Rare Copy Number Variants Disrupt Genes Regulating Vascular Smooth Muscle Cell Adhesion and Contractility in Sporadic Thoracic Aortic Aneurysms and Dissections

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“The control data sets used for the analyses described in this manuscript were obtained from dbGaP at http://www.ncbi.nlm.nih.gov/gap through accession numbers phs000092.v1.p1, phs000093.v2.p2, phs000004.v1.p1, phs000142.v1.p1, and phs000182.v1.p1. Funding support for the Study of Addiction: Genetics and Environment (SAGE) was provided through the NIH Genes, Environment, and Health Initiative (GEI) (U01 HG004422). SAGE is one of the GWASs funded as part of the Gene Environment Association Studies (GÉNEVA) under GEI. Assistance with phenotype harmonization and genotype cleaning, as well as with general study coordination, was provided by the GÉNEVA Coordinating Center (U01 HG004446). Assistance with data cleaning was provided by the National Center for Biotechnology Information (NCBI). Support for collection of data sets and samples was provided by the Collaborative Study on the Genetics of Alcoholism (U10 AA008401), the Collaborative Genetic Study of Nicotine Dependence (P01 CA089392), and the Family Study of Cocaine Dependence (R01 DA013423). Funding support for genotyping, which was performed at The Johns Hopkins University Center for Inherited Disease Research, was provided by the NIH GEI (U01HG004438), the National Institute on Alcohol Abuse and Alcoholism, the National Institute on Drug Abuse, and the NIH contract “High throughput genotyping for studying the genetic contributions to human disease” (HHSN268200782096C). Funding support for the National Institute of Neurological Disorders and Stroke (NINDS) Repository Neurologically Normal Control Collection was provided by the NINDS. Funding support for “A Whole Genome Association Scan for Myopia and Glaucoma Endophenotypes using Twin Studies” was provided by the Wellcome Trust, the Guide Dogs for the Blind Association (UK), the National Health and Medical Research Council (350415), the Ophthalmic Research Institute of Australia, the Clifford Craig Medical Research Trust, the National Glaucoma Research Organization, the American Health Assistance Foundation, and the National Eye Institute (NEI) (R01EY018246). Funding support for the GWAS of Lung Cancer and Smoking was provided through the NIH GEI (Z01 CP 010200). The human subjects participating in the GWAS are from the Environment and Genetics in Lung Cancer Etiology (EAGLE) case-control study and the Prostate, Lung Colon, and Ovary Screening Trial, and these studies are supported by intramural resources of the National Cancer Institute. Assistance with phenotype harmonization and genotype cleaning, as well as with general study coordination, was provided by the GÉNEVA Coordinating Center (U01 HG004446). Assistance with data cleaning was provided by the NCBI. Funding support for genotyping, which was performed at The Johns Hopkins University Center for Inherited Disease Research, was provided by the NIH GEI (U01HG004438). Funding support for the NEI Study of Age-Related Macular Degeneration (NEI-AMD) was provided by the NEI. We would like to thank NEI-AMD participants, the NEI-AMD research group, and the Twins Eye Study research group for their valuable contribution to this research.”

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