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A novel recovery: Characterisation of Progranulin in FTD human pluripotent stem cells and inhibition of nonsense-mediated RNA decay

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FTD is a presenile neurodegenerative disorder, genetically FTD is complex and up to 40% of patients report a family history, which suggests a large genetic element in the aetiology of the disease. Research conducted into characterising the mutations present on chromosome 17q21, has cast doubt and evidence led research groups to exclude MAPT. Considering an alternative explanation, that there may be a second gene on chromosome 17q21, sequencing 80 genes ranked on known function, PGRN was discovered.

Further understanding PGRN is imperative to design potential therapeutic-targets for neurodegenerative disease as it is implemented in multiple processes. Here, we model PGRN-mutations in iPSC-derived neurons, allowing us to demonstrate a disease-specific model for FTD.

Once mutations were sequenced, we found that the novel c.77delG was not present and both mutants were Q337x mutations. A 24-hour drug treatment was conducted, an NMD inhibitor was provided for the treated and DMSO supplement for untreated. Cells were harvested for subsequent qPCR. Immunocytochemistry was conducted to measure changes in intensity.

Results are questionable however it is clear that the NMD inhibitor had a promising effect on recovering levels lost in the untreated samples, that said without another set of data is not possible to confirm. A better understanding of the complexity of progranulin and its role within the brain will help to direct the development of progranulin-modulating therapies not only for FTD familial patients but those with neurodegenerative disease. This finding creates a potential target for pharmaceuticals to mediate the debilitating symptoms FTD presents. Future directions will be considered and discussed.

Biography

Katie Marie Case is a successful Neuroscience Masters graduate, obtaining an additional specialism in Neural stem cells and nervous system repair from King's College London. Conducting her research in neurodegenerative disease progression and potential therapeutic targets using the cutting edge of technology. She is passionate about the potential power of Stem cells and is an advocate for empowering women in science. An experienced STEM communicator she believes that we need to educate and empower the next generation so that they can answer the questions we leave behind.

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