Intermediate Length C9orf72 Expansion In An Als Patient

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Intermediate Length C9orf72 Expansion In
Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology. Beer AM(1), Cooper-Knock J, Higginbottom A, Highley JR, Wharton SB, Ince PG, Milano A, Jones AA, Al-Chalabi A, Kirby J, Shaw PJ. Author information: (1)Sheffield Institute for Translational Neuroscience (SITraN), University of Sheffield , UK. PMID:

Intermediate length C9orf72 expansion in an ALS patient ...
Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology Article (PDF Available) in Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration 16(3-4):1...

(PDF) Intermediate length C9orf72 expansion in an ALS ...
The expansion of a hexanucleotide repeat GGGGCC in C9orf72 is the most common known cause of ALS accounting for ~ 40% familial cases and ~ 7% sporadic cases in the European population. In most people, the repeat length is 2, but in people with ALS, hundreds to thousands of repeats may be observed.

C9orf72 intermediate expansions of 24–30 repeats are ...
The hexanucleotide repeat expansion in C9orf72 is a common cause of amyotrophic lateral sclerosis/frontotemporal dementia and also rarely found in other psychiatric and neurodegenerative conditions. Alleles with >30 repeats are often considered an expansion, but the pathogenic repeat length threshold is still unclear.

C9orf72 hexanucleotide repeat length in older population ...
Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology ALEXANDER M. BEER 1, JOHNATHAN COOPER-KNOCK 1, ADRIAN HIGGINBOTTOM 1, J. ROBIN HIGHLEY 1, STEPHEN B ...

Intermediate length C9orf72 expansion in an ALS patient ...
Intermediate C9orf72 repeat lengths, however, remain uncertain. We systematically reviewed the role of intermediate C9orf72 alleles in C9orf72-related neurological disorders. We identified 49 studies with adequate available data on normal or intermediate C9orf72 repeat length, involving subjects with FTD, ALS, Parkinson's disease (PD), atypical parkinsonism, Alzheimer's disease (AD) and other aetiologies.

Intermediate C9orf72 Alleles in Neurological Disorders ...
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Intermediate Length C9orf72 Expansion In An Als Patient
Intermediate length repeat expansions of CAG (polyQ) repeat in the ATXN2 gene have also been reported to increase the risk of developing ALS in North America and Europe. We screened 131 ALS patients and 127 healthy controls from India for C9orf72 and ATXN2 repeat expansions.

C9orf72 hexanucleotide repeat expansions and Ataxin 2 ...
addition to the C9orf72 expansion, we observed an ATXN2 polyQ intermediate length expansion, and OPTN p.Met468Arg in patients who exhibited ALS and FTD or bvFTD. We conclude that the C9orf72 expansion likely explains much of the ALS-FTD phenotype; however, inheritance of these additional variants likely modifies the

OPTN p.Met468Arg and ATXN2 intermediate length polyQ ...
We demonstrate here that intermediate expansions of the C9ORF72 gene (>20RCs) are associated with increased risk for clinically diagnosed PD. Moreover, combining two different clinical datasets, we found seven PD but no controls having over 23RCs.

C9ORF72 Intermediate Repeat Copies Are a Significant RK ...
C9orf72 intermediate repeats are associated with corticobasal degeneration, increased C9orf72 expression and disruption of autophagy. Microsatellite repeat expansion disease loci can exhibit pleiotropic clinical and biological effects depending on repeat length. Large expansions in C9orf72 (100s-1000s of units) are the most common genetic cause of amyotrophic lateral sclerosis (ALS) and frontotemporal degeneration (FTD).

C9orf72 intermediate repeats are associated with ...
The frequency of ATXN1 alleles with ≥33 polyglutamine repeats was particularly high in the group of ALS patients carrying the C9orf72 expansion (12/59, 20.3%). We confirmed this result in an independent cohort of C9orf72 Italian patients (10/80 cases, 12.5%), thus finding a cumulative frequency of ATXN1 expansion of 15.82% in C9orf72 carriers ...

ATXN1 intermediate-length polyglutamine expansions are ...
Patient carriers of a C9orf72 repeat expansion exhibit remarkable heterogeneous clinical and pathological characteristics suggesting the presence of modifying factors. In accordance with other repeat expansion diseases, repeat length is the prime candidate as a genetic modifier. Observations of earlier onset ages in younger generations of large families suggested a mechanism of disease ...

Relationship between C9orf72 repeat size and clinical ...
While depletion of C9ORF72 only has a partial deleterious effect on neuron survival, it synergizes with the toxicity of Ataxin-2 carrying intermediate length of polyglutamine expansions to induce motor neuron dysfunction and neuronal cell death.

203228 - Gene ResultC9orf72 C9orf72-SMCR8 complex subunit ...
Since intermediate repeat expansions result in decreased transcription of the gene, we explored the hypothesis that C9orf72 intermediate alleles could be a genetic risk for autoimmune conditions. We genotyped 69 systemic lupus erythematosus (SLE) and 77 rheumatoid arthritis (RA) patients, with 68 expansion-negative ALS patients, as control.

C9orf72 Intermediate Alleles in Patients With Amyotrophic ...
C9orf72 expansion (202 ALS, 63 FTD, and 57 FTD-ALS). Results: We found a significant association with intermediate repeat size ($29 CAG) in patients with ALS (both familial and sporadic) and, for the first time, in patients with familial FTD-ALS. Of interest, we found the co-occurrence of pathogenic C9orf72 expansion in 23.2% of ATXN2

Contribution of ATXN2 intermediary polyQ expansions in a ...
In addition to the C9orf72 expansion, we observed an ATXN2 polyQ intermediate length expansion, and OPTN p.Met468Arg in patients who exhibited ALS and FTD or bvFTD.

OPTN p.Met468Arg and ATXN2 intermediate length polyQ ...
Another interesting feature of the GGGGCC repeat expansions in the C9orf72 gene is the length variability between different tissues in single individuals. This seems to intrinsically suggest that the repeat is highly unstable. However, the largest repeats are not seen in fast-replicating tissues, such as blood cells or endothelial cells.

C9orf72 - an overview | ScienceDirect Topics
Specifically, 59 had an expansion in C9orf72, 25 a variant in SOD1, 18 in TARDBP, and 4 in FUS. Patients with ATXN1 intermediate-length ≥33 repeats were significantly more frequent in the whole group of ALS patients (9.16%) than in controls (vs. 5.48%) (p = 0.003).