Discovery of single gene mutation could shine light on three different rare diseases

I have Ehlers-Danlos Syndrome (EDS), Postural Orthostatic Tachycardia Syndrome (POTS), and Mast Cell Activation Syndrome (MCAS)—a trifecta of weird diseases. POTS, EDS, and MCAS are so obscure that many doctors have never even heard of them. But a study published [Oct. 17] in *Nature Genetics* might help change that: Researchers have found a genetic mutation that links all three conditions.

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People who have EDS-HT often also have POTS or MCAS or both, yet the relationships between the three remain murky…It's hard to study conditions that look different in every patient…and have few, if any, quantifiable symptoms…All three conditions are far more common in women, a trait long associated with meager research funding and minimal medical concern.

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Consequently, there are no FDA-approved tests for these diseases, and there are certainly no cures.

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Fortunately, the cost of DNA sequencing has continued to drop, and clusters of researchers around the world are beginning to take a look.

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It's "interesting work," says Lawrence Afrin, a hematologist at the University of Minnesota. He told me the study represents "early progress toward further unraveling these illnesses."

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: One Gene Mutation Links Three Mysterious, Debilitating Diseases