

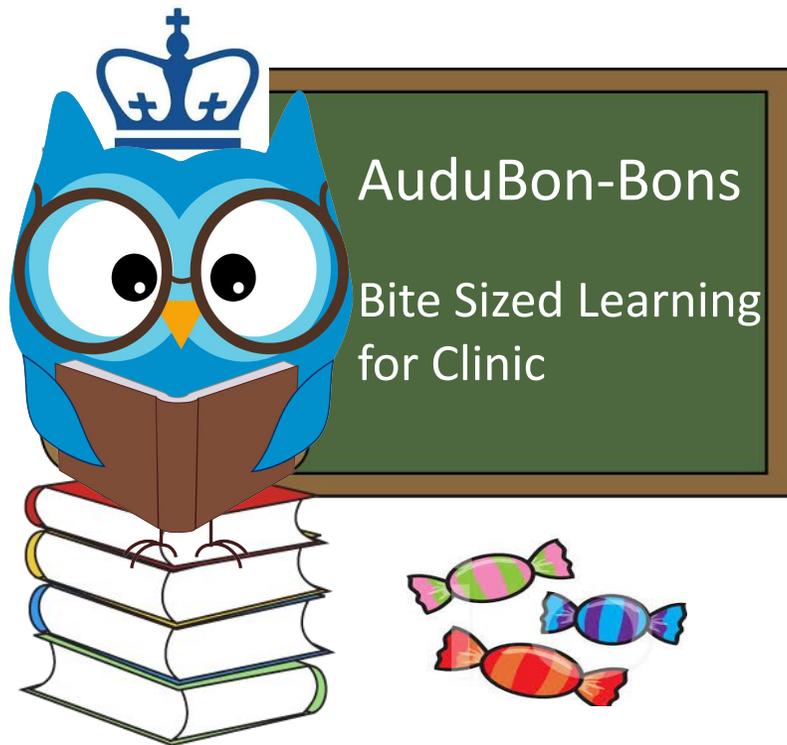
# PRENATAL CARE: NEURAL TUBE DEFECTS

Week 3

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Reading Assignment

<https://www.obgproject.com/2018/08/01/neural-tube-defects-a-guideline-based-approach/>



# LEARNING OBJECTIVES



- Review the different types of neural tube defects and their etiology and pathophysiology
- Be able to identify the methods for screening and diagnosis of NTDs
- Be comfortable initiating a conversation with patients about NTDs and how to make preventive recommendations based on risk stratification
- Be comfortable making an antenatal plan for patients with a fetus diagnosed with an NTD



# CASE VIGNETTE

- Ms. A. Ma is a 32yo G2P0010 woman at 13 weeks EGA who presents to clinic for her second prenatal visit. She is doing well and has no complaints. This is a planned pregnancy and they are very excited.
- You have just finished reviewing the lab results from her initial visit, which were all normal. She wants to discuss what kind of testing can be done to make sure the baby has no spinal cord abnormalities



# FOCUSED HISTORY

## What elements of the patient's history are most relevant?

- **POB:** 1 TOP at 19 weeks due to anencephaly
- **PGYN:** Regular menses  
No STI/cysts/fibroids  
No abnormal paps
- **PMH:** Denies
- **PSH:** Denies
- **Meds:** PNV
- **All:** NKDA
- **Soc:** Denies toxic habits; works as a teacher; lives with her husband
- **FHx:** No genetic abnormalities; FOB has no family members with genetic abnormalities



# PERTINENT PHYSICAL EXAM FINDINGS

**What elements of the patient's physical exam are most relevant?**

- **P:** 80 **BP:** 116/70    **Wgt:** 60kg **Hgt:** 160cm    **BMI:** 23.4
- **Abd:** Soft, NT/ND
- **FHR:** 145
- **Ext:** NT b/l

**What will be pertinent in her prenatal labs?**

- **Normal First Trimester Screen**



# PATHOPHYSIOLOGY

**What developmental process when compromised, results in NTDs?**

- The neural tube commences closure at the cervical region and extends in both cranial and caudal directions
- Failure of this closure at any point results in different NTDs



# TYPES of NEURAL TUBE DEFECTS

- What structures are impacted by NTDs?
  - Brain
  - Spinal cord
  - Meninges
  - Vertebra



# TYPES of NEURAL TUBE DEFECTS

- What are examples of NTDs involving the structures below?

- **Brain**

Anencephaly — Absence of all/part of brain/skull/skin

Exencephaly — Failure of scalp/skull formation

Encephalocele — Failure of complete skull formation

- **Spinal cord & Meninges**

Spina bifida — Exposed spinal cord and/or meninges

Myeloschisis — Exposed flattened neural tissue mass

Meningocele — Exposed meninges

Holorachischisis — Exposed spinal cord in entirety

Myelomeningocele — Exposed meninges and neural tissue

Craniorachischisis — Anencephaly + open NTD

- **Vertebra**

Iniencephaly —

Abnormal brain tissue

&

extreme retroflexed cervical and upper thoracic spine



# CLINICAL SEQUELAE

- What are important clinical features to consider when discussing the impact of NTDs in the following areas?

CNS

Ambulation

Bladder function

Bowel function

*NB:*  
Approximately  
**30%** of  
individuals  
with NTD will  
have a severe,  
life-threatening  
latex allergy



# RISK FACTORS/ETIOLOGY

What are factors that increase the risk of having a fetus with an NTD?

## Environmental factors

(present during first 28 days of development)

- Geographic regions
- Toxin exposure
- **Hyperthermia**
  - Fever
  - Heat exposure (hot tubs)
- Medications (that deplete folic acid)
  - e.g. **Antiepileptic drugs**
- **Folate deficient diet**

## Genetic factors

- Chromosomal abnormalities (T13, T18, Triploidy)
- Single gene disorders (22q11.2 deletion syndrome)

## Family history

- Personal/family history of NTD
- Previous NTD-affected pregnancy

## Maternal conditions

- Pregestational diabetes
- Obesity



# SCREENING/DIAGNOSIS

**What is the primary prenatal screening method for NTDs?**

- Maternal serum alpha-fetoprotein ([MSAFP](#))
  - Secreted by [Fetal yolk sac and liver](#)

**What is the utility of MSAFP for detecting closed NTDs?**

- MSAFP is not usually increased with closed NTDs

**Is the presence of ultrasound findings consistent with NTD at 18-22 weeks gestation considered diagnostic?**

- Yes

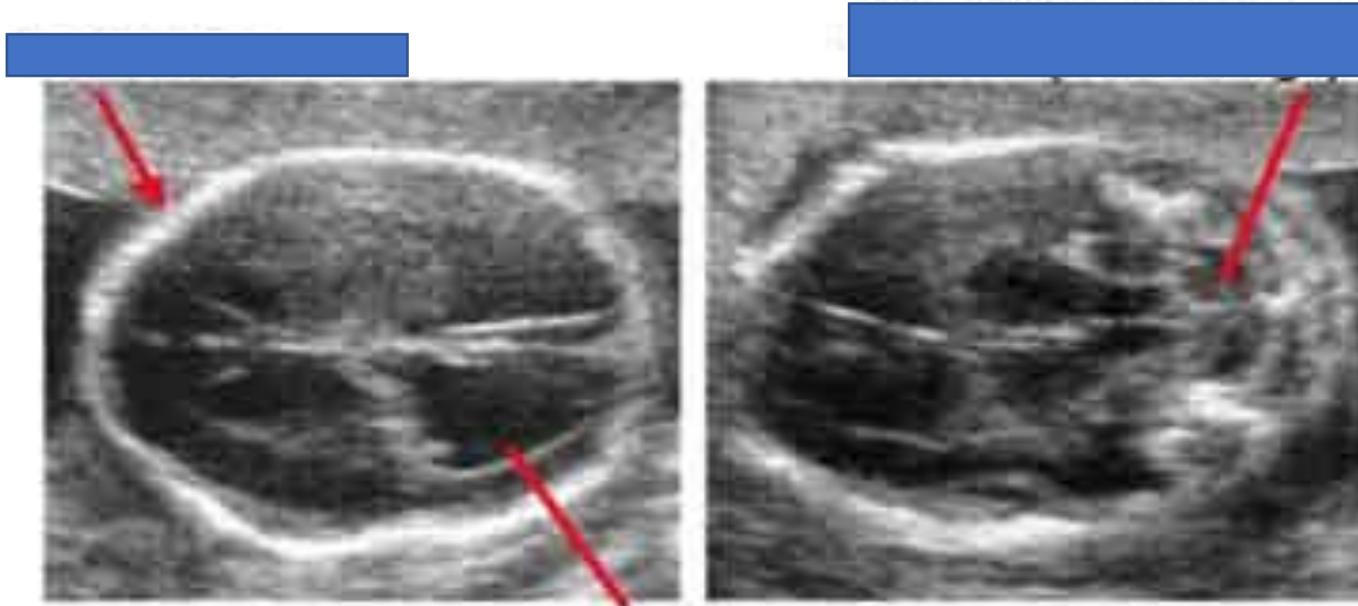
**What can differentiate open from closed NTDs in unclear cases?**

- Amniocentesis with measurement of [acetylcholinesterase](#)



# ULTRASOUND FINDINGS

- Can you identify these ultrasonographic findings?



<http://www.obgyn.upenn.edu/res/ntd.htm>

- What NTD are they typically associated with?
  - Open spina bifida



# ANTENATAL MANAGEMENT

- What are the recommendations for folic acid intake for the following populations?
  - All women planning a pregnancy
    - 400mcg supplementation daily from 1 month before pregnancy through first 12 weeks
  - Women at high risk of NTDs
    - 4mg supplementation daily from 3 months before pregnancy through 12 weeks gestation
- If an NTD is detected, what is the appropriate management plan?
  - MFM referral for further evaluation, counseling, and management
- For patients proceeding with pregnancy,
  - Fetal echo
  - Amniocentesis, including microarray
  - Care team should include MFM, Neonatology, Pediatric neurosurgery, & Genetics



# COUNSELING

**What are key factors to consider when counseling a patient after a NTD is detected?**

- Counseling should be individualized and specific
- Include anticipated survival, outcome and variability of these
  - i.e. Open spina bifida - highly variable outcomes dependent on lesion size/location, other anomalies & associated genetic abnormality
- Offer full spectrum of management
  - Termination, expectant management with neonatal surgery, in utero fetal surgery for appropriate candidates



# DELIVERY PLANNING

- **In what scenarios does a NTD result in a late-preterm or early-term delivery?**
  - If **in utero fetal surgery** has been performed, the increased risk of uterine rupture will require a Cesarean **delivery timed similarly to a patient with a prior classical Cesarean** delivery
  - Delivery prior to term may be indicated in the event of **rapidly increasing ventriculomegaly** to allow **for placement of a VP shunt**
- **What is the recommended mode of delivery for a fetus with NTD in cephalic presentation?**
  - The best route for delivery remains **controversial**
  - Current published evidence suggests vaginal delivery does not adversely affect neonatal outcome
  - However, due to lack of clarity, decisions regarding timing/route of delivery should be **individualized with a multidisciplinary approach**



# SOCIAL DETERMINANTS OF HEALTH

Compared to non-Hispanic women, Hispanic women are:

- Less likely to take prenatal folic acid supplements
- Have lower blood folate concentrations
- Have a higher prevalence of spina bifida and anencephaly



- Patients in this community would benefit for stronger outreach to encourage preconception counseling.
- A larger patient education effort needs to be made for our Latinx patients during GYN visits and preconception counseling to ensure they are adequately informed about the importance of folic acid intake prior to and during early pregnancy; this should include information about NTDs and associated sequelae.
- Regulations regarding folic acid fortification of grains need to account for variability of staple grains in different communities and ensure that regulatory policies are inclusive of them.



# EPIC .PHRASE

## .B BonNTDPCounseling

### Description: Preconception folic acid counseling

The patient was informed of the importance of folic acid intake during early pregnancy and the preceding months; she was also informed of the risk of neural tube defects and associated sequelae with inadequate folic acid intake during this time.

#### Folic Acid Supplementation

\*\*\*As the patient has no risk factors for increased risk of NTDs, we discussed intake of 400mcg supplementation daily from 1 month before pregnancy through first 12 weeks. She was informed that this is the amount available in most PNV formulations.

\*\*\*Given the patient's increased risk for pregnancy affected by NTDs, she was counseled on the recommendation of 4mg supplementation daily from 3 months before pregnancy through 12 weeks gestation. She was informed that this amount exceeds that found in most PNV formulations and would require supplementation in addition to a daily PNV tablet.



# CODING AND BILLING

- ICD-10

- **Z31.69** - Encounter for other general counseling and advice on procreation
- **O35.0** - Maternal care for (suspected) central nervous system malformation in fetus
- **Q05.9** - Spina bifida unspecified
- **Q76.0** - Spina bifida occulta
- **Q05.4** - Unspecified spina bifida with hydrocephalus



# EVIDENCE

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