

## RSF Spotlight: Aleksandra Djukic, MD, PhD, Tri-State Rett Syndrome Center, Montefiore Medical Center, Albert Einstein College of Medicine

To end this year, it is with great pleasure to feature Dr. Aleksandra Djukic in this installment of the Investigator Spotlight series. Dr. Djukic is the Director of the Rett Syndrome Center at the Montefiore Medical Center and is also appointed as an Associate Professor of Neurology at Albert Einstein College of Medicine of Yeshiva University. She attended the University of Belgrade in Yugoslavia-Serbia for both medical and graduate school, received training as a Pediatric Resident and Neurology Fellow at the Albert Einstein College of Medicine in Bronx, NY, and has an extensive background as a neurologist and neuropsychologist. Dr. Djukic's commitment to Rett syndrome is evident by her vision of developing the Rett Syndrome Center at the Montefiore Medical Center. It was recently announced that the Montefiore Medical Center and Albert Einstein College of Medicine have secured a grant from the National Institute of Neurological Disorders and Stroke (NINDS) to establish a clinical site for the Network for Excellence in Neuroscience Clinical Trials (NeuroNEXT). NeuroNEXT was created to facilitate Phase II clinical trials for rare neurological diseases such as Rett syndrome, which face significant challenges securing funding from industry, as well as with recruiting and retaining participants. These additional funds will support the mission of the Rett Syndrome Center to provide state-of-the-art services for patients with Rett syndrome through intervention, education, and research aimed at effective treatments and cures. The specific research goals of the center entail a systematic and uniform documentation of the cognitive phenotype, development of objective outcome measures, and ultimately, development of the approach to education and communication in the Rett syndrome population.

Dr. Djukic has been a long-time advisor for IRSF as a member of the Scientific Review Board and participates on the International Consortium of Rett Syndrome Clinical Researchers (RettSearch). She has also been awarded two HeART awards for her work in developing techniques using eye tracking technology to examine cognitive ability in RTT girls. This translational research will specifically study the ability of these girls to distinguish different emotional expressions, an important aspect of their social skills. Dr. Djukic has also recently received funding from the NIH to study how much RTT girls understand spoken language. Together, these studies are integral components of her mission "to focus on what girls with RTT can do".

In her spare time, she conceived the "Blue Sky Girls" event during October's Rett syndrome awareness month, where the girls and their families gathered together in different parts of the world at the same time to climb to the top of a set of

magnificent stairs as a symbolic gesture of overcoming the difficulties they face every minute of their lives. Dr Djukic, also endearingly called “Dr. Sasha” by her patients and families, is a highly motivated, passionate clinician scientist who treats girls diagnosed with Rett syndrome, performs research to understand the their nonverbal cognitive abilities, and raises awareness for Rett syndrome. Thank you, Dr. Sasha, for your dedication to Rett syndrome.

**What prompted you to begin a career in research?**

My orientation towards research originates in my curiosity to understand how things happen. Understanding how something as abstract as human thought originates from something very physical, the brain, was the main reason I choose to dedicate my professional life to the field of neurology, and focus my research interest on behavioral neurology.

**What is the single most rewarding aspect of conducting Rett syndrome research?**

As a clinician-scientist, my conviction that clinical and translational research will lead to the substitution of breath holds, anxiety, and pain with smiles in those I deeply care for, and that this will happen in a foreseeable future.

**Identify a potential positive outcome of the research you are conducting that is specific to this proposal.**

Vision and gaze are considered to be the most important ways in which patients with RTT relate to the world. The objective of this pilot study is to assess visual attention and memory of patients with Rett Syndrome in a structured way, using eye gaze and eye tracking technology. I expect this study to provide better insight into intellectual abilities of individuals with Rett syndrome, to allow physicians and educators to develop more appropriate educational strategies, and provide objective outcome measures for scientists for the upcoming treatment trials.

**If you could pick any one symptom of Rett syndrome to prevent or to provide relief for, what would it be?**

Apraxia, because of its devastating impact on both:

- a. Most fundamental biological functions ( feeding, moving)
- b. Most fundamental human functions (speaking).

**What other diseases does your research focus on?**

Krabbe disease, which is another rare genetic disease which affects children.

**Besides your role as principal investigator on this project and as a Rett syndrome investigator, what other roles do you currently hold that are specific to the field of Rett syndrome research?**

IRSF Grant Reviewer, RettSearch member.