Evaluation of the potential association of *SOHLH2* polymorphisms with non-obstructive azoospermia susceptibility in a large European population

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Non-obstructive azoospermia (NOA) or spermatogenic failure is a complex disease with an important genetic component that causes infertility in men. Known genetic factors associated with NOA include AZF microdeletions of the Y chromosome or karyotype abnormalities; however, most causes of NOA are idiopathic. During the last decade, a large list of associations between single-nucleotide polymorphisms (SNP) and NOA have been reported. However, most of the genetic studies have been performed only in Asian populations. We aimed to evaluate whether the previously described association in Han Chinese between NOA and two SNPs of the *SOHLH2* gene (involved in the spermatogenesis process) may also confer risk for NOA in a population of European ancestry. We genotyped a total of 551 NOA patients (218 from Portugal and 333 from Spain) and 1,050 fertile controls (226 from Portugal and 824 from Spain) for the genetic variants rs1328626 and rs6563386 using TaqMan assays. To test for association, we compared the allele and genotype frequencies between cases and controls using an additive model. A haplotype analysis and a meta-analysis using the inverse variance method with our data and those of the original Asian study were also performed. No statistically significant differences were observed in any of the analyses described above. Therefore, considering the high statistical power of our study, it is not likely that the two analysed *SOHLH2* genetic variants are related with an increase susceptibility to NOA in the European population.

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