

Day to Day: Quality of Life Issues

Judy Barrish, B.S.N., R.N.

Patient Manager and Project Coordinator

The Blue Bird Circle Rett Center

Department of Pediatrics

Baylor College of Medicine

Texas Children's Hospital



Texas Children's Hospital



Rett Syndrome: Diagnosis

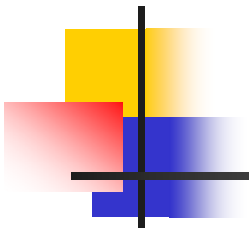
- Remains a Clinical Diagnosis
- Of Those With Classic RTT, 95% Have an Identifiable MECP2 Mutation (Xq28)
- For Atypical RTT, 65% Have an Identifiable MECP2 Mutation
- Analysis
 - Sequencing analysis
 - Southern analysis for larger mutations (Deletion/Insertion)



Rett Syndrome: Comprehensive Care

THE TEAM APPROACH

- Parents
- Pediatrician, Family Practice, Internist
- Specialists: Neurologist, GI/Nutrition, PM&R, Orthopedic, Geneticist, Cardiologist, Surgeon, Anesthesiologist, Dentist, etc.
- Teacher, therapist, school nurse



Who Should
Have the
MECP2 Test?



RETT SYNDROME: GENETIC TESTING

- GIRLS MEETING THE DIAGNOSTIC CRITERIA FOR CLASSIC RTT OR VARIANT FORM OF RTT
- YOUNG GIRLS (FIRST 1-2 YEARS OF LIFE) WITHOUT ALL CRITERIA BUT WITH DEVELOPMENTAL DELAYS OF UNKNOWN CAUSE, ESPECIALLY IF REGRESSIONS IN DEVELOPMENT, STEREOTYPIC REPETITIVE HAND MOVEMENTS OR DECELERATION OF HEAD SIZE GROWTH



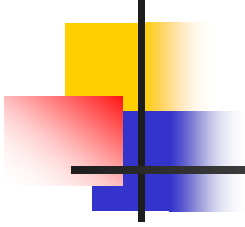
RETT SYNDROME: GENETIC TESTING

- MOTHERS WITH DAUGHTERS WITH RTT AND *MECP2* MUTATION
- MOTHERS WHO TEST NEGATIVE AND THEIR DAUGHTERS TEST POSITIVE SHOULD HAVE TESTING DURING THEIR PREGNANCIES



RETT SYNDROME: GENETIC TESTING

- BOYS WITH NEUROLOGICAL DISORDERS THAT INCLUDE DEVELOPMENTAL DELAYS, MOTOR PROBLEMS LIKE TREMORS AND BEHAVIORAL ABNORMALITIES OF UNKNOWN CAUSE ESPECIALLY IF OTHER MALE RELATIVES WITH SIMILAR PROBLEMS
- NOT INDICATED FOR NORMAL FATHERS OR BROTHERS



Why Does My Daughter Scream?



Why Texans are SO tough...



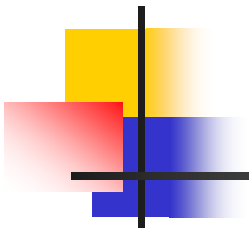
Rett Syndrome: Screaming/Crying Episodes

- A Form of Communication
- The Usual Suspects
 - Gastro-esophageal Reflux/Esophagitis
 - Constipation
 - Bone Fracture



Rett Syndrome: Screaming/Crying Episodes

- The Usual Suspects (continued)
 - Gall Stones
 - Infections
 - Urinary tract infections
 - Sinus infections
 - Hungry
 - Dental Problem
 - Menstrual Discomfort
 - Angry, Sad (Depression), Anxiety



My Daughter has
Osteopenia, Does
She Need Calcium?



Rett Syndrome: Osteopenia

- Osteopenia occurs at an early age
- Rett girls are at increased fracture risk
- Bone mineral loss greater in Rett than in other neurological disabilities



Rett Syndrome: Bones

- Dietary sources of Calcium
- Vitamin D – achieved with 10 minutes exposure to sun per day
- Promote Weight Bearing

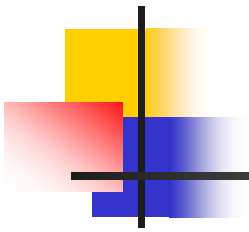


Does Our Daughter Need an EEG?



What's the Question?

- Abnormal in almost all girls with Rett syndrome after 2 years of age
- Indicates slowing EEG – not unexpected
- Includes markers indicating risk of seizures
- Not all behaviors are epileptic seizures
- Video EEG helps differentiate between Rett behaviors and seizures



In What Stage is
My Daughter?
Will She
Deteriorate?



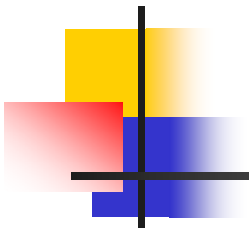
Rett Syndrome: Clinical Stages

I: Pre-Regression ("Delays")

II: Regression ("Rapid")

III: Stable ("Improvement")

IV: Motor Deterioration – not necessarily going to die at this stage



How Long Will
My Daughter
Live?



SURVIVAL STUDY: GIRLS

“BETTER THAN EXPECTED”

IMPACT OF EARLY DIAGNOSIS AND
AGGRESSIVE INTERVENTIONS

CAUSES OF MORBIDITY/RISK FACTORS:

- SUDDEN UNEXPECTED DEATH: LONG QT_c
- SEPSIS/PNEUMONIA
- SWALLOWING DYSFUNCTION
- NUTRITION



Rett Syndrome: Seizures

- Most are brief and do not cause injury or death. Most injuries and/or deaths are related to seizures which occur in a bath or other at risk activities.
- Prolonged seizures (a single episode lasting greater than an hour) are cause for concern of injury to brain or heart.



So What Can We
Do?



QUALITY OF LIFE FOR YOUR DAUGHTER

- **Nutrition:**
 - Monitor her weight
 - Intervention
- **PT/OT/Alternative communication**
- **Aggressive management of scoliosis**
- **Appropriate management of seizures**
- **Hygiene:**
 - Birth control, time training
- **Guardianship**
- **Involvement in Family Life:**
 - Residential care v.s. Home Care



QUALITY OF LIFE FOR YOU

- Parent Health
- Parent Time Out
- Have a Plan