

ASSOCIATIONS BETWEEN VOCAL SYMPTOMS AND GENETIC VARIANTS IN THE OXYTOCIN RECEPTOR AND VASOPRESSIN 1A RECEPTOR GENE

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Most research regarding the etiology of voice disorders focus on environmental risk factors and the genome as a contributing factor is relatively unexplored. A twin study has shown that 35% of the variability in dysphonia between individuals is explained by genetic effects with the rest (65%) being explained by nonshared environmental effects. These results indicated genetic effects on voice, providing reason to further explore the role of genetics in the occurrence of vocal symptoms. The hormones oxytocin and arginine vasopressin have been associated with stress regulation and oxytocin and vasopressin receptor genes have been associated with stress reactivity. Since stress is regarded as a risk factor for vocal symptoms, we wanted to explore the association between the oxytocin (*OXTR*) and arginine vasopressin 1A receptor gene (*AVPR1A*) single nucleotide polymorphisms (SNP) and self-reported vocal symptoms. We also wanted to explore whether such effects might be mediated by cortisol since both oxytocin and vasopressin influence cortisol levels.

A population based sample ($N = 657$) of Finnish twins (born 1965-1989) completed a web-questionnaire on the occurrence of vocal symptoms. A total of ($n = 170$) participants submitted saliva samples for hormone analysis. A total of 20 *OXTR* and *AVPR1A* SNPs were analyzed. The data were analyzed using the Generalized Estimated Equations (GEE) method, which takes into account the dependent structure of family data.

Three *OXTR* polymorphisms (rs2270465, rs2268493, rs7632287) and two *AVPR1A* polymorphisms (rs1587097, rs1042615) showed nominal effects on vocal symptoms, of which one (rs1587097) remained significant after correcting for multiple testing. Of the nominally significant effects, the results suggested potential mediation of the effect of the *OXTR* rs2268493 polymorphism on vocal symptoms through levels of cortisol.

Our results show associations between variants of *OXTR* and *AVPR1A* indicating that oxytocin and vasopressin influence vocal symptoms. The effect of oxytocin seems to be partly mediated through cortisol actions.