

OXYTOCIN RECEPTOR (OXTR)

The Love Gene

Biology Background

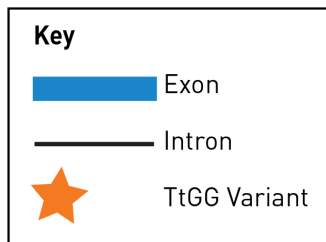
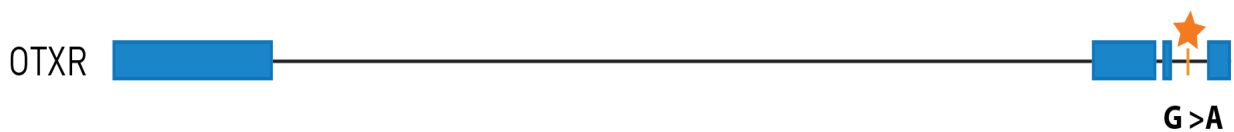
- The Oxytocin Receptor (OXTR) gene produces the OXTR protein, which functions as a receptor for the hormone and neurotransmitter oxytocin.
- The OXTR protein is an integral membrane protein of the family of G protein coupled receptors.
- OXTR has been demonstrated to exhibit its strongest effects in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



Brain

Genomic Locus

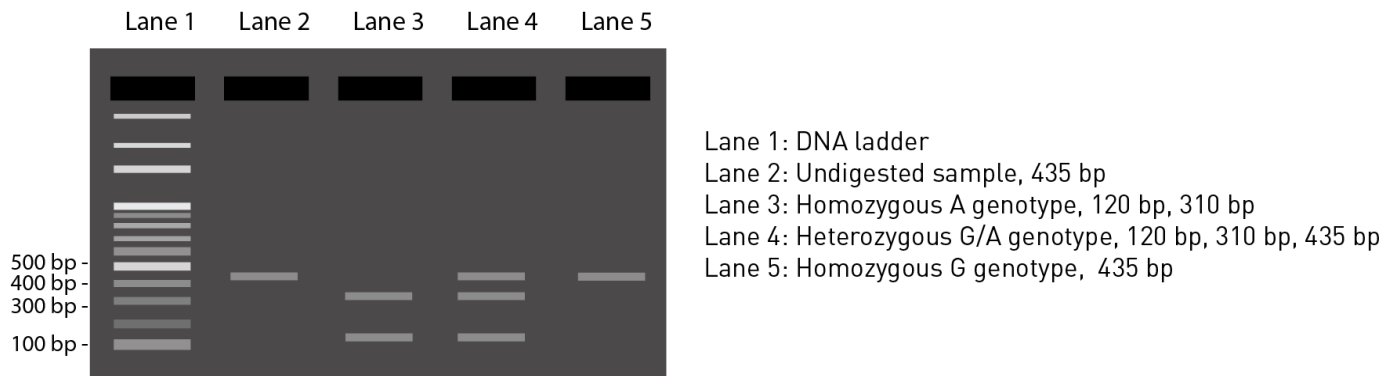
The OXTR gene is located on chromosome 3. It is 19,206 base pairs in length and consists of 4 exons and 3 introns.



The TtGG Variant

- In this assay you are examining a G>A polymorphism (SNP) in the third intron of the OXTR gene (see star).
- The A allele 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



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, GG or GA genotypes were correlated with emotional support-seeking behaviors, whereas homozygous AA individuals had a tendency to become recluses during times of high emotional stress.

Influence on Human Health

- Variants such as these 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 (and its action through OXTR) are involved in many human behaviors including social bonding and fear reduction.
- Decreased levels of oxytocin or OXTR have been associated with disorders such as depression and 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>
- 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]
- Human Protein Atlas