

# OXYTOCIN RECEPTOR (OXTR)

## The Love Gene

### Biology Background

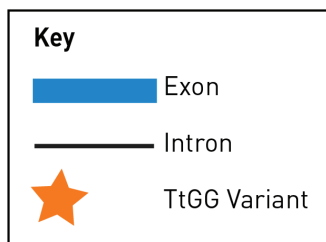
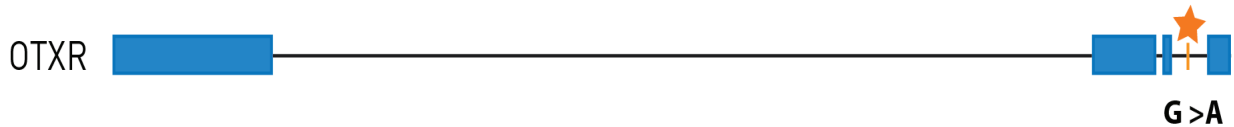
- The Oxytocin Receptor (OXTR) gene produces the OXTR protein, which functions as a receptor for the hormone and neurotransmitter oxytocin.
- The OXTR protein is an integral membrane protein of the family of G protein coupled receptors.
- OXTR has been demonstrated to exhibit its strongest effects in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



Brain

### Genomic Locus

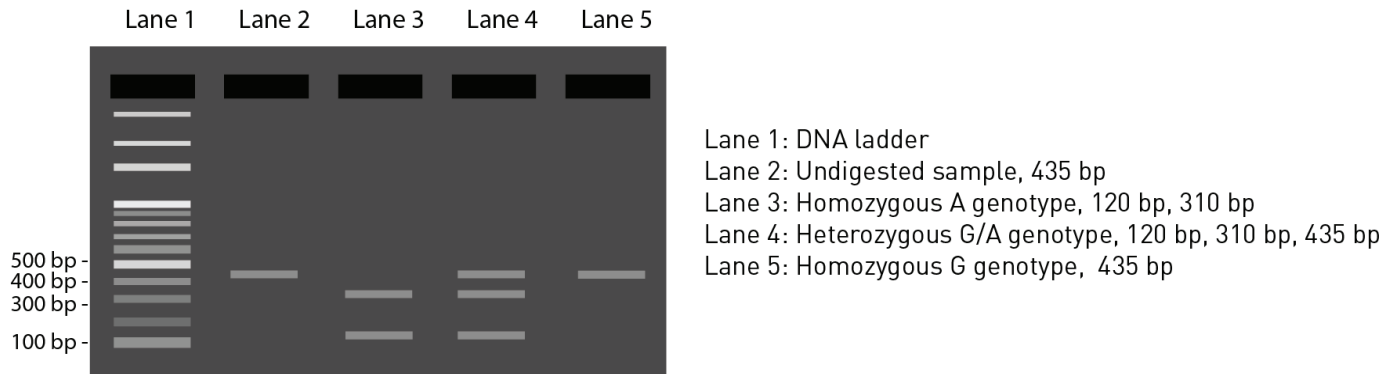
The OXTR gene is located on chromosome 3. It is 19,206 base pairs in length and consists of 4 exons and 3 introns.



### 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