Close to home: Biologist was studying gene now linked to daughter’s rare illness

By the time her mother received the doctor’s email, Yuna Lee was already 2 years old, a child with a frightening medical mystery. Plagued with body-rattling seizures and inconsolable crying, she could not speak, walk or stand.

“Why is she suffering so much?” her mother, Soo-Kyung Lee, anguished. Brain scans, genetic tests and neurological exams yielded no answers. But when an email popped up suggesting that Yuna might have a mutation on a gene called FOXG1, Soo-Kyung froze.

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For years, Soo-Kyung, a developmental biologist at Oregon Health and Science University, had worked with the FOX family of genes.

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She also knew harmful FOXG1 mutations are exceedingly rare and usually not inherited — the gene mutates spontaneously during pregnancy. Only about 300 people worldwide are known to have FOXG1 syndrome, a condition designated a separate disorder relatively recently. The odds her own daughter would have it were infinitesimal.

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Balancing the missions of science and motherhood, Soo-Kyung has begun doing what she is uniquely positioned to do: aiming her research squarely at her daughter’s disorder. With Jae’s help, she is studying how the FOXG1 gene works and why mutations like Yuna’s are so devastating.

“Our ultimate goal is to find a better treatment for FOXG1 syndrome patients,” she said. Her day-to-day goal is helping Yuna make slivers of developmental progress.

Read full, original post: A Scientist Finds Her Child’s Rare Illness Stems From the Gene She Studies