Investigator Spotlight: Huda Y. Zoghbi, MD, Howard Hughes Medical Institute, Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital and Baylor College of Medicine

It is hard to believe that the 7th World Rett Syndrome Congress has come and gone with great success! Now with summer upon us, IRSF continues to move the spotlight to the committed scientists who have made the World Congress an impressive, high quality meeting with outstanding presentations and discussions. This month we are honored to focus on Dr. Huda Zoghbi who had co-chaired the Basic Research Symposium at the World Congress along with Dr. Gail Mandel. Together, they had produced an exciting lineup of speakers who were encouraged to present new, unpublished data in an effort to foster new ideas that will help chart the course for Rett syndrome research.

Dr. Zoghbi is a Howard Hughes Medical Institute (HHMI) Investigator, the Director of the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital, and Professor of the Baylor College of Medicine in Houston, TX. She received her medical degree from Meharry Medical College and completed residency training in pediatrics and neurology at Baylor College of Medicine where she encountered her first Rett syndrome patient in 1983. Dr. Zoghbi was inspired to receive additional research training in the area molecular genetics and upon completion she joined the faculty of Baylor College of Medicine.

In 1999, Dr. Zoghbi and collaborators including research fellow Ruthie Amir made a major breakthrough for Rett syndrome. They had discovered that mutations in MECP2, the gene encoding methyl-CpG-binding protein 2, causes Rett syndrome. The discovery that the Rett-causing gene is on the X chromosome proved beyond any doubt that the mostly sporadic Rett syndrome is a genetic disorder and X-linked—a finding that also helps explain why it is usually found primarily in girls.

Dr. Zoghbi and her laboratory use genetic, behavioral, physiological, and cell biological approaches to explore many neurological disorders including the inherited degenerative balance disorders (spinocerebellar ataxias) in addition to Rett syndrome and the MECP2 Duplication syndrome. Their lab has generated a mouse model for Rett syndrome and mice that overexpress MECP2 at twice the normal levels. Studies of these mice and other models have shown than too little or too much MeCP2 can increase phenotypic severity.

In addition to Dr. Zoghbi's role in this year's World Rett Syndrome Congress, she is working on a project titled "Therapeutic Interventions to Modulate the GABAergic System in Animal Models of Rett Syndrome" that was funded by an ANGEL grant awarded to her in 2011. She has made significant contributions for Rett syndrome research and is dedicated to moving research towards treatments and a cure for Rett syndrome. For her noteworthy and admiring dedication, Dr. Zoghbi was presented with the Circle of Angels Award for Outstanding Research in Rett syndrome in 2009.

What prompted you to begin a career in research?
Meeting patients with Rett syndrome.

What is the single most rewarding aspect of conducting Rett syndrome research?
Knowing that the work will help the girls one of these days.

If you could pick any one symptom of Rett syndrome to prevent or to provide relief for, what would it be?
Communication. If we can restore or preserve verbal communication I know restoring other deficits will be within reach.

What other disease(s) does your research focus on?
Spinocerebellar ataxia type 1; Shank3 disorders.

Provide any other interesting information about yourself or your work that you would like the Rett syndrome community to know about you.
Reading, working out, gourmet cooking (especially with my daughter, a superb dessert chef), and the opera.

For more information on Dr. Zoghbi, please visit:
www.nri.texaschildrens.org/about_nri/leadership/zoghbi.aspx
www.bcm.edu/genetics/?pmid=11053
www.hhmi.org/research/investigators/zoghbi_bio.html

For a list of Dr. Zoghbi's publications, please visit:
www.ncbi.nlm.nih.gov/pubmed?term=zoghbi%20h20JI