

GEPHE SUMMARY

GTF2l ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=GTF2l^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=GTF2l^#gephebase-summary-title</a> )	Gephebase Gene	GP00002673	GepheID
Draft	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

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 Category
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
low sensibility to training	Trait State in Taxon A
high sensibility to training; human-directed canine hypersociability	Trait State in Taxon B
Taxon A	Ancestral State
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> )	Taxonomic Status

Taxon A	Latin Name	Taxon B	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> )	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> )	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> )	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	dog	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms
subspecies	Rank	subspecies	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	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
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	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
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	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
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

-	Generic Gene Name	P78347NULL ( <a href="http://www.uniprot.org/uniprot/P78347NULL">http://www.uniprot.org/uniprot/P78347NULL</a> )	UniProtKB
-	Synonyms	()	GenebankID or UniProtKB
-	String		
-	Sequence Similarities		
-	GO - Molecular Function		
-	GO - Biological Process		
-	GO - Cellular Component		
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> )		Presumptive Null
			Molecular Type

Unknown (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Unknown^#gephebase-summary-title>)

Aberration Type

Insertion (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=^Insertion^#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=^Association+Mapping^#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<sup>®</sup>, 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 ( $\hat{I}^2 = 0.23$ , 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.

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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+Taxon+ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

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