Gastro Intestinal Manifestations in Rett Syndrome

CELEBRATING THE TORCH WITHIN
IRSF 25TH ANNUAL EDUCATION & AWARENESS CONFERENCE

Anil Darbari, MD

Pediatric Gastroenterologist
Kennedy Krieger Institute

Assistant Professor of Pediatrics

The Johns Hopkins University, Baltimore, MD

Gastro Intestinal Manifestations in Rett Syndrome

Anil Darbari, MD

Director GI Motility Center
Medical Director Pediatric Feeding Disorders Program

CELEBRATING THE TORCH WITHIN
IRSF 25TH ANNUAL EDUCATION & AWARENESS CONFERENCE

OUTLINE OF PRESENTATION

Introduction
- Rett Syndrome- Overview
- Gastrointestinal Manifestations
- Clinical Presentations

Diagnostic Testing
- Gastroesophageal Reflux
- Swallowing Problems
- Feeding Problems

Management Options
- Gastroesophageal Reflux
- Swallowing Problems
- Feeding Problems

Conclusion

RS Clinical Patients (N = 278)

Rett Syndrome: Clinical Presentation

Rett Syndrome Fact Sheet

Four typical stages of the disorder

- Stage I: Early Onset
- Stage II: Rapid Destructive
- Stage III: Plateau or pseudo-stationary
- Stage IV: Late Motor Deterioration

Rett Syndrome Staging System

Stage I: early onset stagnation	Onset age 6 mo to 1.5 yr
Duration: wks to mos	Developmental progress delayed
	Developmental pattern still not significantly abnormal
Stage II: developmental regression	Onset age 1-3 or 4 yr
Duration wks to mos, possibly 1 yr	Loss of acquired skills/communications
	Mental deficiency appears
Stage III: pseudostationary period	Onset: after passing stage II
Duration: yrs to decades	Some communicative restitution
Stage IV: late motor deterioration	Onset: when stage III ambulation ceases
Duration: decades	Complete wheelchair dependency
	Severe disability: wasting and distal distortion

PERSPECTIVES IN PRACTICE

Eating difficulties in girls with Rett syndrome compared with other developmental disabilities

JANET SUGARMAN ISAACS, PhD, RD; MARIANNE MURDOCK, PhD, RD; JANE LANE, RN; ALAN K. PERCY, MD

Clinical Features	Possible nutritional implications
Growth Failure	Decline in growth percentiles over time for weight, stature and head size
Initial hypotonia	If muscles of face and neck are affected, there may be decreased texture tolerance
Abnormal breathing	Likely to interfere with eating, because swallowing takes place during expiration
Seizures	Medication side effects such as sedation or nausea may change appetite or meal pattern
Ambulation, limited or lost	Unclear. High energy needs do not appear related to overall activity
Movement disorder	May increase energy requirements
Scoliosis	Generally no nutritional impact, unless related to a change in GI function- like gastric emptying
Constipation	May decrease appetite if unmanaged. Food sources of dietary fiber are not effective, stimulants more effective
Self-abuse	No effect, unless wound healing is substantial or chronic

DISABILITY AND REHABILITATION, 2001 ; VOL. 23, NO. 3/4, 118–128

Growth and nutrition in Rett syndrome

SHEENA REILLY† and HILARY CASS‡

† Faculty of Health Sciences, La Trobe University and the Royal Children's Hospital, Melbourne, Australia
‡ Great Ormond Street Hospital London, UK

Feeding Problems in 60 females with Rett Syndrome		
Oral stage: oral motor dysfunction	N=25	41%
Oral-pharyngeal stage	N=12	20%
Esophageal stage	N=9	15%

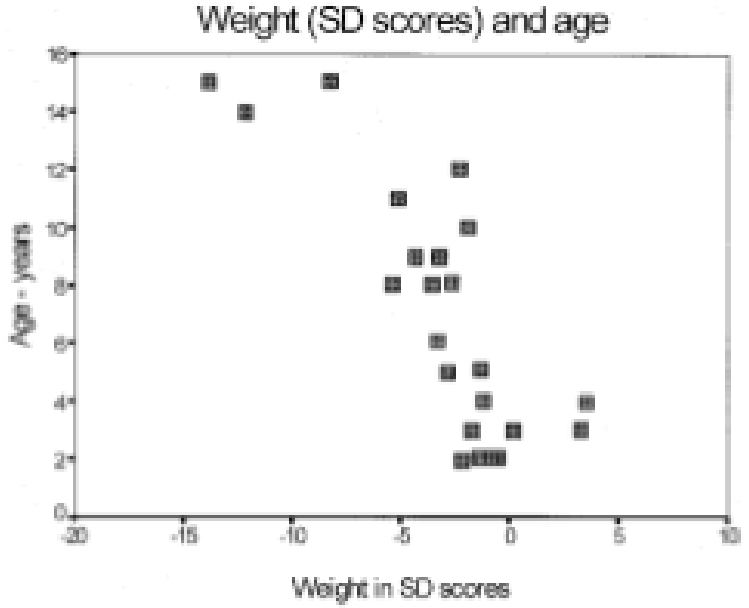


Figure 2 Weight (SD scores) for a subsample of girls (n = 27) correlated with age.

Conclusions

GI Manifestations in Rett Syndrome

Feeding Problems

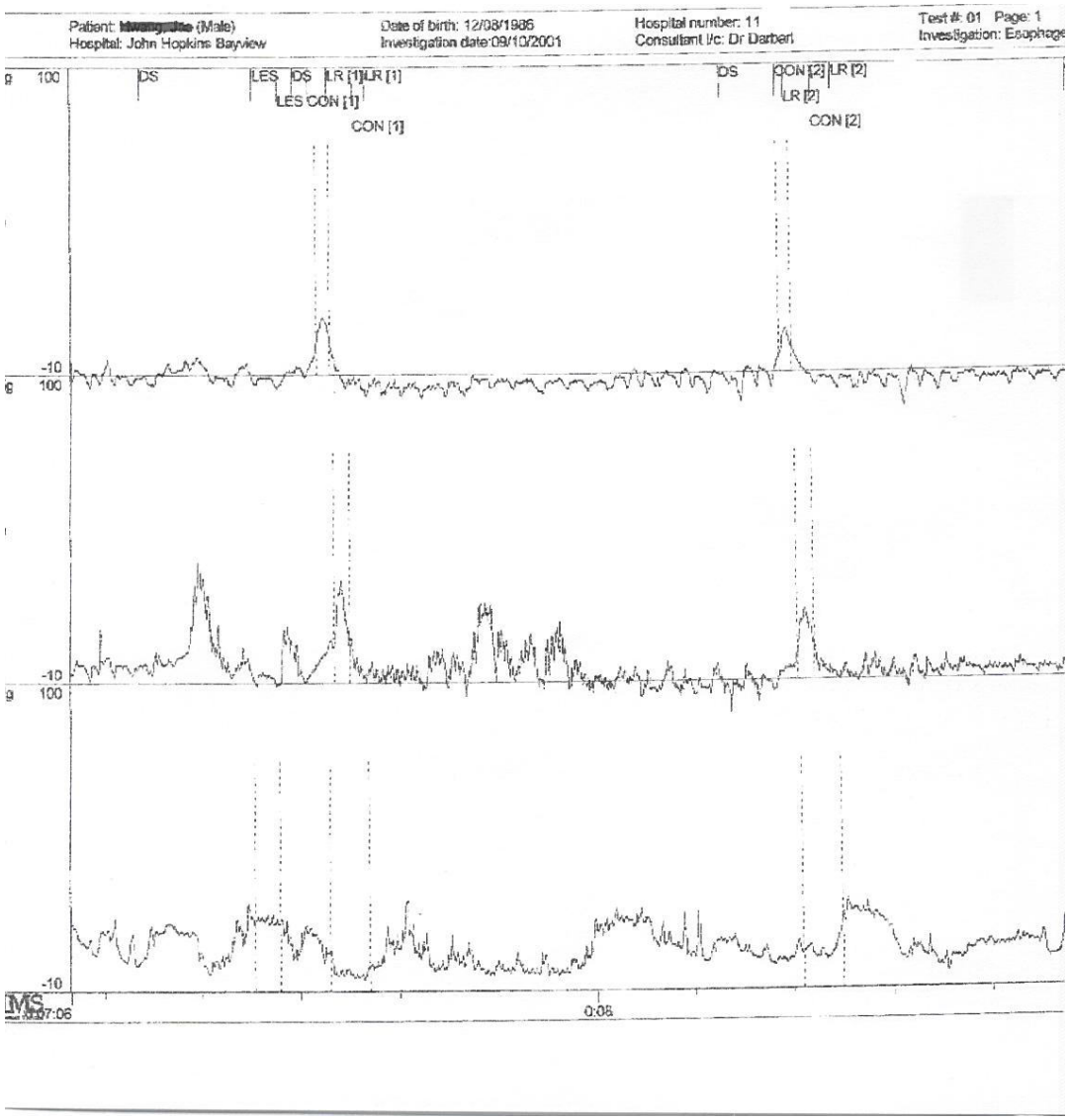
- Long meal times
- Difficulty in chewing
- Difficulty in swallowing

Failure to Thrive

- Feeding problems
- Swallow dysfunction
- High activity levelss

Gastroesophageal Reflux

Constipation



Nutritional Goals

Correct Weight Loss

Replete nutritional deficiencies

Support normal growth for age

What is the best approach to nutrition support?

Support oral feeds

Multidisciplinary programs

medical, oral motor, nutrition, and behavioral support for feeding

Tube feeds

Gastric tubes- NG tubes, G tube

Intestinal tubes- NJ tubes, G-J tubes

Parenteral Nutrition

Pathophysiology of GE Reflux

Increased Frequency of Reflux episodes

- LES Hypotonia
- Defective perisphincteric support
- Transient LES Relaxations
- Increased Gastric pressure
- Increased Gastric volume

Decreased Esophageal Clearance

- Gravity effects
- Defective salivation
- Peristaltic dysfunction or infrequent swallowing