

**TITLE**

**Insomnia linked to a T2D Gene**

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**Objective:** Insomnia is associated with increased risk for T2D. The chromosome 12q24 locus is linked type 2 diabetes (T2D) and depression. Insomnia often accompanies depression. *PSMD9* on the 12q24 locus is linked to MODY3, T2D, T2D-microvascular and macrovascular complications, and to hypercholesterolemia. Of note, *PSMD9* common variants contribute to the anti-depressant therapy response. Our goal was to determine whether *PSMD9* is linked to insomnia and sleep hours in Italian T2D families.

**Methods:** We phenotyped the Italian families' members for presence and/or absence of insomnia and for average sleep hours per night. The phenotype was described as unknown in all cases in which the diagnosis was unclear or data were not available. We tested in the 200 Italian families for evidence of linkage of the *PSMD9* single nucleotide polymorphisms (SNPs) *IVS3+nt460A>G*, *IVS3+nt437T>C* and *E197G A>G* with insomnia and average sleep hours. We performed non-parametric linkage analysis, linkage disequilibrium model analysis, single SNP analysis, and parametric analysis cluster-based, and variant component analysis, by using the Merlin software. To rule out results due to random chance, 1000 replicates were executed for the significant data.

**Results:** The *PSMD9* SNPs studied and/or any gene variants in linkage disequilibrium are linked to insomnia in our Italian families.

**Conclusions:** This is the first report of *PSMD9* linkage to insomnia in T2D.

**Biography:** Dr. Claudia Gragnoli is an MD/PhD endocrinologist who since early on in her academic career has focused on the genetics of type 2 diabetes (T2D) and maturity-onset-diabetes of the young (MODY). She graduated in Medicine and Surgery in 1992 and became an Endocrinologist in 1998 at the U. La Sapienza of Rome. She performed her research at U. La Sapienza, at the U. of Chicago as JDFI Fellow and Research Associate, at the U. Tor Vergata, where she obtained her PhD in 2003, and at Harvard Medical School, where she was Research Fellow, Instructor (2001-2005), International Rotary Ambassador, NIH Training Fellow and taught/tutored Human Genetics of Complex Disorders, Endocrinology, Chemistry and Cell Biology (2000-2005). Since 2006, she holds a tenure-track position as Assistant Professor in Medicine at Penn State U. College of Medicine Milton S. Hershey Medical Center. She is a member of the Faculty Committee of the Penn State U. PhD Programs in Physiology, Genetics, Biomedical Sciences, and of the MD/PhD Program. Since 2007, she is Joint Assistant Professor in Biology at Temple U. College of Science and Technology, Philadelphia, PA and in Cellular & Molecular Physiology at Penn State U. College of Medicine. Since 2009, she is Joint Assistant Professor in Public Health Sciences at Penn State U. College of Medicine. Since 2008, she is Joint Associate Professor in Biology at Temple U. College of Science and Technology, Philadelphia, PA.