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

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Points of Discussion

- Child and adult preventive care
- Finding the care you need
- 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 diagnosis, then annually
  - Scoliosis screening at age 4, then as indicated
  - Nutritional assessments including BMI
  - Bone health by x-ray or DEXA if at risk
  - Physical therapies and seating assessments
Adult Preventive Care

● Primary care routine visits
● Vision, hearing, and dental
● Immunizations
  ● Not just for children & per physicians’ recommendations
● Infectious disease screening
  ● Hepatitis C – if high risk
  ● TB – if high risk
● Other recommended screenings
  ● Cholesterol
  ● Diabetes type 2
  ● Hypertension
Adult Preventive Care

- Cancer screening - consider family history
  - Colon
    - Rectal exam for mass and occult blood or ultrasound every 2-3 years if low risk
  - Skin
    - Look over during bath
  - Breast exams
    - Monthly “at home” and more extensive as needed
  - Gynecological exams
    - No need for pelvic or PAP unless increased risk or symptomatic
    - Screening for ovarian cysts if high risk
    - Exam may require sedation
Adult Preventive Care

- Rett syndrome/MECP2-specific considerations
  - EKG screening annually
  - Nutritional assessments including BMI
  - Physical therapies
  - Bone mineralization by xray or DEXA as indicated
  - Mental health – depression
From whom do you get your medical information?
Whom do you hire when you are building a house?

An Architect
Whom to go to when you need to have some fun?

Friends!
Whom should you ask when you have medical questions?

A physician!!!
Finding a good “doctor”

- Ask other families of special needs children
- Consider your own physician
- Ask the pediatrician early about transitioning to adult physician
- Consider nurse practitioner
- It’s okay to educate
Maintaining a therapeutic relationship

- Make regular appointments and keep them
- Be considerate of cancelations
- Try to arrive on time
- Supply medical records
- Write down questions
- Ask for clarification
- Refrain from cell phone use during appointment

In Adulthood – pediatrician or “adult doctor”

- **Advantage of pediatrician**
  - Familiar with you and you with them
  - Familiar with Rett syndrome

- **Advantage of “adult doctor”**
  - Familiar with non-Rett syndrome health issues of adulthood

- **Transitioning**
  - May be required by pediatrician or insurance
  - Controlled transitions are better

Specialists whom you might encounter

- Neurologist – seizures
- Geneticist – testing, diagnosis, and counseling
- Orthopedist – scoliosis, contractures, dystonia, hip dysplasia
- Gastroenterologist – reflux, constipation, nutrition, gallbladder, extreme gas
- Pulmonologist – sleep apnea, asthma
- Cardiologist – prolonged QT interval
- Physiatrist/Physical Medicine/Rehab – spasticity treatment and mobility

WHEN TO CALL A DOCTOR
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
When to go the ER

- High fever
- Difficulty breathing
- Seizure lasting more than 10 minutes
- Unresponsiveness
- Severe pain
- Stomach distended, hard and unremitting
- Limb is swollen or red

Surgical considerations

- Pre and post op – Propofol paper*
  - Dispelling Rett folklore
  - May require less anesthesia
  - May take longer to awaken post op
  - Breath holding associated with RTT goes away in sleep

- Discharge
  - Stay within your comfort zone for discharge
    - Bleeding management
    - Fluid management
    - Pain management

* NM Tofil et al. *J Child Neurol* 2006;21:210-213
VACCINATIONS

Teaching without Preaching
Why *should* we vaccinate?

- **Vaccinations work**
  - Used for over 50 years
  - Save lives and decrease disease

- **Vaccinations are safe**
  - Monitored by unbiased health agencies
  - No credible evidence of linkage to autism

- **Vaccinations are necessary**
Why should we *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
Wakefield’s “autism link” study
Parents’ responsibilities about vaccine decisions

- To make INFORMED decisions

- Listen with a discriminating ear to those you trust and who understand the facts
  - not to the sensational media
  - not to those who will gain personally
  - not to someone who doesn’t understand the “science”
Providers’ responsibilities

- Provide *accurate* information about vaccines and the diseases they are preventing
- Provide *accurate* information about risks of vaccinating versus not vaccinating
- Give you the *opportunity* to ask questions about your concerns
For more information about vaccines

- 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
Remember to complete your gallbladder study questionnaire!!
The end!

NO TRUCKS OR BUSES ON JANE LANE
Finding the care you need