Updated 05/01/2024

OXYTOCIN RECEPTOR (OXTR)

The Love Gene

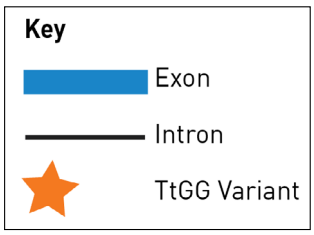
Biology Background

- The Oxytocin Receptor (OXTR) gene produces the OXTR protein, which functions as a receptor for the hormone and neurotransmitter oxytocin.
- The OXTR protein is an integral membrane protein of the family of G protein coupled receptors.
- OXTR has been demonstrated to exhibit its strongest effects in the brain.

Brain

Genomic Locus

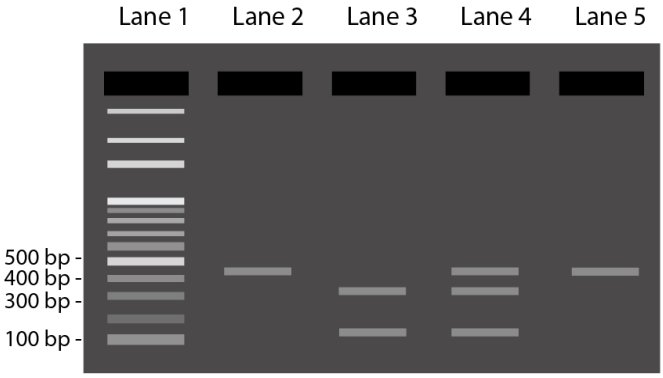
The OXTR gene is located on chromosome 3. The OXTR gene is 19,233 base pairs in length and consists of 4 exons and 3 introns.



The TtGG Variant

- In this assay, you are studying a single nucleotide polymorphism (SNP) in the third intron of the OXTR gene (see star). The nucleotide at this position is typically either a G or an A.
- Since this SNP is in the intron of the gene, it does not directly affect the amino acid sequence of the protein.
- The A variant creates a site for the restriction enzyme BamHI to cut the DNA segment. Cut versus uncut DNA segments can be detected on a gel.

OXTR Gel



Lane 1: DNA ladder
Lane 2: Undigested sample, 435 bp
Lane 3: Homozygous A genotype, 120 bp, 315 bp
Lane 4: Heterozygous GA genotype, 120 bp, 315 bp, 435 bp
Lane 5: Homozygous G genotype, 435 bp

Population Genetics

- The A allele has been associated with structural changes in the brain and was correlated with low scores in tests that measure social ability.
- In other studies, the G allele was linked to emotional sensitivity.
- Additionally, homozygous G and heterozygous GA genotypes were correlated with emotional support-seeking behaviors, whereas homozygous A individuals had a tendency to become recluses during times of high emotional stress.

Influence on Human Health

- Variants that are associated with complex, multifactorial traits, such as behavior, likely contribute only a small amount of effect, with many other genetic and environmental factors playing a significant role.
- Increased oxytocin levels (which act through OXTR) are involved in many human behaviors, including social bonding and fear reduction.
- Decreased levels of oxytocin or OXTR are associated with depression and with autism.

Sources

» Online Mendelian Inheritance in Man (OMIM) http://www.omim.org/entry/167055
» National Center for Biotechnology Information (NCBI) Gene http://www.ncbi.nlm.nih.gov/gene/5021
» NCBI Reference SNP (rs) report https://www.ncbi.nlm.nih.gov/snp/rs53576
» Review on OXTR and behavioral impacts see: Kumsta and Heinrichs Oxytocin, stress and social behavior: neurogenetics of the human oxytocin system. Current Opinion in Neurobiology. (2013)
» The Human Protein Atlas
» UCSC Genome Browser