Restless legs syndrome: epidemiological and clinicogenetic study in a South Tyrolean population isolate.

Abstract:
Genetic contributions to restless legs syndrome (RLS) have been consistently recognized from population and family studies. To determine the clinical and genetic features of RLS in a population isolate and explore linkage to three previously described susceptibility loci on chromosomes 12q, 14q, and 9p, respectively, an isolated population in the South Tyrolean Alps was identified and 530 adults participated in the study. Using a two-step strategy, 47 patients with idiopathic RLS were ascertained. The prevalence in the population was 8.9%. Twenty-eight patients (59.6%) had at least one affected first-degree relative and were classified as hereditary cases. In a single extended pedigree, linkage to known RLS loci was investigated specifying autosomal dominant and recessive models; parametric and nonparametric multipoint linkage scores were computed. None of the calculated linkage scores was suggestive of linkage between RLS and any of the three investigated loci. This study was conducted in a population isolate providing for a homogeneous genetic and environmental background. The absence of a suggestive linkage signal at the three known RLS susceptibility loci is indicative of further locus heterogeneity of this frequent disorder and encourages further studies to unveil the genetic causes of RLS.