Scientist chases cure for her rare brain disorder using ‘clear genetic blueprint’

In 2011, Sonia Vallabh was handed a genetic report that contained a death sentence. But it also held a map for how to escape. Her body, she learned, harbored a gene mutation, a single wrong letter of DNA in her “prion” gene, that would eventually lead to a rare brain condition called fatal familial insomnia.

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After the diagnosis of the genetic time bomb, [she and her husband] dropped out of their careers in law and engineering and became scientists dedicated to defusing it. They expect to get PhDs next spring.

Now, after seven years, they think they have found a treatment that can do it. It’s called an antisense drug, a type of mirror-image molecule that, if it reaches the brain, could greatly reduce the amount of the prion protein. That could potentially forestall the mysterious chain reaction of misfolding proteins that characterize prion diseases.

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Two weeks ago, on their blog, Vallabh announced she and [Eric] Minikel were working with a commercial partner, the California biotech company Ionis, which specializes in antisense compounds. “For the first time, I am optimistic about a specific therapeutic strategy,” she wrote. It’s “plausible” that antisense could treat the disease “in our lifetime.”

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“The big picture,” [Vallabh] says, “is we are moving much more quickly than anyone could expect due to this beautifully clear genetic blueprint we were handed.”

Read full, original post: One woman’s race to defuse the genetic time bomb in her genes