Frontotemporal dementia has distinct clinical and pathological features linked to C9ORF72 mutations.

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Introduction

The studies reveal a strong link between C9ORF72 mutations and psychosis, implying that clients with C9ORF72 mutations have qualitatively different behavioural traits. C9ORF72 gene mutations will be a key cause of late onset psychosis as well as frontotemporal dementia with neuromuscular disorders. Some concerns are relevant to the larger problem of FTD–place MND's in FTLD. FTLD is caused by a wide range of pathologies. The pathophysiology of TDP-43 seems heterogeneous. TDP-43 immunoreactive pathogenic alterations inside neurons and/or neurites have been divided into distinct categories. The discovery of a hex nucleotide repeat expansion in the C9ORF72 gene as the cause of chromosome 9-linked frontotemporal dementia and motor neuron disease opens up the possibility of learning more about the connections between such diseases but other clinical types of frontotemporal lobar degeneration. We looked for mutations in the C9ORF72 gene in 398 patients with frontotemporal dementia, progressive non-fluent aphasia, semantic dementia, or a combination of these diseases. Nonetheless, the studies reveal a strong link between C9ORF72 mutations and psychosis, implying that patients with C9ORF72 mutations have qualitatively different behavioural traits. C9ORF72 gene mutations may be a key cause of late onset psychosis as well as frontotemporal dementia with neuromuscular disorders [1,2].

Procedures in genetics, clinical medicine, and pathology

As part of a molecular genetic study of neurodegenerative illnesses approved by the local Research Ethics Committee, blood samples from patients were taken with informed written consent. Demographic, neurological, behavioural, and cognitive findings were documented for the 398 individuals who took part in the study. Where post-mortem brain tissue became usable, brains were fixed for at least 3 months in 10% buffered formaldehyde, and 14 standard blocks were taken following fixation, including the frontal and temporal cortex (with hippocampus), cerebellar cortex, medulla oblongata (at level of hypoglossus nucleus), but also spinal cord [3,4].

Symptoms and signs of the nervous system, as well as findings from neuroimaging

In all but one of the nine MND patients, bulbar neurons were implicated in the development of sickness. The neurological indications in two of these individuals were restricted to the cranial nerves, indicating increasing bulbar palsy. Some people had limb involvement, with upper and lower motor neuron symptoms that fit the amyotrophic lateral dementia criteria [5].

Conclusion

However many but not all instances of FTD–MND are caused by mutations in the C9ORF72 gene. Patients with the mutation present with a variety of symptoms, the most common of which are somatic, behavioural, or linguistic. Malignant traits vary as well. Despite this, there are diagnostic features that differentiate the patients from other FTD patients. The strong link between C9ORF72 mutations and psychosis shows that C9ORF72 mutations may play a key role not just in FTD–MND but also in the development of delayed psychosis.

References

1. Baborie A, Griffiths TD, Jaros E, et al. Pathological correlates of frontotemporal lobar degeneration in the elderly. Acta Neuropathol. 2011;121:365-71.
2. Baker M, Mackenzie IR, Pickering-Brown SM, et al. Mutations in Progranulin cause tau-negative frontotemporal dementia linked to chromosome 17. Nature. 2006;442:916-19
3. Caselli RJ, Windebank AJ, Petersen RC, et al. Rapidly progressive aphasic dementia and motor neuron disease. Ann Neurol. 1993;33:200-07.
4. DeJesus-Hernandez M, Mackenzie IR, Boeve BF, et al. Expanded GGGGCC Hexanucleotide repeat in noncoding region of C9ORF72 causes chromosome 9p-linked FTD and ALS. Neuron. 2011;72:245-56.
5. Josephs KA, Hodges JR, Snowden JS, et al. Neuropathological background of phenotypical variability in frontotemporal dementia. Acta Neuropathol. 2011;122:137-53.

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