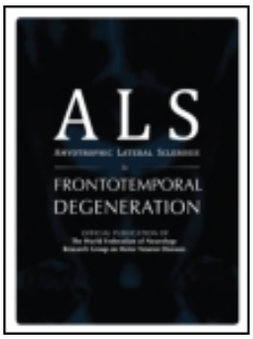

Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration

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Correction to: Residual association of chromosome 9P21 SNPS with ALS after exclusion of C9ORF72 mutated cases [Abstract]

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ERRATUM

Correction to: Residual association of chromosome 9P21 SNPS with ALS after exclusion of C9ORF72 mutated cases [Abstract]

Jones A, Shatunov A, Ellis CM, Leigh PN, Shaw CE, Al-Chalabi A. Residual association of chromosome 9P21 SNPS with ALS after exclusion of C9ORF72 mutated cases [Abstract C29]. Amyotroph Lateral Scler. 2012;13(s1):18.

Following publication of the abstracts from the *23rd International Symposium on ALS/MND* within the 2012 supplement of '*Amyotrophic Lateral Sclerosis*', some errors were identified in abstract C29 – Residual association of chromosome 9P21 SNPS with ALS after exclusion of C9ORF72 mutated cases.

An incorrect version of this abstract appeared in the printed supplement and the online PDF. The correct version is shown below. The title of the abstract has also been updated.

Residual association, haplotype analysis, and significant epistasis for chromosome 9p21 SNPS with and without ALS C9ORF72 mutated cases

Pathological expansion of a hexanucleotide repeat in an intron of the C9ORF72 gene is a cause of about 10% of all ALS, and was identified through a series of linkage and association analyses. The risk SNPs and haplotype that tag the mutation are frequent in the general population. It is possible that the mutation is not the only disease causing variation in this region. For example, in Parkinson's disease, a situation exists in which linkage is seen in families to the same genomic region as those with no family history, but is caused by two different genetic lesions; one a Mendelian, high penetrance mutation, the other, common variation at a SNP.

Aim: To identify whether the 9p21 variation still associates with the disease when the mutation has been accounted for.

To remove the ALS cases with the mutation from our dataset to elucidate other SNPs genome-wide that associate with ALS.

Method: We screened case samples previously analysed in a genome-wide association study (GWAS) for the pathological expansion of C9ORF72. We stratified further analyses by the presence or absence of the expansion, examining the locus for residual association, analysing for genome-wide associations, confirming the 9p21 haplotype association with the mutation, and testing for epistasis.

Results: There were 599 case samples and 4142 controls. A total of 39 of the cases were expanded. Controlling for the presence of the expansion, there was residual association at chromosome 9p21, (top SNP rs903603 $p = 1.87e^{-5}$). Genome-wide, no new loci were identified in the more mutation-only sample using association analyses but we did identify an interesting significant hit in the epistasis analysis ($p = 8.03e^{-6}$). A previously identified ALS-associated 9p21 haplotype did not show association once the expanded cases were accounted for.

Conclusions: There may be further disease-causing variation at the chromosome 9 locus and epistasis interactions with the mutation.

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