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Rettsyndrome.org Funds Research to Provide Hope for Females and Males Affected by MECP2 Mutation.

CINCINNATI, OH – June 3 - Rettsyndrome.org announced their funding of two new research projects today. Jeannie T. Lee, MD, Ph.D. of Massachusetts General Hospital is awarded a two-year ANGEL Grant for $600,000 to focus on reactivating the silent X chromosome for Rett syndrome and Davut Pehlivan, MD of Baylor College of Medicine is awarded a two-year Mentored Clinical Fellowship for $250,000 to study MECP2 Duplication syndrome.

Rett syndrome is caused by a mutation of the MECP2 gene. Affected girls have one X chromosome with a defective MECP2 gene and one X chromosome with a healthy copy of the gene. In nearly all, the healthy X chromosome is inactivated or silenced. Dr. Lee will aim to activate the silent X chromosome to restore MECP2 levels in the brain. Her team has already concluded that even partial restoration of MECP2 levels has a positive impact on Rett symptoms in mice. This is cutting edge research of paramount importance to those living with Rett syndrome and the second grant from Rettsyndrome.org awarded to Dr. Lee for her work in reactivating the silent X. Dr. Lee's team has developed of a female Rett mouse model that better replicates the symptoms and severity of Rett syndrome and will be used in the new study.

"I believe that a specific treatment for Rett Syndrome is within reach," shared Dr. Lee. "We are very encouraged by [our] findings and believe that an X-reactivation approach can be both efficacious and safe. Through [Rettsyndrome.org's] ANGEL award, we hope to develop and optimize new therapeutics for the treatment of Rett. It is an honor to be an ANGEL award recipient and I look forward to the day when a specific treatment will improve the lives of Rett Syndrome patients."

Rettsyndrome.org Executive Director Melissa Kennedy, commented, ""We are very proud to continue funding Dr. Lee's exciting work in X reactivation. She is one of the most respected and promising researchers in the field, and we are hopeful that her research will lead to a treatment or a cure for thousands living with Rett syndrome."

MECP2 Duplication Syndrome (MDS) is a rare disorder, accounting for 1-2% of X-linked intellectual disability cases in males. Those affected often have low muscle tone, severe developmental delays with absent speech, impaired or absent gait, and restricted hand use, and seizures. Like Rett syndrome, there is no cure for this disorder, and little information on MDS is available to families with an affected individual.

Dr. Pehlivan will work with Dr. Hoda Zogbhi and Dr. Daniel Glaze at Texas Children's Hospital and the Blue Bird Rett Clinic to develop an assessment tool that can aid in MDS diagnosis, help gauge severity and disease course, and be used to evaluate treatment efficacy. In addition, they aim to develop an MDS biomarker to assess disease severity and progression and

potentially be used to evaluate a treatment in clinical trials.

Dr. Glaze shared, “The fellowship that Rettsyndrome.org has funded will make a world of difference for Dr. Pehlivan and the Rett community.”

Kennedy shared, “Rettsyndrome.org believes it is essential to invest in future researchers and clinicians to ensure hope for everyone living with Rett syndrome or a Rett-related disorder. We are proud to be supporting the clinical fellowship of Dr. Pehlivan and his work in MECP2 Duplication syndrome. ”

About Rettsyndrome.org
Rettsyndrome.org (The International Rett Syndrome Foundation) is one of the leading private funders of Rett syndrome research, investing over $49 million to date. The mission of the organization is to accelerate full spectrum research to cure Rett syndrome and empower families with information, knowledge and connectivity. Rettsyndrome.org recently earned Charity Navigator’s prestigious 4-star rating for its strong financial health and commitment to accountability and transparency. Further information about Rettsyndrome.org can be found at www.rettsyndrome.org.

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