Laura was diagnosed with Rett Syndrome in September 2002.

Rett syndrome is a progressive neurological disorder that affects only one in every 10,000 to 15,000 live female births, occurring in all racial and ethnic groups worldwide.

After a normal birth and normal development during the first 6-18 months of life, the girls then enter a short period of developmental stagnation followed by rapid regression in language and motor skills. The hallmark of the disease is the loss of purposeful hand use and its replacement with repetitive stereotyped hand movements. In addition, screaming fits and inconsolable crying are common by 18-24 months of age. Additional characteristics include autistic features, panic-like attacks, bruxism, episodic apnea and/or hyperpnea, gait ataxia and apraxia (the inability to perform motor functions including interfering with every body movement, including eye gaze and speech), tremors and acquired microcephaly. After this period of rapid deterioration, the disease becomes relatively stable, but the girls will likely develop dystonia and foot and hand deformities as they grow older. Seizures occur more frequently when the disease stabilizes.

The disorder was identified by Dr. Andreas Rett, an Austrian physician who first described it in a journal article in 1966. It was not until after a second article about the disorder was published in 1983 (this time in English) that the disorder was generally recognized. Only in late 1999 the *MECP2* gene responsible for Rett Syndrome identified.

While Rett syndrome is a genetic disorder almost all cases occur spontaneously, which means the mutation occurs randomly and is not inherited.

Why are some cases more severe than others?

Because girls have two copies of the X chromosome and need only one working copy for genetic information, they turn off the extra X chromosome in a process called "X inactivation". This process occurs randomly so that each cell is left with one active X chromosome. The severity of Rett syndrome in girls is a function of the percentage of cells with a normal copy of the MECP2 gene after X inactivation takes place.

Because boys have only one X chromosome they have no protection from the harmful effects of the disorder and die shortly before or after birth.

Is treatment available?

There is no cure. Treatment for the disorder is symptomatic — focusing on the management of symptoms — and supportive, requiring a multidisciplinary approach.

What is the outlook for those with Rett syndrome?

Despite the difficulties with symptoms, most girls with Rett syndrome continue to live well into middle age and beyond, but with a significantly higher incidence of sudden, unexplained death. Because the disorder is quite rare, very little is known about long-term prognosis and life expectancy.

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