Genetic factors causing sporadic and familial amyotrophic lateral sclerosis (ALS/MND) with or without frontotemporal dementia.

https://neurodegenerationresearch.eu/survey/genetic-factors-causing-sporadic-and-familial-amyotrophic-lateral-sclerosis-alsmnd-with-or-without-frontotemporal-dementia/

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Country

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Title of project or programme

Genetic factors causing sporadic and familial amyotrophic lateral sclerosis (ALS/MND) with or without frontotemporal dementia.

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Swedish Research Council

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Start date of award

01-01-2013

Total duration of award in years

5

The project/programme is most relevant to:

Motor neurone diseases

Keywords
Research Abstract

ALS is a fatal neurodegenerative syndrome characterized by progressive loss of motorneurons in the brain and spinal cord resulting in paralysis and death. 230 cases are diagnosed annually

in Sweden. 10% have a dominantly Mendelian-inherited family history of ALS and 15 genes have been identified, most common are mutations in SOD1 and the recently found C9ORF72 with a GGGGCC-repeat expansion. Since 1993, 167 SOD1 mutations have been found, 43 by us and 18 in Nordic ALS cases. 6% of ALS cases have a SOD1 mutation. The D90A is the only SOD1 mutation inherited as a recessive trait and causes an unique type of ALS with long survival suggesting that D90A patients have co-inherited a modifying protective gene which is absent in other mutants. Our objectives are to find the D90A-modifying factor, to study the prevalence of C9ORF72-repeat expansions in patients with ALS, ALS+FTD, and FTD, characterize the phenotype and penetrance for patients with different sizes of C9ORF72 expansions in blood and autopsy tissue specimens from different parts of the CNS. Our southern blot results shows ALS patients to have >800 repeats but <1600, while most controls <<30. We have found a few controls with intermediate expansions and two patients with homozygous expansions. Comparative SOD1 and C9ORF72 expressions studies will be performed in fibroblasts cultures established from patients with C9ORF72, SOD1, FUS, NF-H, VAPB and Alsin mutations, and in vitro antisense studies performed.

Lay Summary
Further information available at:

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Investments > €500k

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Motor neurone diseases

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Database Categories:
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