Pediatric and Adult Preventive Care in Rett Syndrome, MECP2 Duplications and Rett-related Disorders

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OUTLINE

- Finding the care you need
- Child and adult preventive care
- When to call the doctor and surgery considerations
- Vaccinations
Finding the care you need
Thou shalt not keep a pediatrician into adulthood

*Small person ≠ child*

As they may not
- have admitting privileges to adult hospital
- be equipped to care for adult problems
- know adult subspecialists
- understand Medicare
- accept your “adult” insurance
And avoid the temptation to only see the primary when there is illness

- So that providers may know them in a ‘healthy state’ to recognize ‘unhealthy’ one

- Screenings may detect a unknown problem
And *remember* that not all health issues are related to Rett syndrome or related disorders

- trust that adult providers know their business and be willing to educate them when necessary
- don’t overwhelm them with “syndrome care” information if not the primary problem
- recognize that adult providers’ approach and personalities may differ from pediatricians
Finding a good healthcare provider

- Be proactive about the transition to adult care
- Ask other families and the pediatrician
- Consider your own provider
- Med/Peds and Nurse Practitioner options
- Syndrome specialists are great resource
- Beware of having unrealistic expectations.
  - It’s okay to educate
**Keeping a good provider**

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

List of ‘ologists

- Neurologist – seizures, movement disorders, dystonias, sleep issues
- Orthopedist – scoliosis, contractures, dystonia, hip dysplasia
- Gastroenterologist – reflux, constipation, nutrition and gastrostomy eval, gallbladder eval, extreme gas
- Pulmonologist – sleep apnea, asthma, pneumonia
- Cardiologist – EKG interp/ prolonged QT interval
- Physiatrist/PMR – spasticity treatment and mobility
- Endocrinologist – diabetes, bone health, puberty abn
- Nephrologist – kidney stones, urinary retention
- Urologist – ‘plumbing’ care
- Gynecologist – ‘female’ 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 and related disorder
  - EKG screening at diagnosis, then annually
  - EEG screening at diagnosis, then as needed*
  - 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
  - Labs: CBC, Vitamin D, lipid profile, metabolic profile
Adult Preventive Care

- **Cancer screening**
  - Skin
  - Breast exams - Monthly “at home” exam; mammography
  - Gynecological exams -
    - Bimanual versus speculum and PAP as needed.
    - Individualized GYN care based on need and symptoms
    - Exam may require sedation

- **Dental**

- **Vision and hearing**
Adult Preventive Care

- Labs (minimum)
  - CBC, vitamin D, lipid profile, metabolic profile

- Immunizations
  - Per physicians’ recommendations

- Infectious disease screening
  - Hepatitis C – if high risk
  - TB – if high risk

- Other recommended screenings
  - Diabetes type 2
  - Hypertension
Adult Preventive Care

- Rett syndrome and related disorders considerations
  - EKG screening annually
  - EEG as prescribed
  - Nutritional assessments including BMI
  - Physical therapies
  - Bone mineralization by xray or DXA as indicated
  - Mental health – depression
WHEN TO CALL IN MEDICAL REINFORCEMENTS
Call when there is:

- New onset fever greater than 101°
- 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
- ER is not a primary care clinic!

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

* NM Tofil et al. *J Child Neurol* 2006;21:210-213
VACCINATIONS
Why should you 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
  - Diseases they guard against are deadly
Why should you 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
Early report

Ileal-lymphoid-nodular hyperplasia, non-specific colitis, and pervasive developmental disorder in children

A J Wakefield, S H Murch, A Anthony, J Linnell, D M Casson, M Maik, M Berelowitz, A P Dhillo, M A Thomson, P Harvey, A Valentine, S E Davies, J A Walker-Smith

Summary

Background. We investigated a consecutive series of children with chronic enterocolitis and regressive developmental disorder.

Methods. 12 children (mean age 5 years, range 3–11), 10 boys) were referred to a paediatric gastroenterology unit with a history of normal development followed by loss of acquired skills, including language, together with diarrhea and abdominal pain. Children underwent gastrointestinal, neurological, and developmental assessment, and review of developmental records, ileocolonoscopy and biopsy sampling, magnetic-resonance imaging (MRI), electroencephalography (EEG), and lumbar puncture were done under sedation. Salmonella follow-through radiography was done where possible. Biochemical, haematological, and immunological profiles were obtained.

Findings. Onset of behavioural symptoms was associated by the parents with measles, mumps, and rubella vaccination in eight of the 12 children, with measles infection in one child, and otitis media in one. All 12 children had intestinal abnormalities ranging from lymphoid nodular hyperplasia to strictures. Histology showed patchy chronic inflammation in 11 children and reactive intestinal lymphoid hyperplasia in seven, but no granulomas. Eleven children also had included autism (nine), distractibility (nine), loss of language, poor self-care skills, short attention span, and possible postural or vocal abnormalities. There were no focal neurological abnormalities and the EEG tests were normal. Abnormal laboratory results significantly raised urinary methylmalonic acid compared with age-matched controls in five children, but not in the four children, and normal in the remaining eight children.

Intervention. The observation of gastrointestinal symptoms and developmental regression in a group of closely related children, which was generally associated in time with possible environmental triggers. Lancet 1998; 351: 637–41

See Commentary: 'Wakefield's paper: Wakefield's paper'

Inflammatory Bowel Disease Study Group, University Departments of Medicine and Histopathology (A Wakefield, S H Murch, N C Anthony, J Linnell, D M Casson), A P Dhillo, M E Davies, and the University Department of Paediatric Gastroenterology (S H Murch, D M Casson, M Maita, M Maita, M A Thomson, J A Walker-Smith) and the Child and Adolescent Psychiatry (M Berelowitz) and Neurology (P Harvey) and Radiology (A Valentine), Royal Free Hospital and School of Medicine, London NW3 2QG, UK

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Introduction

We saw several children who, after a period of apparent normality, lost acquired skills, including language, together with abdominal pain, diarrhea, and vomiting, and, in some cases, food intolerance. We noted the clinical features and gastrointestinal symptoms of these children.

Patients and methods

12 children, compared, were referred to our department of paediatric gastroenterology with loss of acquired skills and intestinal symptoms. Abdominal, gastrointestinal, and food intolerance were investigated. All children were admitted to the ward usually, accompanied by their parents.

Clinical investigations

These included dietary histories, details of immunisations and vaccine adverse reactions, physical examination of the abdomen, and assessment of the children. In 11 children, the diagnostic procedures obtained by the senior author (J W W). Neurological and psychiatric assessments were done by consultant psychiatrist (D W M) with neuropsychological testing. Developmental assessment included a review of prospective developmental records from the parents, and developmental contacts and general practitioners. Four children were not referred to psychiatric assessment in hospital; all had been assessed professionally elsewhere, so these assessments were used as the basis for their developmental diagnosis.

After bowel preparation, ileocolonoscopy was performed by SHM or MA T under sedation with midazolam and pethidine. Frozen and formalin-fixed mucosal biopsy samples were taken from the terminal ileum, descending, sigmoid, and rectum. The procedure was recorded by video or still images, and compared with images of the previous seven consecutive paediatric colonoscopies (four normal colonoscopies and three children with ulcerative colitis), in which the physician reported normal appearances in the terminal ileum. Barium follow-through radiography was possible in some cases.

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Also under sedation, cerebral magnetic resonance imaging (MRI), electroencephalography (EEG) including visual, brain stem auditory, and sensory-evoked potentials (where possible) were measured.

Laboratory investigations

Thyroid function, serum long-chain fatty acids, and cerebrospinal-fluid lactate were measured to exclude known causes of childhood neurodevelopmental disease. Urinary methylmalonic acid was measured in random urine samples from eight of the 12 children and age-matched normal controls, by a modification of a technique described previously. Chromatograms were scanned digitally on computer, to analyse the methylmalonic acid zones from cases and controls. Urinary methylmalonic-acid concentrations in patients and controls were compared by a bootstrap approach. Urinary creatinine was estimated by routine spectrophotometric assay.

Children were screened for antiendomysial antibodies and boys were screened for fragile-X if this had not been done.
Parents’ responsibilities about vaccine decisions

- To make INFORMED decisions
- Be responsible to those vulnerable
- Listen with a discriminating ear to those you trust and who understand the science
  - 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
QUESTIONS? jlane@uab.edu

NO TRUCKS OR BUSES ON JANE LANE