families as they navigate the health care system.

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