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In the news

RETT SYNDROME SCIENTIST HONOURED

The 2014 March of Dimes Prize in Developmental Biology has been awarded to Huda Y. Zoghbi (Baylor College of Medicine, Texas Children's Hospital and Howard Hughes Medical Institute) for her groundbreaking research on the genetic mutations that underlie Rett syndrome and other neurological disorders. The prize recognizes researchers whose work has advanced our understanding of the science that underlies birth defects.

Rett syndrome — named after Austrian paediatrician Andreas Rett, who first described the syndrome in 1966 — is estimated to affect one in every 10,000 to 15,000 live female births, which makes it one of the most common causes of intellectual disability in girls. Other symptoms, which typically develop in infants aged 6–18 months and can vary substantially, include the loss of speech, motor coordination and social skills, as well as inconsolable crying and breathing irregularities.

In 1999, Zoghbi and her team identified the culprit: mutations in the X-linked gene methyl CpG-binding protein 2 (*MECP2*). Her subsequent research revealed that *MECP2* mutations and duplications can produce a range of disorders from neonatal encephalopathy to classic autism, and led others to identify an early-onset schizophrenia syndrome involving *MECP2*.

Today, Zoghbi's discoveries form the basis for the development of treatments for some of these disorders. "Dr Zoghbi's contributions to our understanding of several entirely different neurological disorders, including her finding of the genetic basis of Rett syndrome, have opened new areas of research," says Joe Leigh Simpson, senior vice president for Research and Global Programs at the March of Dimes. "Her work influences the entire field of autism and other neuropsychiatric disorders."

Linda Koch

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