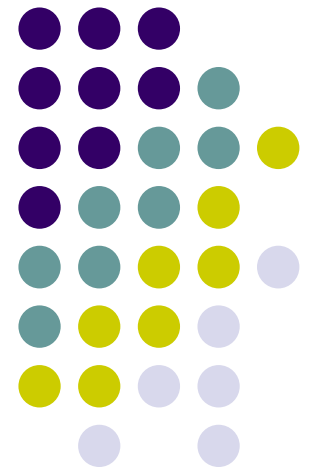
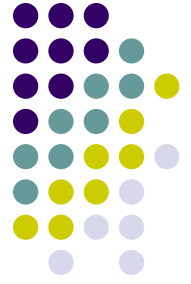


Pediatric and Adult Preventive Care in Rett Syndrome and *MECP2* Mutations

Jane Lane, BSN, RN
University of Alabama at Birmingham
Colorado Springs, CO
May 29, 2010

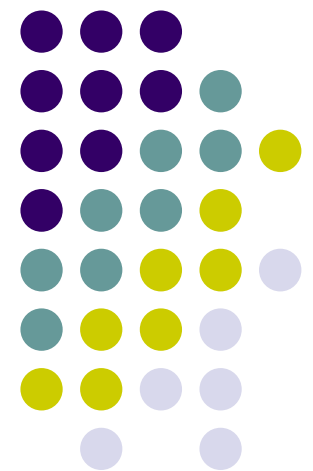




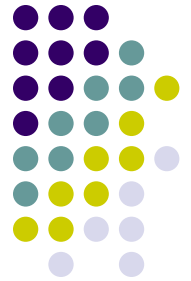
Points of Discussion

- Child and adult preventive care
- When to call the doctor and surgery considerations
- Vaccinations

PREVENTIVE CARE

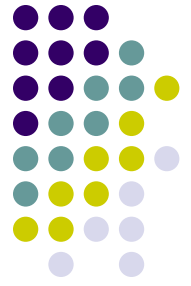


Childhood Preventive Care



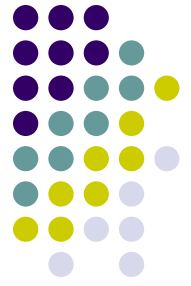
- Well child visits with primary care provider
- Dental care every 6 months or as needed
- Vision and hearing as indicated
- Vaccinations as per AAP guidelines
- Rett syndrome/*MECP2* specific considerations
 - EKG screening at dx, then annually
 - Scoliosis screening at age 4, then as indicated
 - Nutritional assessments including BMI
 - Bone health by xray or DEXA if at risk
 - Physical therapies and seating assessments

Adult Preventive Care



- Cancer screening
 - Skin
 - Breast exams - Monthly “at home” exam; mammography
 - Gynecological exams -
 - Rectal exam for mass and occult blood or ultrasound every 2-3 years if low risk.
 - No need for pelvic or PAP unless increased risk or symptomatic
 - Screening for ovarian cysts if high risk
 - Exam may require sedation
- Dental
- Vision and hearing

Adult Preventive Care



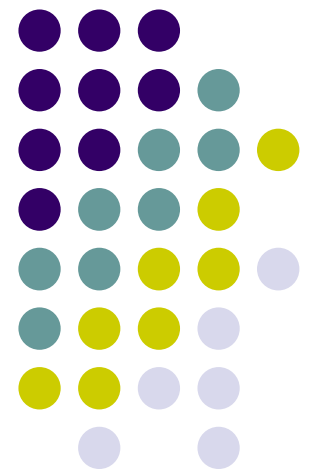
- Immunizations
 - Influenza 18-50 if desired; recommended annually after 50
 - Zoster (shingles) after age 60 regardless of history of chicken pox
 - Meningococcal polysaccharide after age 55
 - Others depending on childhood vaccination history
- Infectious disease screening
 - Hepatitis C – if high risk
 - TB – if high risk
- Other recommended screenings
 - Cholesterol
 - Diabetes type 2
 - Hypertension



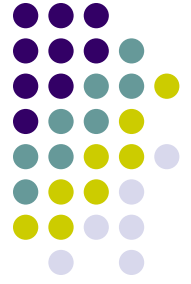
Adult Preventive Care

- Rett syndrome/*MECP2*-specific considerations
 - EKG screening annually
 - Nutritional assessments including BMI
 - Bone mineralization by xray or DEXA
 - Physical therapies
 - Mental health – depression

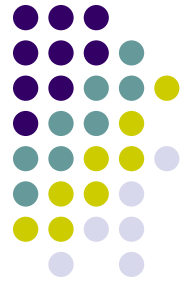
WHEN TO CALL A DOCTOR



Advice....

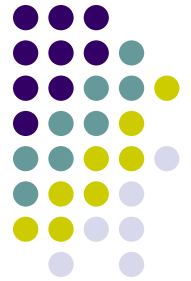


- You wouldn't buy stock based on your family doctor's advice, so why would you get medical advice from your stock broker (or friend)?
- The definitive word when you are worried should be your health care provider



Call when there is

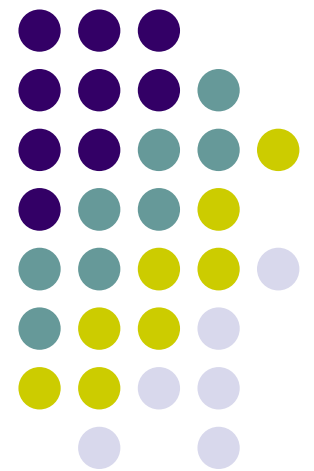
- New onset fever greater than 101° rectally
- Change in consciousness, activity level, fainting
- New onset, increase or change in type of seizures
- Change in limb use or appearance
- Reduced urine output, smelly urine
- Fussy, agitated, inconsolable
- Not eating or drinking, weight loss
- Prolonged vomiting or bilious vomiting
- Rash
- Easy bruising
- Foul (really bad!) breath

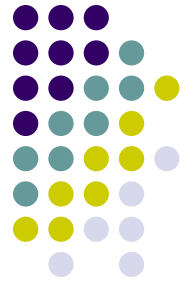


And when there is...

- Change in behavior with new medication
- Blood from any opening
- Anything crawling inside or out
- Established menstrual cycles have stopped
- Distended, tender abdomen
- Change in bowel or bladder pattern
- Anything swollen and/or hot
- Discharge from breasts or vagina
- Skin breakdown at pressure points - knees, ankles, elbows, tailbone

**or when things just
don't feel right**





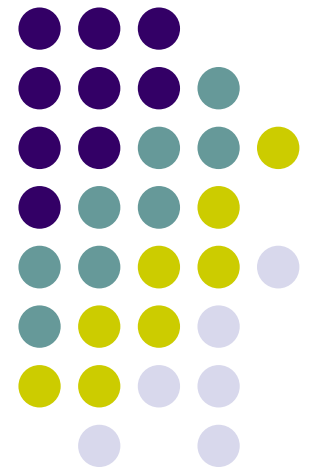
Surgical considerations

- Pre op
 - May require less anesthesia – Propofol study
 - May take longer to awaken post op
 - Breath holding associated with RTT goes away in sleep
- Post op and discharge
 - Stay within your comfort zone for discharge
 - Bleeding management
 - Fluid management
 - Pain management

VACCINATIONS

“.... people who choose not to vaccinate don't just put themselves and their children at risk, but also their communities, which includes infants who are too young to immunize”

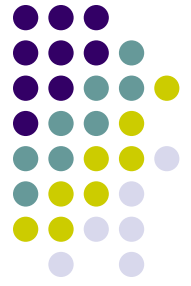
Jane Seward, MD
CDC





Why should I *not* vaccinate?

- NO REASON
 - Research has proven that vaccines do not cause autism
 - Talk over your concerns with a trusted medical person
- Special considerations
 - No live virus vaccines with cancer, oral or injected steroid use, or immune deficiency
 - May delay pertussis part of DTaP with recent history of seizures



Why should I vaccinate?

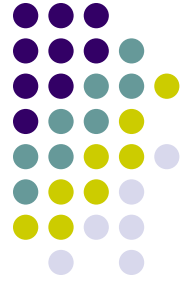
- **Vaccinations work**
 - Used for over 50 years
 - Save lives and decrease disease
- **Vaccinations are safe**
 - Monitored by FDA, CDC, AAP, AAFP
 - No credible evidence of linkage to autism
- **Vaccinations are necessary**

Without Vaccinations in the US



- ***Polio*** would paralyze **10,000** children
- ***Rubella (German measles)*** would cause birth defects and mental retardation in **20,000**
- ***Measles*** would infect **4 million** children, killing **3,000**
- ***Diphtheria*** would be one of the **most common causes of death** in school-aged children
- ***Haemophilus influenzae type b (Hib)*** would cause meningitis in **15,000** children
- ***Pertussis*** would **kill thousands** of infants
- ***H1N1*** is responsible for **36,000** hospitalized and **540** deaths *in children* in the US as of 11/09

Resources For Information



- Food and Drug Administration
 - www.fda.gov
- American Academy of Pediatrics
 - www.aap.org
- Centers for Disease Control and Prevention
 - www.cdc.gov/vaccines
- The Children's Hospital of Philadelphia Vaccine Education Center
 - www.chop.vaccine.edu

That's All!

