

## **Oral Calcium Supplementation to Treat Osteopenia in Rett Syndrome**

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### **What do we know?**

Dietary factors may play an important role in bone health in Rett syndrome (RTT) and may be helpful in treating osteopenia (bone mineral loss), and bone fractures which frequently occur in girls and women with RTT. From an initial study we carried out, in which nearly 1,000 families responded to a survey questionnaire, we know that 30% of girls and women with RTT have bone fractures by the time they reach 20 to 30 years of age. In a follow up cross-sectional study on the natural history osteopenia, we found that bone mineral deficits occur in nearly 50% of females with RTT and these deficits increase with advancing age in individuals with RTT.

Preliminary data from our IRSF funded study titled "Randomized, Placebo-controlled Trial of Oral Calcium Supplementation for Osteopenia in Girls and Women with Rett Syndrome", suggests that dietary calcium and vitamin D may be associated with the bone mineral status of girls and women with RTT. Both nutrients work together to support bone health; calcium is known to be important for the structural integrity of bones and vitamin D is important in facilitating calcium absorption from the gastrointestinal tract for bone use. From our natural history study, we know that girls and women with RTT consume, on average, only one or two glasses of milk daily -an amount that does not fully support the body's needs for bone health. We also know that approximately one-fifth of these individuals have vitamin D deficiency when measured by examining blood serum levels of 25-hydroxyvitamin D. Currently, every effort is being made to ensure adequate dietary calcium and vitamin D intake in girls and women with RTT.

### **What do we want to know?**

In order to improve bone health outcomes, we want to know if a dietary calcium supplement will halt the progression of bone mineral deficits and improve bone mineral deposition in girls and women with RTT.

### **Where are we going?**

To address our question, we are conducting a research study that will measure the change in bone mineralization after one year of dietary calcium supplementation in girls and women with RTT. We are asking families whose daughters are affected with RTT to consider their daughters' participation in our study. In our study, we will give an oral calcium or placebo (sugar substitute) supplement in a random manner (like flipping a coin) to 52 girls and women with RTT for one year. We will measure bone mineral content by dual energy x-ray absorptiometry (x-ray scanner) before and one year after using the calcium or placebo supplement. We will obtain blood and urine samples to measure markers of bone calcium metabolism. We will monitor the safety of the calcium or placebo supplement by reviewing 3-day food records and measuring the amount of supplement that was taken by each individual. We will obtain blood samples to monitor the safety of calcium supplementation with respect to kidney function. The information that we obtain from this study is important because we need to know if supplemental dietary calcium treatment will improve bone mineralization in girls and women with RTT, and ultimately, prevent bone fractures in these individuals.

### **Where is the study being done?**

The study is being conducted at the Texas Children's Hospital General Clinical Research Center, Houston, TX. The study requires the participant to stay overnight in the General Clinical Research Center. The facility can accommodate the parent's overnight stay as well. Parking and a small stipend are provided.

### **How can we participate?**

Contact Judy Barrish, R.N., research study coordinator, for enrollment information or questions at 832-822-RETT.