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New genes (since 2020) in FTD and their clinical features

Frontotemporal lobar degeneration (FTLD) represents a group of disorders with highly heterogeneous clinical and pathological presentation. Genetic research over the past two decades in FTLD families led to the identification of three common FTLD genes (microtubule-associated protein tau, progranulin, and chromosome 9 open reading frame 72) and a small number of rare FTLD genes, explaining the disease in almost all autosomal dominant FTLD families. The identified genes have triggered many studies and broaden our understanding of the disease process. Even though this is a highly heterogenous disorder, some correlation between genetic variants and pathological subtype of FTLD exist. Importantly, the majority of so-called sporadic patients are not explained by the known genes. Thanks to the rapid development of next generation sequencing and omics technologies, new strategies were and are being used to tackle that issue. Such approaches have increased the number of genes associated with disease onset with the TANK Binding Kinase 1 gene as the most prominent example shedding light onto unsuspected disease mechanisms. In this presentation, we will review current knowledge of genetics of FTLD and provide an update on on-going large scale genomics projects.