Meet Huda Zoghbi, pediatric neurologist working on rare diseases

Huda Zoghbi has uncovered the molecular mechanisms of normal neurodevelopment and neurodegeneration by probing the complexities of rare neurological diseases.

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She was frustrated by the fact that medical science could only ease the symptoms of the many children she worked with who suffered from untreatable neurological disorders. It was then that a patient caught her attention: a girl with Rett syndrome, a rare, poorly characterized disorder that leads to severe learning disability and motor impairments.

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Over the next 10 years, she and her lab members began to collect tissue samples from families with two affected sisters, systematically comparing each of their X chromosome genes.

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[I]n 1999, Zoghbi and her collaborators identified the exact gene, MECP2, which is mutated in Rett syndrome sufferers. The researchers showed that Rett was indeed an X-linked dominant disorder, meaning that just one mutated copy of MECP2, which normally encodes a methyl-CpG-binding protein, was enough to cause the disorder.

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Recently, in collaboration with a biotechnology company, Zoghbi's lab has developed a potential therapy for decreasing MECP2 expression.

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"When I started working on Rett, most researchers didn't think that sporadic disorders could be genetic, but here we found a disease that is genetic but a result of a de novo, not an inherited, mutation," she explains. "This has opened up the search for other genetic forms of disabilities that are sporadic but still caused by a genetic defect."

Read full, original post: Genetic Neurologist: A Profile of Huda Zoghbi