

Allelic discrimination in determination of sociality level by SNP of oxytocin receptor gene

Due to human nature social interactions are playing important role throughout human life. But there are differences in human degree of sociality. Whereas one part of people enjoys an active social life and doesn't have many problems with social interactions another part avoids social interactions and have difficulty in dealing with people. Previous studies have shown that the oxytocin system plays an important role in human socially related personality traits and behaviors, referred to as "sociality"[1, 2]. For example, high levels of plasma oxytocin (OXT) have been associated with behaviors indicative of enhanced sociality, such as increased physical contact with a partner and trustworthiness. Differences in sociality can be depended on oxytocin genes variations. One of the genes that are included in oxytocin functioning cycle is oxytocin receptor gene (OXTR), that is located in the human genome on chromosome 3p25. Receptor status can be the reason of different influence of oxytocin on cells.

A single nucleotide polymorphism (SNP) —a single base pair difference —is the most commonly occurring variation in DNA. Because of the omnipresence of SNPs, this particular type of variant is important to understanding the correlation between genetic variation and its effect on the observed phenotype. There is one SNP in the third intron of OXTR gene, rs53576 (G/A), which is interesting for research. rs53576 is the silent G to A change in the OXTR gene. Studies have demonstrated that individuals with the G allele are more empathetic, feel less lonely, employ more sensitive parenting techniques and have lower rates of autism. People with the G-G genotype are better able to discern the emotional state of others than those who carried the A-allele. Real-time quantitative PCR is common method used for effective genotyping of SNP. In our experiment we use the real-time genotyping assay known as TaqMan for discrimination between two alleles of the specific SNP. TaqMan probes are short oligonucleotides, that anneal within DNA in a region containing SNP under study. A part of DNA near SNP amplified by a specific set of primers during Polymerase Chain Reaction (PCR). During amplification each uniquely labeled with fluorophore and quencher probe binds preferentially to one of the two alleles of the SNP of interest with different affinity. As amplification proceeds, the Taq polymerase enzyme cleaves probe, that bounds with high affinity. Degradation of the probe releases the fluorophore from it and breaks the close proximity to the quencher, thus relieving the quenching effect and allowing fluorescence of the fluorophore. The level of fluorescence is measured and the conclusion about the type of nucleotide is made.

We developed the system of allelic discrimination for the rs53576 polymorphism using the set of two primers and two probes, which differ by one nucleotide. The probe for A-allele is FAM-nAn-Q, and for G-allele is Rox-nGn-Q. During the study of 20 volunteers using our allelic discrimination system, we had shown that the system we developed successfully differs each of 3 possible variants that can be present in the human genome - homozygotes (A-A, G-G) and heterozygote (A-G). For all three variants, the data obtained by allelic discrimination were confirmed by sequencing and restriction analysis. Our results indicate that this system can be used to accurately and fast determination of rs53576 in OXTR gene and can be put into practice (population genetics, individual genetic consultations, prediction of specific character features, etc.).

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2. Meyer-Lindenberg A, Domes G, Kirsch P, Heinrichs M. Oxytocin and vasopressin in the human brain: social neuropeptides for translational medicine. *Nat Rev Neurosci*. 2011; 12: 524–538. doi: 10.1038/nrn3044

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