

Announcing: A DNA repository for Rett Syndrome

Since the seminal discovery in 1999 that mutation in the gene encoding *Methyl-CpG binding Protein 2* (*MECP2*) is the causative factor of Rett syndrome (RTT), it has been reported that the majority (95%) of individuals with RTT have some type of *MECP2* mutation. While there are distinct clinical features of RTT, there has been much observation that clinical severity is not due to specific mutations of *MECP2*. Instead, the severity of these clinical features is widely variable in those diagnosed with RTT, and it is this variation that indicates genes other than *MECP2* may contribute to clinical severity.

This month's Investigator Spotlight features physician scientist Dr. Jeffrey Neul of the Baylor College of Medicine and the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital who will create a DNA repository for Rett syndrome. The IRSF Board of Directors has recently voted to fund this important venture that will yield significant discoveries in other underlying genetic components of RTT.

To create the repository, blood will be collected from people enrolled in the Rett Syndrome Natural History study and from the parents of the affected individuals. The blood samples will be de-identified and labeled with a unique identifying study number linked to accumulated clinical research information. DNA will be isolated using standard techniques and stored for qualified investigators to collaborate on genetic testing for specific hypotheses or open-end genetic screening. The overarching goal of this DNA repository will allow for the identification of other genetic causes of RTT and the determination of the role various genetic factors play in modifying the clinical severity in RTT.

For those enrolled in the Rett Syndrome Natural History Study, families will be contacted in the spring to determine their willingness to participate in this important endeavor.