



Should I Participate in the Rett Natural History Study

A Rettsyndrome.org Publication

Since coming to Rettsyndrome.org in January of 2012, I have been asked many questions related to Rett syndrome (RTT). The one question that has surfaced in many of my conversations with Rett families is “Should I participate in the Rett Natural History Study?” This is a great question, and each family has to weigh the cost and benefit to them as individuals. However, the benefits of a better understanding of Rett syndrome far outweigh the costs.



To answer this question, Janice Ascano, Paige Nues and Jennifer Endres and I have put our thoughts down for you to think about. From our perspective, there are five compelling reasons to participate in a natural history study in addition to the general benefit it gives to the biomedical research community.

1. A natural history study is the single best avenue to study the hallmarks of RTT and how these hallmarks progress over time. The fact that an isolated population can be followed in depth by the same group of medical specialists will unfold the obvious and subtle traits of RTT. Observing these traits over time gives rise to ways and ideas to address the biology associated with RTT. Since the traits of our Rett patients can differ from patient to patient, these in depth studies start to unfold patterns that otherwise would go unnoticed.



2. Through a natural history study, a significant number of patients are gathered into a single study. This gathering can be the start of a patient registry. Patient registries have become the coin of the realm when starting clinical trials with drug companies and the biotechnology industry. Having an established patient registry is one of the first things a drug company looks for when fielding a new compound. They are always interested in clinical trials that are statistically powered with the numbers of enrolled patients so that the study can withstand the scrutiny of the regulatory agencies.

3. A natural history study provides an environment to establish both tissue and blood repositories. These repositories provide academic and industrial researchers the opportunity to study any syndrome at either a cellular or molecular level. Studies with the repositories will shed light on pathways and screening procedures that otherwise would go untouched.



4. Natural history studies often disclose the best outcome measures for a clinical trial. This cannot be emphasized enough. By studying the small details associated with RTT, the physician scientist become fully aware of what small changes might occur under any given treatment. These outcome measures become the foundation upon which a trial is deemed success or not. Without a natural history study in place defining the progression of RTT, it might be difficult to determine how any experimental drug made a difference in the progress of symptoms in RTT.



About Rettsyndrome.org

Rettsyndrome.org is accelerating research for treatments and a cure for Rett syndrome. As the world’s leading private funder of Rett research, we have funded more than \$36.8 million in peer-reviewed research grants and programs to date. We empower families to make a difference. We are a 501(c)3 organization, earning Charity Navigator’s most prestigious 4-star rating.



5. Natural history studies provide an environment that readies the family for a clinical trial. This cannot be underestimated. The natural history study provides a setting where physician scientists constantly interact with patients and bring the latest understanding of RTT to the families. This fact tunes the families into where trials are and what is being tested. Through these interactions, families see where they might participate in a particular study and understand both the commitment and cost of participating in a clinical trial.

Lastly, a natural history study gives great value to biomedical research in general, providing a foundation for future physician scientists. Natural history studies create an environment where young physician scientists are close to a large group of patients. The natural history study helps build a future health care provider work force that is committed to their condition of interest.



In conclusion, natural history studies have focused our attention on the right medical conditions at the right time. As we move forward with research unraveling the molecular and cellular events associated with Rett syndrome, the role of the natural history will only increase. It will provide the vehicle for us to translate our basic discovery work into new clinical treatments to modify the pathology associated with Rett. We look forward to hearing from you!

Steven Kaminsky, Ph.D., Janice Ascano, Ph.D. Paige Nues and Jennifer Endres
Rettsyndrome.org



Rettsyndrome.org is accelerating research for treatments and a cure for Rett syndrome. As the world's leading private funder of Rett research, we have funded more than \$36.8 million in peer-reviewed research grants and programs to date. We empower families to make a difference. We are a 501(c)3 organization, earning Charity Navigator's most prestigious 4-star rating.