Forgotten by science: Four decades ago, medical researchers launched studies on rural Colombian families with fatal Huntington’s disease. They are just now following up

Huntington’s is a hereditary neurodegenerative disease caused by excess repetitions of three building blocks of DNA — cytosine, adenine, and guanine — on a gene called huntingtin. The mutation results in a toxic version of a key brain protein, and a person’s age at the onset of symptoms relates, roughly, to the number of repetitions the person carries.

The disease is relatively rare, but in the late 1980s a Colombian neurologist, Jorge Daza, began observing a striking number of cases in [Colombia].

In recent years, a group of researchers at the Universidad Simón Bolívar, in Barranquilla, have taken on the daunting task of rekindling clinical and genetic studies that stalled after the death of their colleague Dr. Daza. It felt, said neuropsychologist Johan Acosta, who is leading the effort, “like starting from zero.”

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