

# Coping With Rett Syndrome

A debilitating disorder that strikes only girls, Rett Syndrome challenges physicians, patients, and families.

One symptom of Rett Syndrome is profound growth failure. Pediatrician and nutritionist Kathleen J. Motil with the USDA-ARS Children's Nutrition Research Center (CNRC) in Houston, Texas, is finding innovative ways to promote adequate nutrition, improve health, and enhance growth for some of these patients.

Rett Syndrome occurs in 1 out of 23,000 live births. The onset is baffling: A healthy, active infant gradually stops developing normally. Typically, she regresses, losing her speech and walking skills, as well as the ability to play with toys. Repetitive hand-wringing and hand-washing movements are common, as are breathing abnormalities. Growth failure and muscle wasting may occur as early as 1 year of age. Motil and others have done studies suggesting this poor growth and wasting may be linked in part to the girls' need for special nutritional care.

Andreas Rett, the medical doctor who first described this neurodevelopmental disease in 1966, noted the symptoms of wasting and slowed growth. Growth retardation is one of the factors supporting the diagnosis of Rett Syndrome. Other researchers, including Motil's colleagues, pediatric neurologist Daniel G. Glaze and nurse practitioner Rebecca J. Schultz, have reported in medical journals a deceleration in the rate of gain in head circumference, height, and weight.

Studies in Norway, England, and the United States have suggested nutrition and eating difficulties might be a possible cause of these decreased rates of growth. Indeed, "Part of the problem is that these girls frequently have oromotor dysfunction," says Motil. "They don't chew or swallow properly, and their dietary intake is inadequate to support normal growth."

Motil has had some success fitting her patients with a gastrostomy button, a surgically implanted device that allows nutrients to be delivered into the body while the girls sleep. One 7-year-old patient increased her weight from 31 pounds to 48 in a year. She gained enough strength to sit up by herself for the first time.

While that sounds like good news, an average 7-year-old weighs about 60 pounds.

In addition to helping patients, Motil is doing research to find

out why these girls aren't growing and have less muscle mass than their healthy counterparts.

Part of her research involves comparing the metabolism and physiology of girls with Rett Syndrome to those of healthy girls. For these experiments, small volunteer groups of girls with and without Rett Syndrome spend brief stays at the General Clinical Research Center at Texas Children's Hospital in Houston and undergo tests at the neighboring CNRC.

Motil wondered whether the repetitive arm, hand, leg, and body motions contributed to the development of malnutrition by burning calories that would be otherwise used for growth. Research she published in the February 1998 issue of *Journal of Pediatrics* showed that repetitive motions were not an energy drain.

But Motil noted that there were metabolic differences between the girls with Rett Syndrome and healthy girls. When sleeping or resting quietly, those with Rett syndrome had total body metabolic rates 23 percent lower than normal.

"The lower metabolic rate was caused by a lower lean-body mass, or less muscle mass," said Motil. "The lower body mass may be related to a lower dietary intake."

Although the energy balance of calories consumed, minus calories used, was positive in girls with Rett Syndrome, it was lower than that of age-matched, healthy girls. It could be that a subtle, long-running, energy-deficit diet hampered nutrition and growth.

In a more recent study published as an abstract in *Pediatric Research*, Motil added to evidence that giving girls with Rett's syndrome a liquid nutritional supplement via the gastrostomy button was helpful. The test showed it increased body weight and reversed the downward trend of poor growth in height.

"Nevertheless, we found that the supplements increased body fat much more than lean body mass, even with the presumably adequate protein and energy intake," said Motil. "Our next task is to understand why we are unable to improve muscle mass to the same extent as body fat."—By **Jill Lee**, ARS.

This research is part of the ARS National Program on Nutrient Requirements, Food Composition, and Intake described on the World Wide Web at <http://www.nps.ars.usda.gov/programs/107s2.htm>.

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