

## GEPHE SUMMARY

	Gephebase Gene	GephelD
GTF2I ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=%GTF2I%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=%GTF2I%#gephebase-summary-title</a> )	GP00002673	Main curator
Draft	Entry Status	Courtier

## PHENOTYPIC CHANGE

	Trait Category	
Behavior, Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=%Behavior%and+Trait+Category=%Morphology%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=%Behavior%and+Trait+Category=%Morphology%#gephebase-summary-title</a> )	Trait	
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=%Coloration+(coat)%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=%Coloration+(coat)%#gephebase-summary-title</a> )	Trait State in Taxon A	
low sensibility to training	Trait State in Taxon B	
high sensibility to training; human-directed canine hypersociability	Ancestral State	
Taxon A		Taxonomic Status
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=%Domesticated%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=%Domesticated%#gephebase-summary-title</a> )		
Taxon A	Latin Name	Latin Name
Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=%Canis+lupus+familiaris%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=%Canis+lupus+familiaris%#gephebase-summary-title</a> )		
dog	Common Name	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Synonyms
subspecies	Rank	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus	Lineage	Lineage
Canis lupus (gray wolf) - (Rank: species) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612</a> )	Parent	Parent
9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615</a> )	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

-	Generic Gene Name	UniProtKB
-	P78347NULL ( <a href="http://www.uniprot.org/uniprot/P78347NULL">http://www.uniprot.org/uniprot/P78347NULL</a> )	GenebankID or UniProtKB
-	Synonyms	
-	0	
-	String	
-	Sequence Similarities	
-	GO - Molecular Function	
-	GO - Biological Process	
-	GO - Cellular Component	
-		Presumptive Null
No ( <a href="https://www.gephebase.org/search-criteria?/and+Presumptive+Null=%No%#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Presumptive+Null=%No%#gephebase-summary-title</a> )		Molecular Type

Unknown ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%5EUnknown%5E#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%5EUnknown%5E#gephebase-summary-title))

Aberration Type

Insertion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%5EInsertion%5E#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%5EInsertion%5E#gephebase-summary-title))

Insertion Size

100-999 bp

Molecular Details of the Mutation

Cfa6.66 = insertion of a transposable element at the GTF2I locus. vonHolt et al 2017 show that the insertion is 259 bp and contains a 187-bp TE.

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%5EAssociation Mapping%5E#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%5EAssociation%20Mapping%5E#gephebase-summary-title))

Main Reference

Transposons in the Williams-Beuren Syndrome Critical Region are Associated with Social Behavior in Assistance Dogs. (2023) (<https://pubmed.ncbi.nlm.nih.gov/38091228>)

Authors

Gnanadesikan GE; Tandon D; Bray EE; Kennedy BS; Tennenbaum SR; MacLean EL; vonHoldt BM

Abstract

A strong signature of selection in the domestic dog genome is found in a five-megabase region of chromosome six in which four structural variants derived from transposons have previously been associated with human-oriented social behavior, such as attentional bias to social stimuli and social interest in strangers. To explore these genetic associations in more phenotypic detail-as well as their role in training success in a specialized assistance dog program-we genotyped 1001 assistance dogs from Canine Companions for Independence®, including both successful graduates and dogs released from the training program for behaviors incompatible with their working role. We collected phenotypes on each dog using puppy-raiser questionnaires, trainer questionnaires, and both cognitive and behavioral tests. Using Bayesian mixed models, we found strong associations (95% credibility intervals excluding zero) between genotypes and certain behavioral measures, including separation-related problems, aggression when challenged or corrected, and reactivity to other dogs. Furthermore, we found moderate differences in the genotypes of dogs who graduated versus those who did not: insertions in GTF2I showed the strongest association with training success ( $\hat{\beta} = 0.23$ , CI = -0.04, 0.49), translating to an odds-ratio of 1.25 for one insertion. Our results provide insight into the role of each of these four transposons in canine sociability and may inform breeding and training practices for working dog organizations. Furthermore, the observed importance of the gene GTF2I supports the emerging consensus that variation in GTF2I genotypes and expression have important consequences for social behavior broadly.

© 2023. The Author(s), under exclusive licence to Springer Science+Business Media, LLC, part of Springer Nature.

Additional References

Structural variants in genes associated with human Williams-Beuren syndrome underlie stereotypical hypersociability in domestic dogs. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28776031>)

## RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103)) (<https://www.gephebase.org/search-criteria?/or+TaxonID=%5E9615%5E/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

@TE