W04-04 - **GENETICS AND PATHOPHYSIOLOGY OF CATATONIA**

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Catatonia has attracted increasing attention in relation to basic theoretical problems of psychiatry. Periodic catatonia (MIM 605419) is characterized by qualitative hyperkinetic and akinetic psychomotor disturbances through psychotic episodes, and debilitating symptoms in the long term with psychomotor weakness, grimacing facial movements, and apathy. Our aim was to identify new association loci for periodic catatonia a subphenotype of schizophrenic psychoses. We therefore performed a genome-wide association analysis using SNP Microarray and DNA pooling (SNP-MaP) with 500K Affymetrix arrays. We pooled DNA of 245 cases in three biological replicates (n=84, 84, 77) and 216 controls in two biological replicates (n=108, 108). All pools were processed in three technical replicates. Array data was analysed with a modified version of GenePool. We determined the mean relative allele signal value (RAS) of the technical replicates of each pool and compared each case pool with the combined controls. We used a 5-SNP sliding window approach and defined clusters as potentially associated loci if they overlapped in all biological replicates. We then verified pooling data at single genotype level for associated array SNPs at five selected loci using TaqMan assays in an extended cohort (344 cases, 585 controls). Two of these loci showed significant association at single marker (p=0.0002) and haplotype levels (p=0.0042), even after permutation correction (pc=0.0007; pc=0.0224). Our data show that hybridization intensities of pooled DNA correlate well with individual genotyping results. Thus DNA pooling is a useful strategy for GWAS since we were able to detect significant association loci in a complex disease.

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