

## Looking to Adulthood



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Amanda, age 30, with her mom

When I first met a patient with Rett (RTT) syndrome in 1982, I was informed this disorder only occurs with girls and they never live to adolescence. Well, we've come a long way. We do see a few boys who meet the criteria for the diagnosis and have identified their associated mutations. Here at the Blue Bird Circle Rett Center, we see many women in their 20's, several in their 30's, and a few in their 40's. We no longer have a 50 year old, because this summer she turned 66.

Information gathered from the Natural History of Rare Diseases – Rett Syndrome study has demonstrated that it is typical for women with RTT to survive into their 50's. That's the good news. The existing pediatric care facilities, however, are becoming more and more reluctant to provide continued care for these adults. Unfortunately, as these special ladies have become adults, there is scant adult oriented medical care available to them. Between June 2014 to June 2015, 46 adult women (ages 21-47) were seen in our clinic at Texas Children's Hospital. During that same time period, 10 of these ladies were seen in the TCH Emergency Center, 2 were monitored in the Observation Unit, and 17 were admitted to the hospital for 1 or more overnight stays.

Our goal over the past year to 2 years has been to develop an advocacy program designed to facilitate a safe, effective transfer to adult care and to promote a health care program that is easy for the families to assimilate. To this end we have now initiated a pilot program at Texas Children's Hospital to address this specific need. I have proposed a study to develop a smooth transfer of patient care by identifying and developing relationships with adult care practitioners while coordinating resources already available. Aside from the need for a primary care physician, most women with Rett syndrome will require care from specialty services such as Dentistry, Gastroenterology, Physical Medicine & Rehabilitation, Pulmonary medicine, and some with Cardiology.

In addition to identifying appropriate medical care for their daughters, many of these families have no resources for home care assistance or adult daycare programs. Once they have left the public school system, there are no resources for the women to continue physical occupational and communication therapy programs. As these patients age, their parents are faced with the prospect of providing care for their adult daughters while they themselves are facing increasing health and economic burdens.

Discussions with families regarding the transition process are suggested to begin as early as when the children are 11 to 14 years old. It is always important for parents to establish legal guardianship, recommended between the ages of 18-19, before they transition to adult care. I have long offered education programs to our children's schools, therapy programs, nursing schools. We are now extending this offer to medical practices to introduce the offices and medical staff to the world of RTT.

Like so many other special needs populations, such as cystic fibrosis, I look forward to a time when an adult care community for RTT is well-established; when our ladies are known and their needs anticipated. The key is to identify and implement proactive programs now and not wait, hoping for them to evolve.

