



*The Research for*  
*Rett Syndrome Foundation, Inc.* file

APR 26 1995

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Ms. Carol Rascoe  
Director of Domestic Policy  
The White House  
Washington, D.C. 20500

April 24, 1995

Dear Carol,

I was so honored, as was my daughter Gaynor, to meet you while visiting my cousin, Gaynor McCown in Washington recently. She keeps us abreast of what a magnificent job you are doing at The White House that we feel especially privileged to see your efforts being carried out first hand.

Our trip was educational, enjoyable and productive! My daughter's introduction to Capitol Hill was particularly moving as Congressman Callahan testified in support of bio-medical research in Rett Syndrome to the Labor, HHS Appropriations Subcommittee. Not only was Chairman Porter very receptive to the appeal, other members particularly interested were Nancy Pelosi and Nita Lowey. Congressman Hoyer has also been very supportive of Rett research in years past as he's attended church with a grown girl now with Rett and witnessed her tragic regression since she was very young. This interest results from the harsh realization that Rett is one of only four diseases affecting only females, to date and that this not so rare invisible enemy has no regard for race, creed, color or political preference for that matter.

While visiting Capitol Hill, we met with the new Washington representative of the National Organization of Rare Diseases, Michael Langan. We then attended the ABC reception for the Peter Jennings special entitled, "Children First". I've enclosed the press kit from the event for your further information on the commitment to children, and issues facing parents in America today by the ABC Television Network.

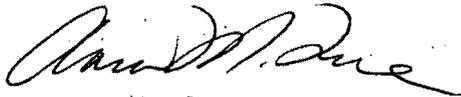
After a day meeting with NIH directors, e.g. Drs. Alexander (NICHD) and Hall (NINDS), and an appeal to the Joseph P. Kennedy, Jr. Foundation, Gaynor McCown was a most gracious hostess showing us the OEGB and the West Wing of The White House.

April 24, 1995

As I'm sure you know my heart well also having a mentally retarded child, please allow me this opportunity to enclose some literature for your further information on Rett Syndrome and the Research for Rett Foundation. Many thanks again for taking time to meet us, it was a real honor for us and truly the highlight of our trip.

May God bless you as you continue your very dedicated efforts on behalf of the children and families in America today.

Sincerely,

A handwritten signature in cursive script, appearing to read "Anna M. Luce".

Anna M. Luce

enc.

## Stage 4 — Late Motor Deterioration

Further deterioration of motor activities occurs, but alertness, interaction and eye contact may remain constant. This stage may last a decade or more.

### HOPE AND PROGRESS

Worldwide collaboration of researchers suggests Rett Syndrome is an X-linked genetic disease. At Baylor College of Medicine, Houston, Texas and other medical meccas, the search is on for a biological marker. Locating this marker will enable researchers to develop a **clinical test to reveal Rett Syndrome**. From there, treatments can be developed to stabilize progression of Rett symptoms and — eventually — cure Rett Syndrome.



Courtney  
Bennington

### FOR NOW

Play therapy, speech therapy, hydro therapy, music therapy and nutrition are the best measures for treating and coping with Rett Syndrome at this time.

It is important to remember that girls with Rett can learn and assessments can be performed around age 4 to determine educational needs.

Each child is different, but all share a common need for love and affection.

### THE NEXT STEPS: WHAT YOU CAN DO

Education and increased awareness are key to successfully defeating Rett Syndrome. There are many things interested individuals can do to further this cause. Lobbying efforts for research are



## RESEARCH FOR RETT, INC.

For more information please write or call:

RFR, Inc.  
P.O. Box 50347  
Mobile, Alabama 36605  
Fax (205) 342-5114  
1-800-422-RETT

Simply sharing information about Rett Syndrome with a friend helps increase awareness, too. The story speaks for itself. And by passing it on, you may help little girls who are losing that ability.

Please make you tax deductible  
donation payable to:  
RFR, Inc.  
P.O. Box 50347  
Mobile, Alabama 36605  
Fax (205) 342-5114  
1-800-422-RETT

RFR, Inc. hosts Medical Research Symposia for international Neuroscientists and Neuropathologists at which significant findings and information is exchanged. These gatherings form the impetus for critical research for Rett Syndrome.

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Special thanks to the following for producing this brochure: Leigh Brown Photography • Colony Photo • Dillard

## RETT SYNDROME

What is it?  
Know the Facts.



girls. Development appears normal up to about six to eighteen months of age. The child with Rett syndrome typically sits independently and finger feeds at the expected time. Some children start the use of single words and word combinations. Many children begin independent walking within the normal range, while others show significant delay or inability to walk independently. A period of stagnation or regression follows, during which the child loses purposeful use of the hands, replacing it with repetitive hand movements which become almost constant while awake. Intellectual development appears to be severely delayed, but true intelligence is difficult to measure. Many children are misdiagnosed with autism or cerebral palsy.

There is no known cause, treatment or cure for Rett syndrome.

For more information, contact The Research for Rett Syndrome Foundation, Inc., P.O. Box 50347, Mobile, AL 36605, 1-800-422-RETT, FAX (205) 342-5114 or (205) 478-8472

*What if . . . your life began seemingly healthy and full of promise . . . your parents imagined the many wonderful things you would accomplish . . . you grew and began learning . . . everything looked perfectly fine... What if one day you found how to walk . . . you noticed the flowers, the birds, the many people who love you . . . you met a "favorite doll" . . . you discovered your voice . . . What if you gradually began to lose your ability to express it all?*

**THE REALITY OF RETT SYNDROME**

For children with Rett Syndrome this scenario isn't pretend. It's very real. Rett Syndrome is a progressive neurological disorder that causes the brain to lose what it has previously learned. To date, its victims are always female.

Surprisingly, Rett Syndrome is **not** rare. It is the **most common** cause of **profound** mental retardation in **females**. Down's Syndrome is recognized as the chief cause of mental retardation in children as a whole population, but that statistic includes both girls **and** boys. Rett limits itself to little girls.

In fact, current data suggests approximately one in every 10,000 to 15,000 live female births is a Rett girl. But this estimate is probably low, because the statistic represents **only** those children who are diagnosed and reported.

Unfortunately, Rett Syndrome is commonly **misdiagnosed** and **misunderstood**. Not until 1983 was it recognized in the American medical community. But even in the '90's, Rett patients are incorrectly diagnosed, wrongly "labeled" and sadly dismissed as "retarded" for unknown reasons."

**RETT: THE BEGINNING**

The roots of this disorder trace to 15th century Sweden; however, in the early 1960's, Dr. Andreas Rett, a Vienna neurologist, discovered and named what today is known as Rett Syndrome. His work continues throughout Europe, where families who have lost girls to Rett willingly participate in research studies. In America, awareness and acceptance of Rett Syndrome is lower than the rest of the world; however, as American awareness increases, research efforts are accelerating. America raises more government-based funding than elsewhere which allows scientists greater flexibility to respond to various needs. International medical cooperation coupled with public education will be powerful weapons in the fight against Rett Syndrome. Knowledge, like the persevering spirit of Andreas Rett, **can** overcome ignorance and reluctance to participate in research.

**RETT: THE SYMPTOMS AND STAGES**

Because Rett Syndrome is relatively "new" to America, very few parents or physicians know how to recognize it. For about the first year of life, development seems normal. Some time around 18 months, the first of four Rett Syndrome stages appears. Signs and stages include:

**Stage 1 — Early Onset Stagnation**  
"Normal" developmental progress such as speech and coordination slows/levels off.

**Stage 2 — Rapid Developmental Regression**  
Several months following stage one, loss of useful hand function and loss of interaction, socialization occurs; handwringing/handwashing begins. This stage lasts about one year.

**Stage 3 — Pseudostationary**  
Eye contact and social interaction may actually improve, but hand movements and breathing irregularities increase. Motor activity gradually decreases, abnormal postures appear. Walking skills decline or stop completely. This stage can last 10 to 20 years.



Anne Stuart Grantham



Annie Leeds



Stephanie Atwell



Aureal Tucker



Lorissa Coulter

*These little girls have Rett Syndrome.*

*Actually, Rett Syndrome has these little girls.*

Vice President/Treasurer,  
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RETT SYNDROME is a progressive neurological disorder affecting only females to date. The Research for Rett Syndrome Foundation, Inc. is the only organization which exists for the sole purpose of raising funds for critical ongoing medical research in Rett Syndrome. The Research for Rett Syndrome Foundation, Inc. is the only organization that exists specifically for funding critical ongoing medical research in Rett Syndrome.



# FACT SHEET

## The Research for Rett Foundation, Inc.

**Mission:** The Research for Rett Foundation, Inc. is a non-profit organization which exists for the purpose of raising funds for critical on-going medical research, hosting medical research symposiums and funding grant applications, expanding public awareness and encouraging RS research to be a high priority with the National Institute of Child Health and Human Development (NICHD) and the National Institute of Neurological Disorders and Stroke (NINDS) within the National Institute of Health (NIH).

- I. **RETT SYNDROME:** Neurological disorder affecting only females, to date. Commonly misdiagnosed as autism or cerebral palsy. **One of four diseases affecting only females.**
- II. **PREVALENCE:**
- A. **1 in 10,000 - 15,000 female births** - estimated by IRSA (International Rett Syndrome Association). Number said to be greater due to cases not reported and/or misdiagnosed. **8,000 - 10,000 estimated undiagnosed in the U.S.**
  - B. **LEADING CAUSE FOR PROFOUND MENTAL RETARDATION IN FEMALES: PERHAPS EVEN GREATER INCIDENCE THAN IN FEMALES WITH DOWN'S SYNDROME.**
  - C. **Greater** incidence than PKU (metabolic disorder for which all infants are screened at birth.)
  - D. Results in **PROFOUND** mental and physical retardation.
  - E. Enormous impact on society with regards to care; institutions commonly needed and life expectancy - 20-40 years.
- III. **IMMEDIATE GOAL:** To increase earmarked federal funding for further research into Rett Syndrome.
- A. **Progress being made at Baylor College of Medicine in Houston, Tx: and the Kennedy-Krieger Institute (Johns-Hopkins) Baltimore, MD**
  - B. **Important Information Generated:**
    1. Natural History
    2. Clinical Neurophysiology
    3. Respiratory Function
    4. Nutritional Requirements and Energy Utilization
    5. Growth Characteristics
    6. Epidemiology in Texas, Survival data in U.S.
    7. Neurotransmitter Analyses in Spinal Fluid and Brain
    8. Critical Neuropathology studies, Molecular Genetic studies
    9. Drug Trial with Opiate Antagonist Naltrexone
    10. Study of Scoliosis and Effect of Treatment Strategies.
  - C. **Impact Research has on Other Diseases:**
    1. Neuropathology studies show new concepts in developmental neurobiology at human level; recognition of impact of developmental arrest at critical stage of brain development.
    2. Molecular genetic studies have led to development of multiple new probes or markers for X chromosome which could be critical for identifying specific X-linked diseases.
    3. Dr. Zoghbi (Baylor) has developed new insights in Aicardi/Goltz syndromes as a result of X chromosome markers she has defined as an example.
    4. Techniques and strategies used are not limited to Rett Syndrome-generalizable to other X-linked diseases.

Submitted by: The Research for Rett Foundation, Inc. April 20, 1995

P.O. Box 50347 Mobile, AL 36605 or 1-800-422-RETT

For more info., contact Anna M. Luce 205-342-5114 or Pat Cofer at 1-800-422-RETT