

TelAbility

Enhancing the lives of children with disabilities



Spina Bifida Update 2011

Joshua J. Alexander, MD
Director
The Spina Bifida Clinic
@ UNC



Objectives

- *Know the proposed etiology of Spina Bifida*
- *Know the importance of folic acid in the prevention of Neural Tube Defects*
- *Be familiar with prenatal diagnosis and management options for Spina Bifida*
- *Know the signs, symptoms, and treatment of primary conditions commonly associated with MMC*
- *Be familiar with secondary conditions associated with MMC*
- *Be familiar with recent research trials in the treatment of spina bifida and its sequelae*

Lecture Outline

- *Prevalence, incidence, etiology and prevention of spina bifida*
- *Prenatal diagnosis and management*
- *Types of spina bifida*
- *Motor effects*
- *Sensory effects*
- *Hydrocephalus and Arnold-Chiari II*
- *Spine abnormalities*
- *Neurogenic bowel and bladder*
- *Secondary conditions*
- *New Approaches to care*

Prevalence

- *2nd most common physical disability in childhood*
- *1/1000 live births nationally*
- *(1-2/1000 live births in North Carolina)*



Incidence

- *Increased risk in those of Irish, German, or Hispanic descent*
- *Decreased risk in Asians and Pacific Islanders*

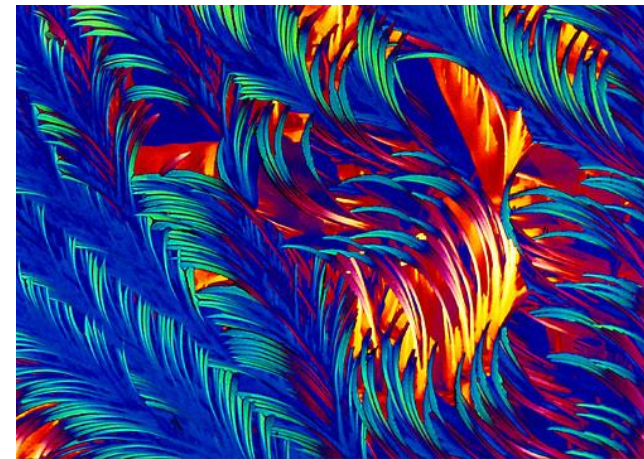
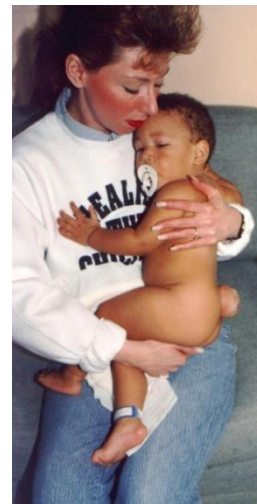
Duke Center for Human Genetics Study
866-385-3683, ntd@chg.duhs.duke.edu

Etiology

- *Polygenic Inheritance*
- *Environmental Influences (Nutrition, heat, valproic acid, Agent Orange, pesticide exposure)*

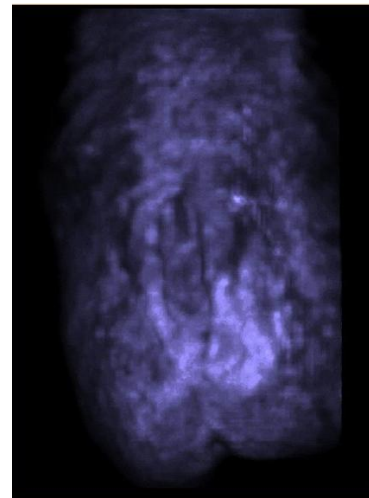
✓ *Maternal pesticide exposure and neural tube defects in Mexican Americans. Ann Epidemiol 2010 January;20(1):16-22*

- *MTHFR*
- *Folic Acid*



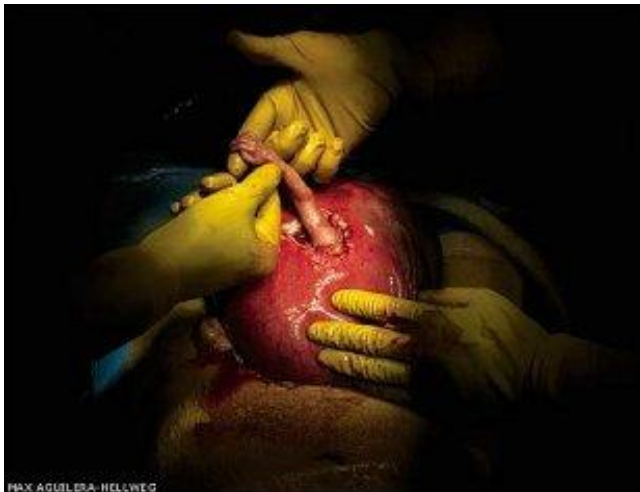
Prenatal Diagnosis

- *Alpha-fetoprotein in amniotic fluid*
- *Acetylcholinesterase in amniotic fluid*
- *Fetal Ultrasound*



Prenatal Options

- *Termination of pregnancy*
- *C-section delivery*
- *Fetal surgery*



MAX AGUILEIRA-HELLWEG

FETUS WITH NORMAL SPINE

▲ FETUS WITH MYELOMENINGOCELE

1. Part of the spinal cord and spinal nerves, usually encased in a sac, protrude through an opening in the back and are exposed to the amniotic fluid.
2. The brainstem (hindbrain) descends, or herniates, into the spinal canal in the neck and blocks the circulation of cerebrospinal fluid. This can cause a damaging buildup of fluid in the brain called hydrocephalus.

▲ FETUS AFTER SURGICAL REPAIR

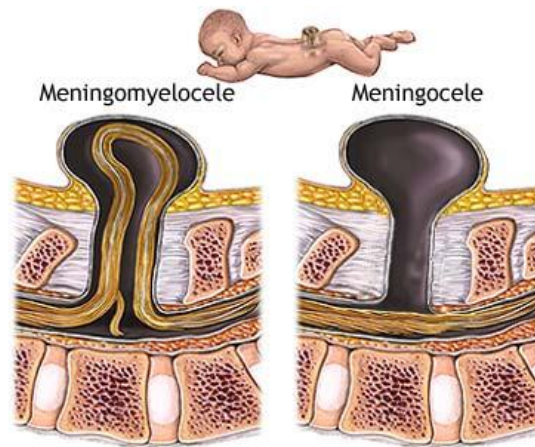
1. Fetal surgery repairs the defect, returning the spinal tissue to its proper place and covering the opening in the fetus' back.
2. The hindbrain herniation gradually reverses after repair, and the brain stem returns to its normal position.

Spina Bifida Occulta

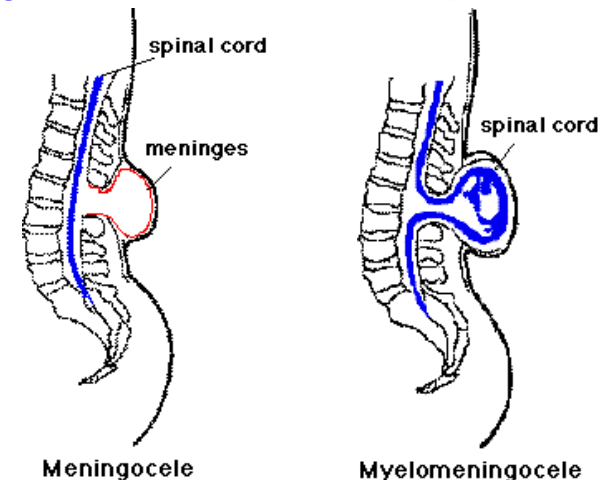
- *1 of 5 people*
- *Failure of vertebrae to fuse (L-S level)*
- *Associated spinal cord or nerve root malformations*
- *Pigmented nevus, angioma, tuft of hair, dimple or dermal sinus*
- *KEY RISK = tethered cord*
- *Not usually associated with Arnold-Chiari malformation*

Spina Bifida Cystica

- **Meningocele** = meningeal cyst filled with fluid (neuro exam may be normal)
Not associated with hydrocephalus
- **Myelomeningocele** = sac also contains dysplastic nerve tissue
✓ (with A/C malformation + Hydrocephalus)



ADAM



Nerve Involvement

- *NOT ALWAYS SYMMETRIC*
- *Increased or decreased muscle tone*
- *Muscle weakness*
- *Decreased sensation*
- *Neurogenic Bowel*
- *Neurogenic Bladder*
- *Vasomotor dysfunction*



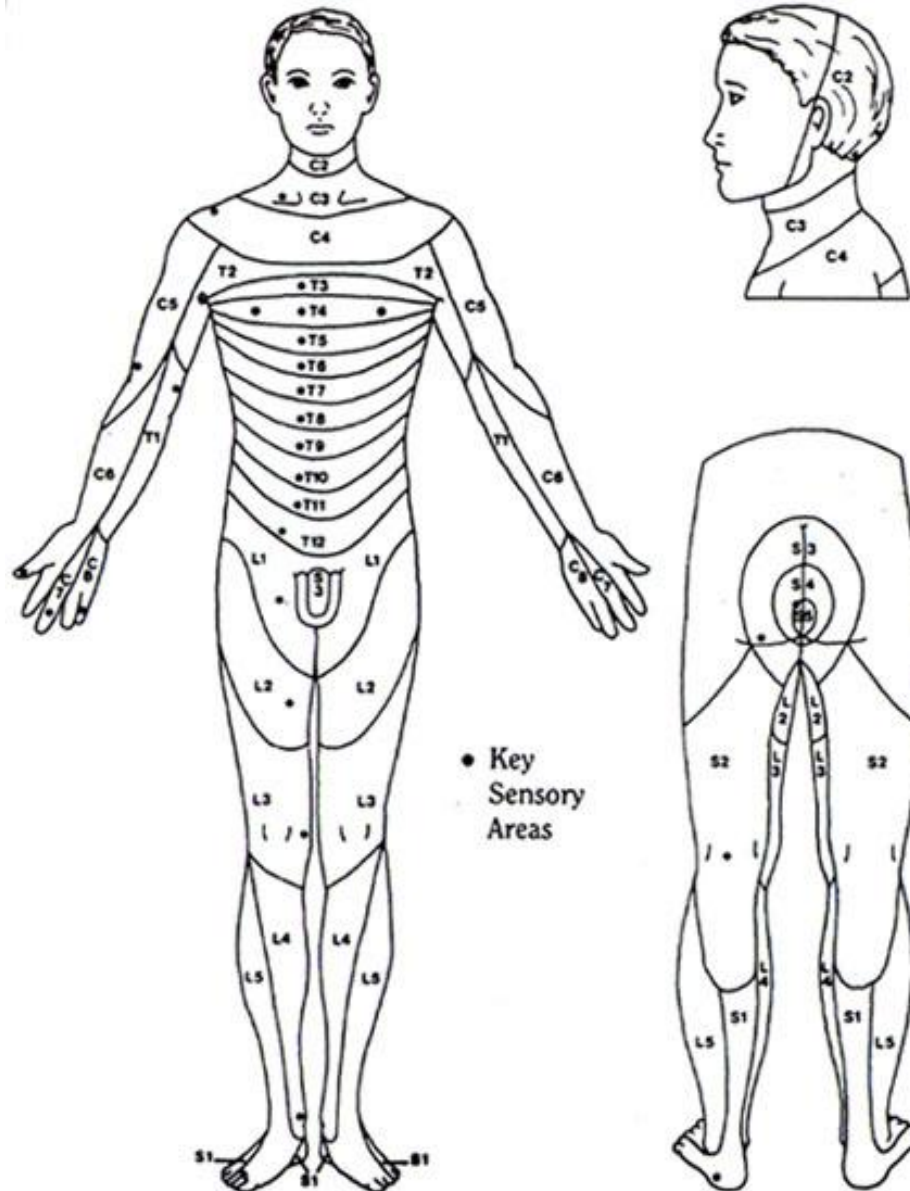
Motor Levels

- *Thoracic*
- *L1-L2*
- *L3*
- *L4*
- *L5*
- *S1*
- *S2-S5*



Sensory Levels

- T4
- T10
- L1
- L2
- L3



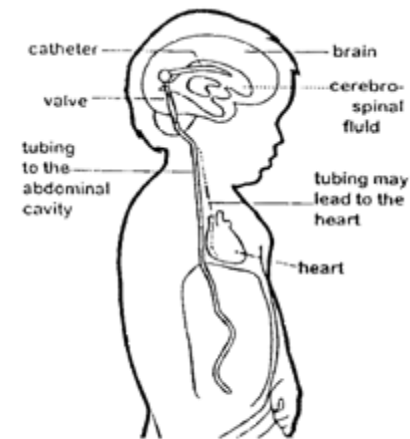
- L4
- L5
- S1
- S2
- S3-S5

Hydrocephalus

- *Most commonly due to obstruction of CSF flow*
- *VP Shunt done in 85-90% of MMC
(1/3 will require revision at some time)*



Children's Hospital, Boston



Shunt System

Hydrocephalus

- *Symptoms: Headache, irritability, N/V*
- *Acute Signs: Large head, bulging fontanelle, prominent scalp veins, somnolence*
- *Chronic Signs: Decreased school performance, personality changes, decreased fine motor skills*

Arnold-Chiari II Malformation

- Definition: Medulla, Pons, 4th ventricle +/- cerebellar vermis herniated into the cervical spinal canal
- Incidence: 80-90% of those with MMC
- Symptomatic: ~20%
- Is it pushed (hydrocephalus), pulled (tethered cord), or crowded out?

Arnold-Chiari II Malformation

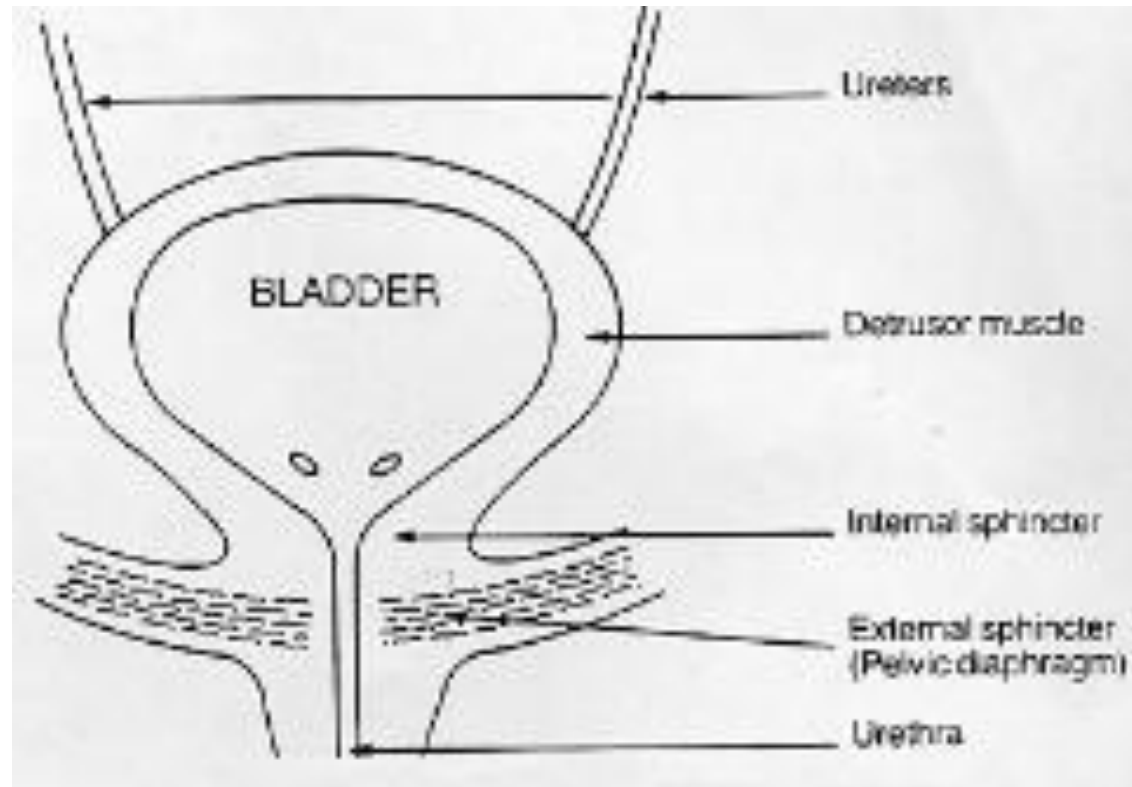
- *Stridor*
- *Laryngeal nerve palsy / vocal cord paralysis*
- *Periodic breathing*
- *Sleep apnea*
- *Dysphagia*
- *Aspiration pneumonia*
- *CENTRAL RESPIRATORY DYSFUNCTION*
(now the most common cause of death in MMC)

Spine Abnormalities


- *Bifid Spine*
- *Vertebral fusion*
- *Hemivertebrae*



Neurogenic Bladder

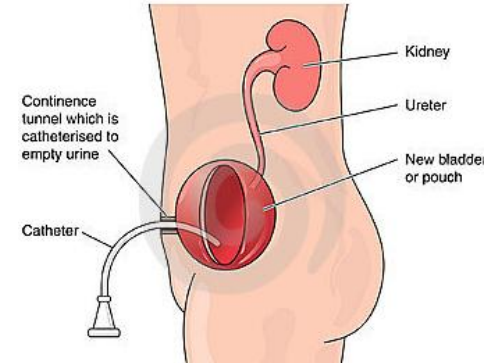


Neurogenic Bladder

- *Incontinence*
 - *UTI*
 - *Hydronephrosis*
 - *Pyelonephritis*
 - *Renal Calculi*
- 
- **INTERMITTENT CATHETERIZATION !**
(Only 5% will void with continence)

Bladder Care

- *Intermittent catheterization*
- *Mitrafanoff Procedure*
- *Hyaluronic acid injections to reduce reflux*
- *Botulinum toxin injections to reduce bladder /sphincter pressures (off-label)*
- *Neuromodulation using transcutaneous approaches with interferential electrostimulation (Experimental)*
- *Nerve re-routing for “re-innervation” (XIAO procedure) (Experimental)*



Neurogenic Bowel

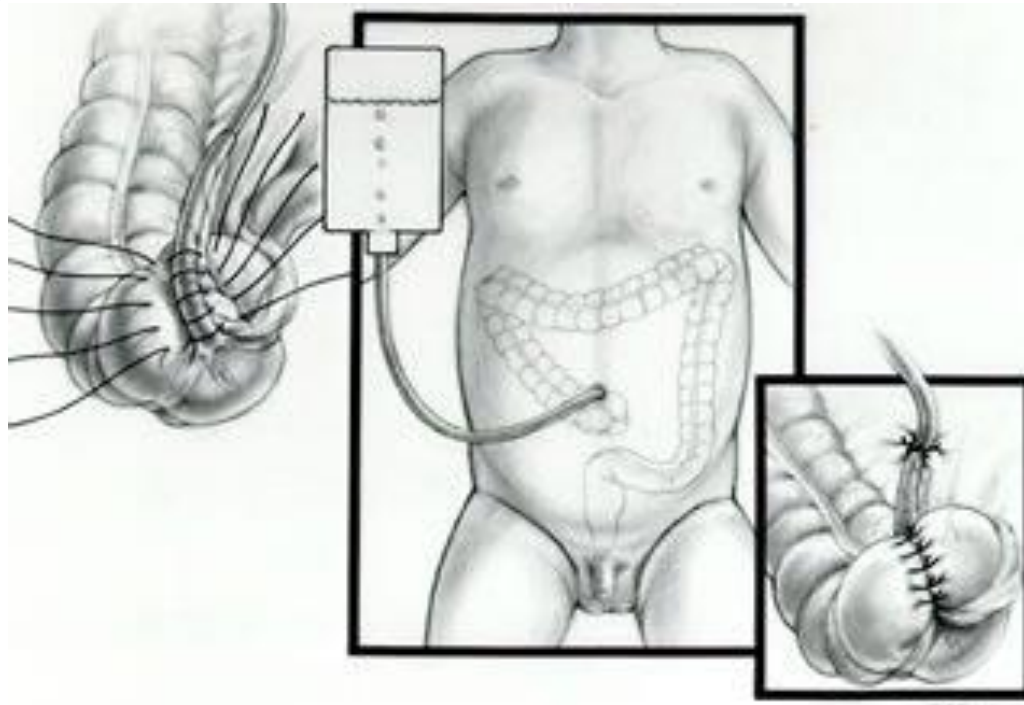
- *Colon, Rectum and internal anal sphincter are affected*
- *Decreased motility*
- *Constipation*
- *Fecal overflow*
- *Incompetent Rectum*
- ***NEED BOWEL TRAINING PROGRAM !***

Neurogenic Bowel

- *High fiber diet*
- *Stool softeners*
- *Gastro colic reflex*
- *Suppositories*
- *Enemas*
- *Biofeedback (if intact anocutaneous reflex)*
- *(M)ACE procedure*



MACE Procedure



Secondary Conditions

- *Fractures*
- *Hypertension*
- *Hip dislocation*
- *Scoliosis*
- *Kyphosis*
- *Foot anomalies*
- *Pressure ulcers*
- *Burns*
- *Obesity*
- *Precocious Puberty*
- *UTI's*
- *Hydronephrosis*
- *Latex Allergy*
- *Syringomyelia*
- *Tethered Cord*
- *Rotator Cuff tears*
- *CTS*
- *Ulnar Neuropathy*

Team Approach

- *PM&R*
- *ORTHOPEDICS*
- *NEUROSURGERY*
- *UROLOGY*
- *PT*
- *OT*
- *MSW*
- *EDUCATOR*

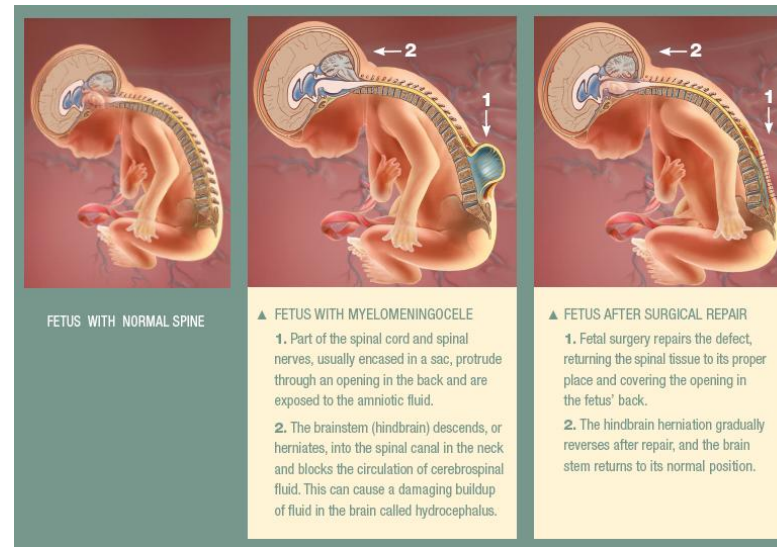
Crucial Periods

- *After diagnosis*
- *After Birth*
- *First Year of Life*
- *Preschool*
- *1st grade*
- *Middle School*
- *High School*
- *Transition to Adulthood*



Fetal Surgery

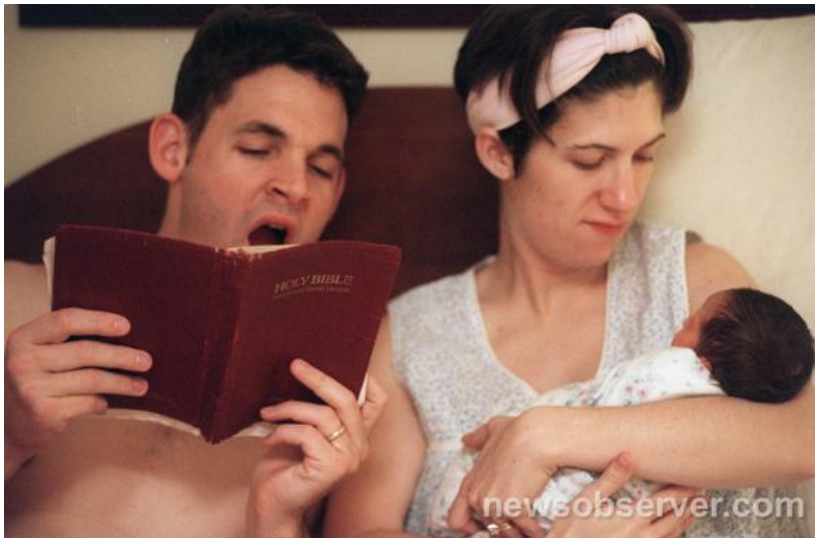
- *Reversal of the hindbrain herniation (the Chiari II malformation)*
- *Reduced Need for VP shunt*
- **Reduced Incidence or severity of motor impairment*



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Susan (& Anna)





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Questions ?