

New findings on fragile X syndrome may explain why some drugs prove ineffectual

Scientists have gained new insight into fragile X syndrome, the most common cause of inherited intellectual disability, by studying the case of a person without the disorder, but with two of its classic symptoms.

Fragile X syndrome results from an inherited genetic error in a gene called FMR1. The error prevents the manufacture of a protein called FMRP. Loss of FMRP is known to affect how cells in the brain receive signals, dialing up the amount of information allowed in. The gene is on the X chromosome, so the syndrome affects males more often and more severely than females, who may be able to compensate for the genetic error if their second copy of FMR1 is normal.

In studying fragile X, researchers' focus long has been on the problems that occur when brain cells receive signals. Like radio transmitters and receivers, brain cells send and receive transmissions in fine tuned ways that separate the signals from the noise. Until recently, most fragile X research has focused on problems with overly sensitive receivers, those that allow in too much information. The new study suggests that fragile X likely also causes overactive transmitters that send out too much information.

"The mechanisms that researchers have long thought were the entirety of the problem with fragile X are obviously still very much in play," Klyachko said. "But this unique case has allowed us to see that something else is going on."

The finding also raises the possibility that drugs recently tested as treatments for fragile X may be ineffective, at least in part, because they only dialed down the brain's receivers, presumably leaving transmitters on overdrive.

Read full, original article: [FMRP: New Genetic Clues Found In Fragile X Syndrome](#)