Visualizing the IWGSC Refseq v1.0 Wheat Assembly in Ensembl Plants







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IWGSC – Wheat Genome Manual and Functional Annotation
PAG XXVII
Jan 15 2018



What I will talk about

- The European Bioinformatics Institute and Ensembl Plants
- Ensembl Plants Genome Browser
- Comparative analysis and the Ensembl Gene Tree
- Variation and the TILLING population in Ensemble
- 10+ genome project



What we do in the European Bioinformatics Institute (EBI-EMBL)

Cambridge (UK)



Hinxton



We provide freely available data and bioinformatics services to the scientific community.

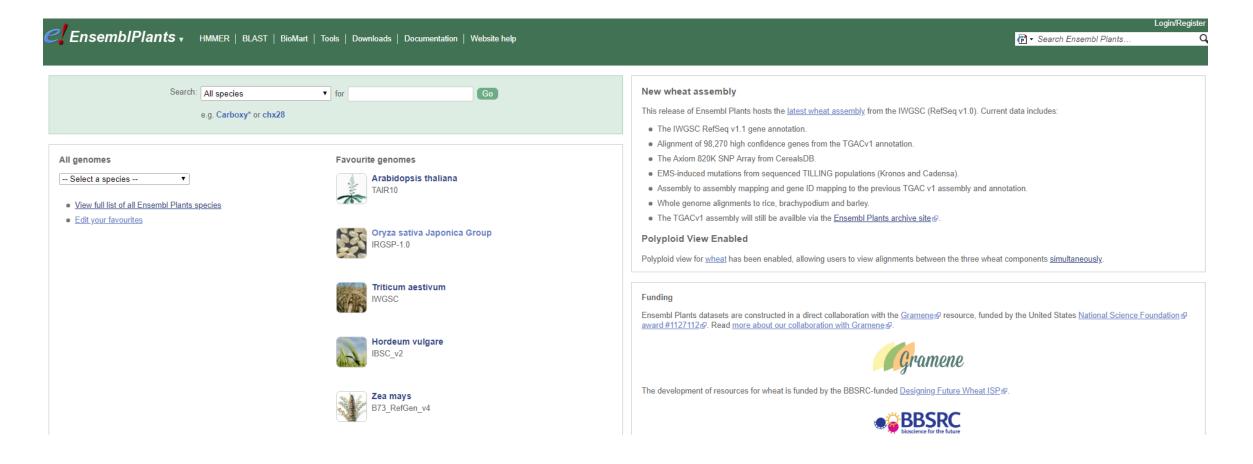


Heidelberg



Barcelona

Ensembl Plants



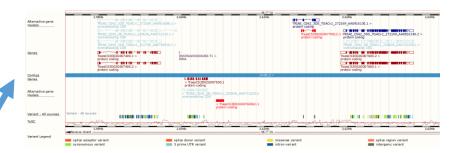
https://plants.ensembl.org

Free and open source



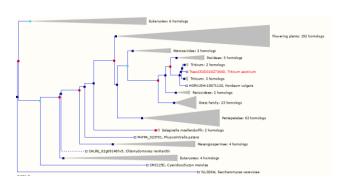
API, VEP, Archive Site, Bulk Download, Release Cycle, Polyploid View, User tracks...

Genome Browser for nearly 60 plants species





Comparative Genomics



Variation Data





Introducing the new IWGSC wheat genome assembly

18TH JULY 2018 BY ERIN (OUTREACH) · COMMENTS OFF



exciting new genome assembly!

One of the biggest highlights of the new Ensembl Plants release 40 is the inclusion of the new Wheat (RefSeq v1.0) genome from the International Wheat Genome Sequencing Consortium (IWGSC).

The path to sequencing the wheat genome has been no easy ride, due to its large and highly repetitive genome. This new assembly from the IWGSC bridges many gaps from the initial genome sequencing effort. Read on to find out more about this



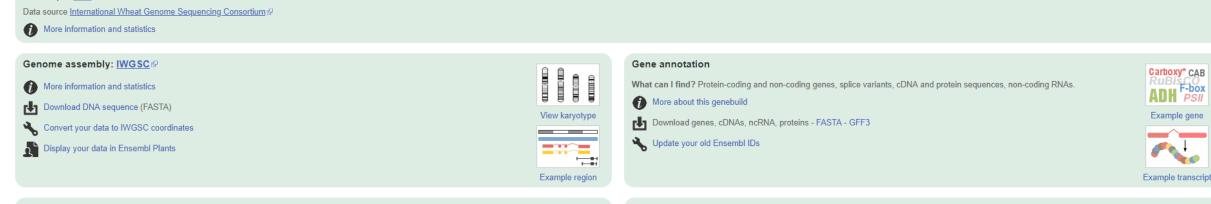
Latest wheat assembly now available in Ensembl

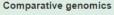


About Triticum aestivum

Triticum aestivum (bread wheat) is a major global cereal grain essential to human nutrition. Wheat was one of the first cereals to be domesticated, originating in the fertile crescent. around 7000 years ago. Bread wheat is hexaploid, with a genome size estimated at ~17 Gbp, composed of three closely-related and independently maintained genomes that are the result of a series of naturally occurring hybridization events. The ancestral progenitor genomes are considered to be Triticum urartu (the A-genome donor) and an unknown grass thought to be related to Aegilops speltoides (the B-genome donor). This first hybridization event produced tetraploid emmer wheat (AABB, T. dicoccoides) which hybridized again with Aegilops tauschii (the D-genome donor) to produce modern bread wheat.

Taxonomy ID 4565 ₽





What can I find? Homologues, gene trees, and whole genome alignments across multiple species.

- More about comparative analyses
 - Phylogenetic overview of gene families
- Download alignments (EMF)
- Genomic alignments [5] [Show»]



Variation

What can I find? Short sequence variants.

- More about variation in Triticum aestivum
- More about variation in Ensembl Plants
- Download all variants GVF VCF VEP

Variant Effect Predictor





https://plants.ensembl.org/wheat

A closer look and examples

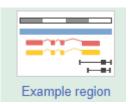
Genome assembly: IWGSC



Download DNA sequence (FASTA)

Convert your data to IWGSC coordinates

Display your data in Ensembl Plants

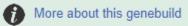






Gene annotation

What can I find? Protein-coding and non-coding genes, splice variants,



Download genes, cDNAs, ncRNA, proteins - FASTA - GFF3



Comparative genomics

What can I find? Homologues, gene trees,

More about comparative analyses

Phylogenetic overview of gene familie:

Download alignments (EMF)

Genomic alignments [5] [Show»]





Variation

What can I find? Short sequence variants.



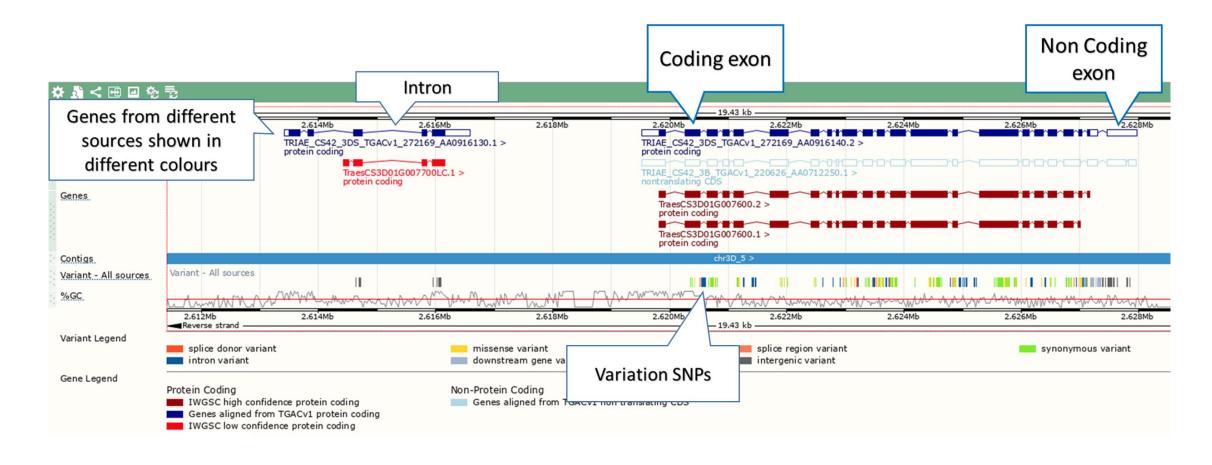
More about variation in Ensembl Plants

Download all variants - GVF - VCF - VEP

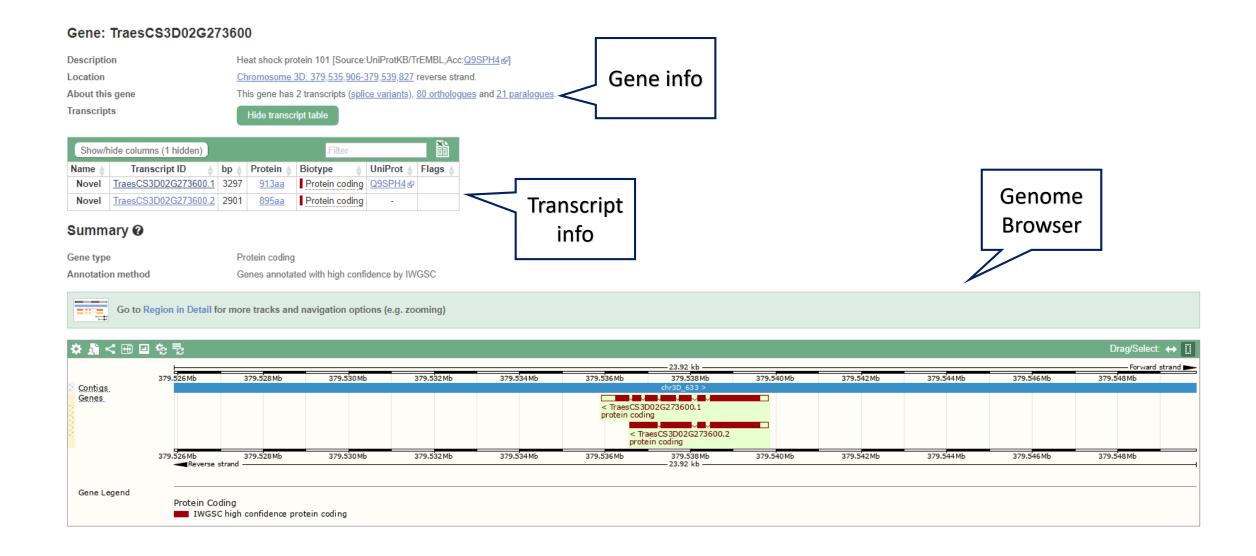
Variant Effect Predictor



The Ensembl Plants Genome Browser



From Genome Browser to Gene



Sequence information

Custom tracks

Share this page

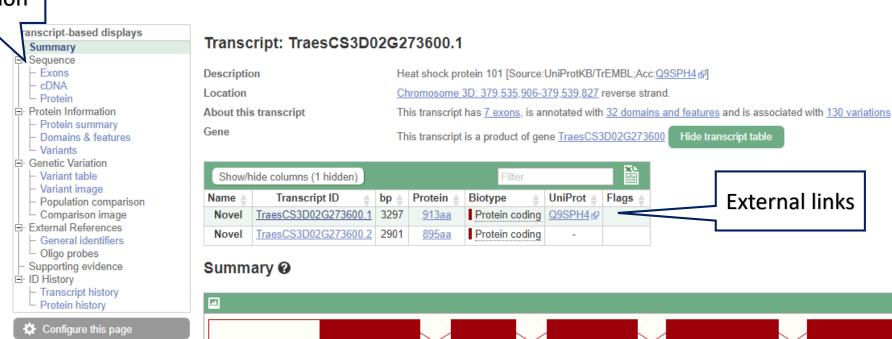
₱ Bookmark this page

Ensembl Plants is produced in

collaboration with Gramene

Export data

From Gene to Transcript



Statistics Exons: 7, Coding exons: 7, Transcript length: 3,297 bps, Translation length: 913 residues

-3.92 kb

Version TraesCS3D02G273600.1.

Type Protein coding

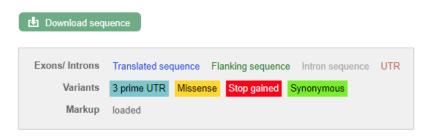
< TraesCS3D02G273600.1

Reverse strand

Annotation Method Genes annotated with high confidence by IWGSC

From Transcript to Exon Sequence

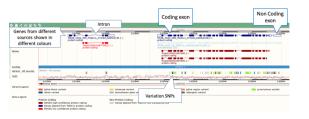
Exons @

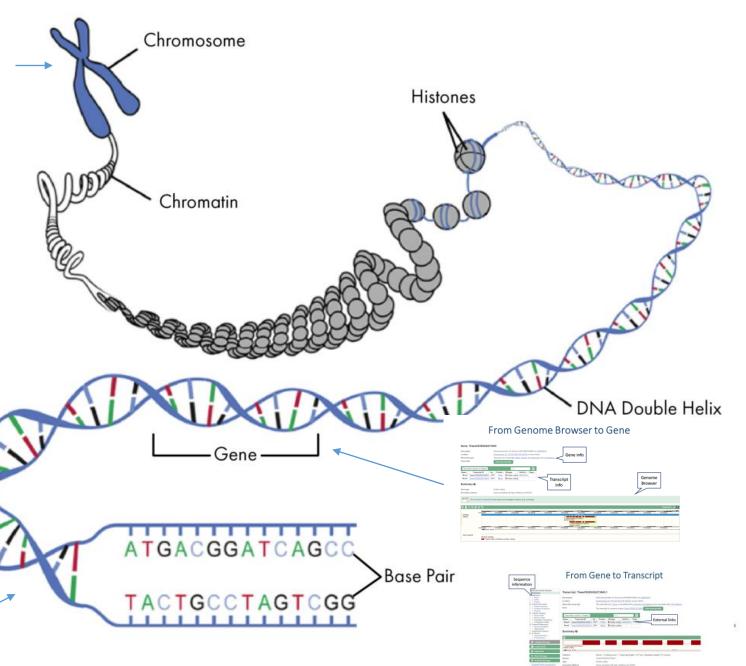


Show All ▼ entries			Show/hide columns			Filter	
No.	Exon / Intron	Start	End	Start Phase	End Phase	Length	Sequence
	5' upstream sequence						cacggttccgggtttttcaaataaaacggttccacactagaatgttctcg
1	TraesCS3D02G273600.1-E1	<u>379,539,827</u>	<u>379,538,459</u>		1	1,369	GACATTCTTGTCCCCCCTAACTCGCCCCCTTCCCGCCGCTTCCACGCCGTTCTAGACCTC CGGCGAATCCTCCCCTCC

Variation in Sequence

The Ensembl Plants Genome Browser





From Transcript to Exon Sequence

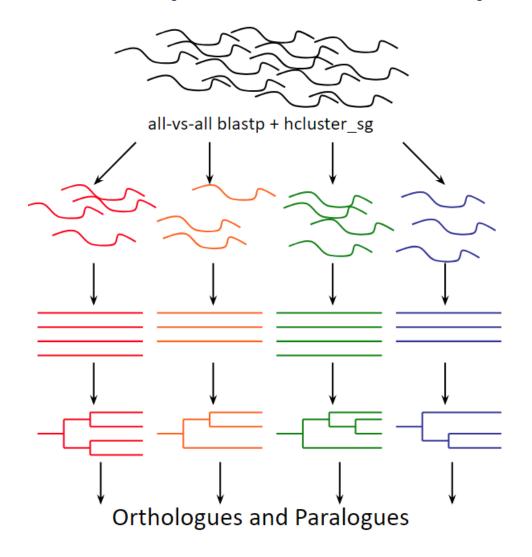
| Company | Comp

What I will talk about

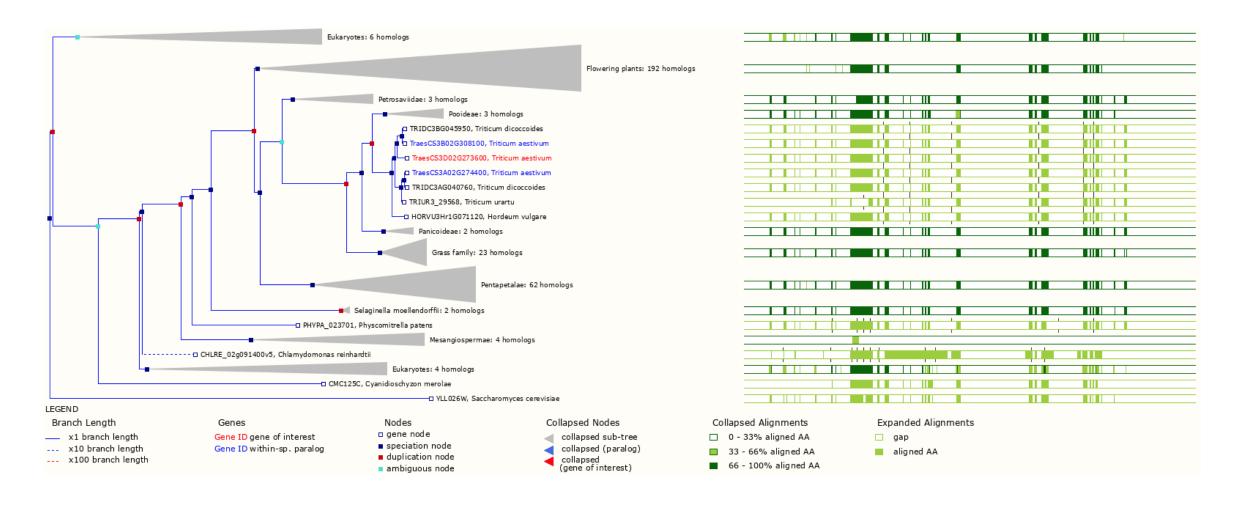
- ✓ The European Bioinformatics Institute and Ensembl Plants
- ✓ Ensembl Plants Genome Browser
- Comparative analysis and gene tree
- Variation and the TILLING population in Ensemble
- 10+ genome project



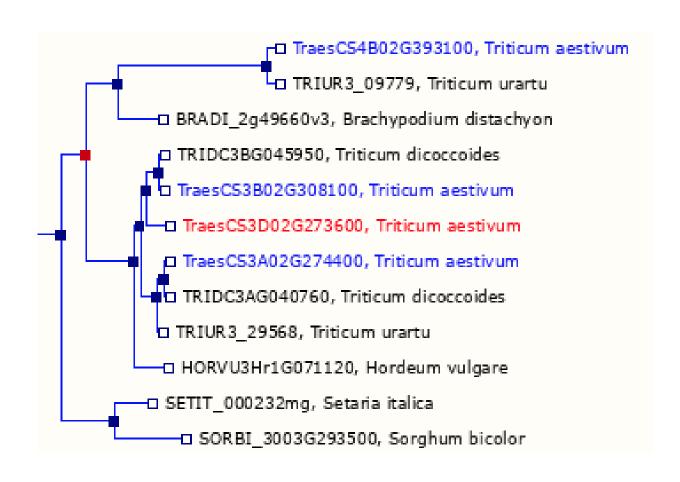
Creating the Comparative Analysis Gene Tree



Gene tree for TraesCS3D02G273600 (Heat shock protein)



Gene tree for TraesCS3D02G273600 (Heat shock protein)



Orthologues @



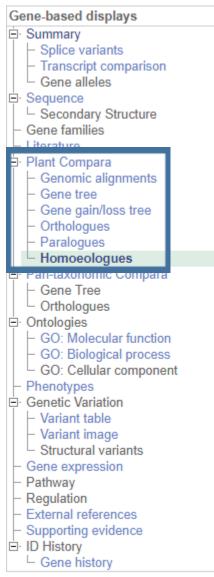
Summary of orthologues of this gene Hide 🔾

Click on 'Show details' to display the orthologues for one or more groups of species. Alternatively, click on 'Configure this page' to choose a custom list of species.

Species set	Show details	With 1:1 orthologues	With 1:many orthologues	With many:many orthologues	Without orthologues
All (49 species)		4	10	14	<u>21</u>
Amborellales (1 species)		0	1	0	0
Bryophyta (1 species)		0	1	0	0
Chlorophyta (2 species)		0	1	0	1
Liliopsida (20 species)		4	0	1	<u>15</u>
Lycopodiophyta (1 species)		0	0	1	0
Rhodophyta (3 species)		0	2	0	1
Eudicotyledons (16 species)		0	5	11	0

Selected orthologues Hide 🔾

Show All ▼ entries		Show/hide columns					Filter	
Species	▲ Type	▲ Orthologue	dN/dS	Target %id	Query %id	△ GOC Score	→ WGA Coverage	High Confidence
Amborella trichopoda	1-to-many	AMTR_s00086p00119290	n/a	83.75 %	83.57 %	n/a	n/a	n/a
	View Gene Tree	Compare Regions (AmTr_v1.0_scaffold00086:1,562,078-1,566,063:-1)						
		View Sequence Alignments						
Arabidopsis lyrata	Many-to-many	ATHSP101 (fgenesh2 kg.2 1695 AT1G74310.1)	n/a	83.42 %	83.24 %	n/a	n/a	n/a
	<u>View Gene Tree</u>	Compare Regions (2:16,230,526-16,234,220:-1)						
		View Sequence Alignments						
Arabidopsis lyrata	Many-to-many	fgenesh2_kg.72872AT4G14670.1	n/a	75.88 %	44.80 %	n/a	n/a	n/a
	View Gene Tree	Compare Regions (7:13,781,642-13,783,448:-1)						
		View Sequence Alignments						
Arabidopsis lyrata	Many-to-many	scaffold_102080.1	n/a	72.09 %	54.87 %	n/a	n/a	n/a
	View Gene Tree	Compare Regions (1:7,827,877-7,830,479:-1)						





Custom tracks

Gene: TraesCS3D02G273600

Description Heat shock protein 101 [Source:UniProtKB/TrEMBL;Acc:Q9SPH4]

Location Chromosome 3D: 379,535,906-379,539,827 reverse strand.

About this gene This gene has 2 transcripts (splice variants), 80 orthologues and 21 paralogues.

Transcripts Hide transcript table

Show/h	ide columns (1 hidden)		Filter					
Name 🍦	Transcript ID 🗼	bp 🌲	Protein	Biotype	UniProt 🍦	Flags 🍦		
Novel	<u>TraesCS3D02G273600.1</u>	3297	<u>913aa</u>	Protein coding	Q9SPH4®			
Novel	<u>TraesCS3D02G273600.2</u>	2901	<u>895aa</u>	Protein coding	-			

Homoeologues

■ Download homoeologues

Show/hide columns		
Species	▲ Type	▲ Homoeologue
Triticum aestivum	1-to-1	<u>TraesCS3A02G274400</u>
	View Gene Tree	Compare Regions (3A:503,751,497-503,755,373:-1)
		View Sequence Alignments
Triticum aestivum	1-to-1	<u>TraesCS3B02G308100</u>
	<u>View Gene Tree</u>	Compare Regions (3B:495,150,368-495,154,251:-1)
		View Sequence Alignments

Polyploid view @ ☆ < □ % </p> Chr. 3D 500.00 kb Forward strand 379.3Mb 379.6Mb 379.7Mb Contigs Genes TraesC\$3D02G273500 > < TraesC\$3D02G273600 Alternative gene TraesCS3D02G362200LC > TRIAE CS42 3B TGAC 1_222109_AA0757460.1 > < TraesCS3D02G362500LC models TRIAE_CS42_3DL_TGACv1_249024_AA0834810.2 > < TraesCS3D02G362600LC TraesCS3D02G362300LC > < TraesCS3D02G362700LC < TRIAE_CS42_3DL_TGACv1_249024_AA0834820.3 < TRIAE CS42_3AL_TGACV1_196076_AA0657130.2 < TRIAE CS42_38_TGACV1_222109_AA0757480.1 < TraesCS3D02G362400LC 379.3Mb 379,4Mb 379.5Mb Gene Legend Protein Coding Non-Protein Coding protein coding IWGSC low confidence non translating CDS Genes aligned from TGACv1 protein coding Genes aligned from TGACv1 non translating CDS IWGSC low confidence protein coding Chr. 3A 500,00 kb -Forward strand 503.6Mb 503.7Mb 503.8Mb 503.9Mb Contigs Genes < TraesCS3A02G274600 TraesCS3A02G274300 : FraesCS3A02G274400 aesCS3A02G274500 > < TraesCS3A02G274700 < TraesCS3A02G274800 Alternative gene < TRIAE CS42 3AL TGACV1 194357 AA063165URIAE CS42 6AL TGACV1 471206 AA1504620 TraesCS3A02G4 TraesCS3A02G405800LC < TraesCS3A02G405600LC models < TRIAE CS42_3DL_TGACv1_253780_AA0895530.2 195163_AA0645580.1 > TRIAE_CS42_3AL_TGACv1_196076_AA0657130.BaesCS3A02G406000LC > TRIAE_CS42_3DL_TGACv1_249024_AA0834820.2 TRIAE CS42 3B_TGACV1_222109_AA0757460RIAE_CS42_3B_TGACV1_221492_AA0741900.3 raesCS3A02G405900LC > < TRIAE_CS42_3AL_TGACv1_194357_AA0631640.2 < TRIAE_CS42_3DL_TGACV1_251190_AA0878590.2 503.9Mb 503.6Mb 503,7Mb 503.8Mb Gene Legend Protein Coding Non-Protein Coding protein coding IWGSC low confidence non translating CDS IWGSC low confidence protein coding Genes aligned from TGACv1 non translating CDS Genes aligned from TGACv1 protein coding Chr. 3B 500 00 kb -Forward strand 494.9Mb 495.0Mb 495.2Mb 495.1Mb 495.3M Contigs Genes TraesCS3B02G308000 > < TraesCS3B02G308100 < TraesCS3B02G476900LC TraesCS3B02G477300LC Alternative gene RIAE_CS42_3AL_TGACv1_195163_AA0645580.2 > models TRIAE_CS42_3B_TGACv1_222109_AA0757460.2 > < TraesCS3B02G477000LC TRIAE_CS42_3DL_TGACv1_249024_AA0834810,1 > < TraesCS3B02G477100LC TraesCS3B02G476700LC > < TraesCS3B02G477200LC B TRIAE_CS42_3AL_TGACV1_196076_AA0657130.1 TRIAE CS42 3DL TGACV1 249024 AA0834820.1 < TRIAE_CS42_3B_TGACv1_222109_AA0757480.2

495.1Mb

495.0Mb

TraesCS3B02G476800LC >

A closer look and examples

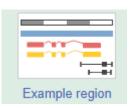
Genome assembly: IWGSC



Download DNA sequence (FASTA)

Convert your data to IWGSC coordinates

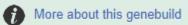
Display your data in Ensembl Plants





Gene annotation

What can I find? Protein-coding and non-coding genes, splice variants,



Download genes, cDNAs, ncRNA, proteins - FASTA - GFF3



Comparative genomics

What can I find? Homologues, gene trees,

More about comparative analyses

Phylogenetic overview of gene familie:

Download alignments (EMF)

Genomic alignments [5] [Show»]







Variation

What can I find? Short sequence variants.



More about variation in Ensembl Plants

Download all variants - GVF - VCF - VEP

Variant Effect Predictor



Variation SNP (820K SNP Array)

BA00249348 SNP

Most severe consequence

Alleles

Location

HGVS names

Synonyms

Original source

About this variant

missense variant | See all predicted consequences

T/C | Highest population MAF: 0.09

Chromosome 4A:714193714 (forward strand) | VCF: 4A 714193714 BA00249348 T C

This variant has 3 HGVS names - Hide □

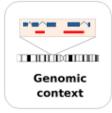
- 4A:g.714193714T>C
- TraesCS4A01G446800.1:c.695A>G
- <u>TraesCS4A01G446800</u>.1:p.Lys232Arg

CerealsDB AX-94599889

Markers from 820k SNP Array provided by CerealsDB

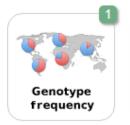
This variant overlaps 1 transcript and has 475 sample genotypes.

Explore this variant @

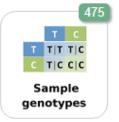


















CerealsDB

Tools for the analysis of the wheat genome

lome Wheat Seq

DArT Markers

SNPs (KASP)

SNPs (Axiom®)

SNPs (iSelect)

SNPs (TagMan®

TGby S

2)1...

Welcome to CerealsDB

The CerealsDB web-site was created by members of the Functional Genomics Group at the University of Bristol. The site provides a range of facilities for the study of the wheat genome. The site has been designed with breeders in mind, and we hope that is will be easy and straightforward to use. Principally the site is aimed at those who wish to obtain information about SNP markers; e.g., the sequence upon which they are based, obtain primers used for their identification, identify the haplotypes of common UK varieties. These publicly funded resources are available as an on-line, searchable database (please see "Important Note" box to the right of this page).

The last update to the database was October 2018.

SNPs databases

The information regarding SNP markers is divided by platform:

.

Axiom® 820K and 35K SNP Arrays

Along with researchers at Affymetrix, we have developed both an '820,000' and a '35,000' feature wheat SNP array. The data sets for these (Affymetrix code, Bristol SNP code and probe sequence containing the SNP ambiguity code) can be downloaded here.

- 3

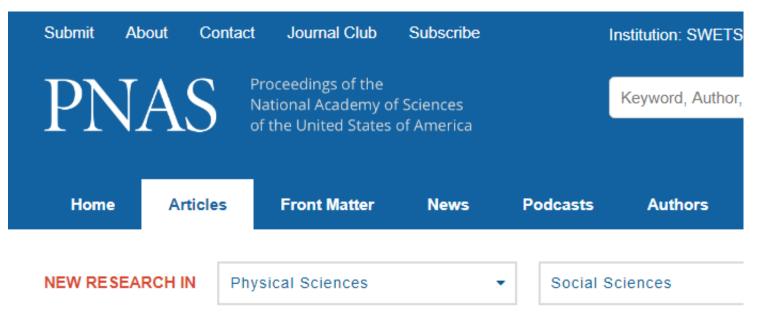
iSelect Array

The iSelect Array contains over 80,000 SNP loci of which 44,000 have been mapped.

.

KASP probes

Wheat TILLING population



Uncovering hidden variation in polyploid wheat



Ksenia V. Krasileva, Hans A. Vasquez-Gross, Tyson Howell, Paul Bailey, Francine Paraiso, Leah Clissold, James Simmonds, Ricardo H. Ramirez-Gonzalez, Xiaodong Wang, Philippa Borrill, Christine Fosker, Sarah Ayling, Andrew L. Phillips, Cristobal Uauy, and Jorge Dubcovsky

Wheat TILLING population

- 2,735 wheat mutant lines were sequenced and developed on a public database
- Lines from Kronos (tetraploid) and Cadenza (hexaploid)
- Database includes more than 10 million EMS induced mutations



Seeds with mutations can be ordered

- Researchers and breeders can study and order seeds with mutation
- A foundation for many wheat experiments and research
- Makes it possible to do double knockouts (Kronos) and triple knockouts (Cadenza) in a few generations
- Knockouts allows you to uncover hidden variation





in silico TILLING mutant resources

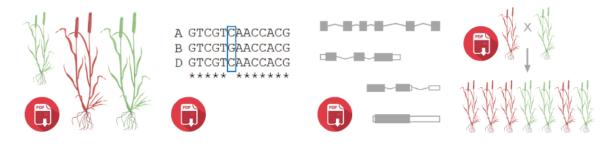
Knock-out mutations are available in most wheat genes providing an invaluable resource for characterising gene function.

<u>Targeting Induced Local Lesions In Genomes (TILLING)</u> is a method of inducing and identifying mutations in specific genes. This section will describe how to use the *in silico* wheat TILLING resource (www.wheat-tilling.com).

This resource consists of TILLING populations developed in tetraploid durum wheat cv 'Kronos' and hexaploid bread wheat cv 'Cadenza' as part of a joint project between the University of California Davis, Rothamsted Research, The Earlham Institute, and John Innes Centre.

The exome sequences of 1,535 Kronos and 1,200 Cadenza mutants have been re-sequenced using Illumina next-generation sequencing, the raw data aligned to the IWGSC Chinese Spring chromosome arm survey sequence, mutations identified, and their effects predicted based on the protein annotation available at the Ensembl Plants archive site.

This section will cover the following four topics:



Selecting TILLING Mutants

Designing Genome Specific
Primers

Improving Gene Models

Designing Crossing Schemes

http://www.wheat-training.com/tilling-mutant-resources/

Cadenza0225.chr3D.379537010 SNP

Most severe consequence missense variant | See all predicted consequences

Alleles G/A

Location Chromosome 3D:379537010 (forward strand) | VCF: 3D 379537010 Cadenza0225.chr3D.379537010 G A

HGVS names This variant has 5 HGVS names - Show

⊞

Synonyms Primer metadata Cadenza0225.chr3D.379537010:snp_type=non-homoeologous;total_contigs=3;A=tgacctcAaaccttggcgC;B=tgacctcAaaccttggcgT;common=cAcgagcaaccttcatCgaA;primer_type=chromosome_specific;product_size=69;EMS-GT=Het

Original source EMS-induced mutations & from sequenced TILLING populations. Seeds can be ordered from UK SeedStor or US Dubcovsky lab & Line identifier is variant name up to dot (e.g Kronos3128).

About this variant This variant overlaps 2 transcripts

Gene and Transcript consequences

Show/hide columns								Filter	X.
Gene	Transcript (strand)	Allele ▲ (transcript allele)	Consequence Type	Position in transcript	Position in CDS	Position in protein	Amino acid	Codons	SIFT Detail
TraesCS3D01G273600	<u>TraesCS3D01G273600.1</u> (-) biotype: protein_coding	A (T)	missense variant	2171 (out of 2742)	2171 (out of 2742)	724 (out of 913)	A/V	GCG/GTG	0.01 Show
TraesCS3D01G273600	<u>TraesCS3D01G273600.2</u> (-) biotype: protein_coding	A (T)	missense variant	2261 (out of 2688)	2261 (out of 2688)	754 (out of 895)	A/V	GCG/GTG	deleterious how

No overlap with Ensembl Regulatory features

SIFT predicts SNP effect on protein function

Based on sequence homology and physical properties of amino acids

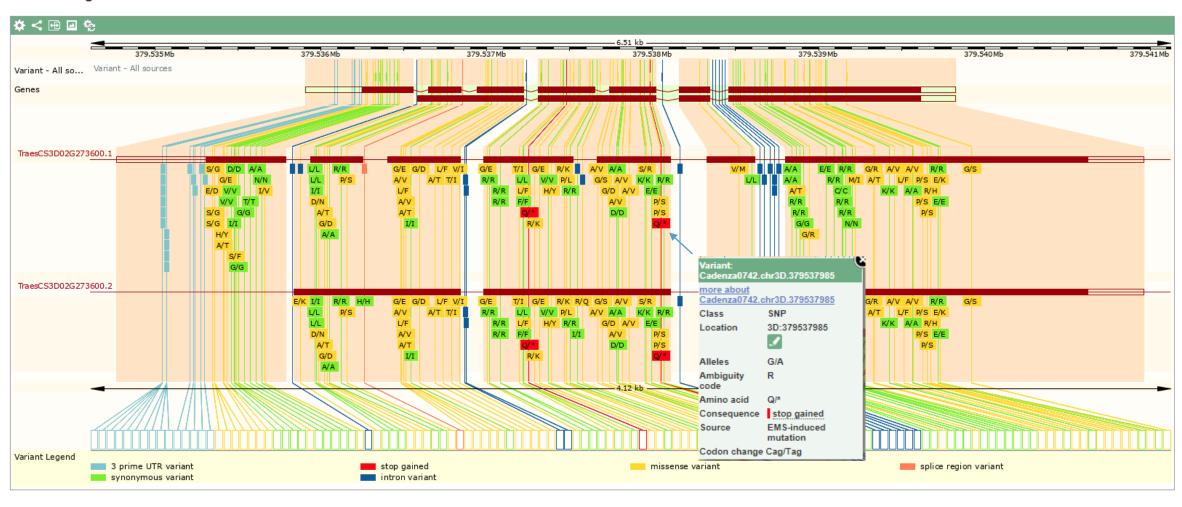
http://sift.bii.a-star.edu.sg/

SIFT predictions for all gene variations

Cadenza0455.chr3D.379536699	3D:379536699	C/T	SNP	EMS-induced mutation	-	-	missense variant	A/T	796	0.37	<u>TraesCS3D01G273600.1</u>
Cadenza1723.chr3D.379536710	3D:379536710	C/T	SNP	EMS-induced mutation	-	-	missense variant	G/D	792	0.37	<u>TraesCS3D01G273600.1</u>
Cadenza1241.chr3D.379536718	3D:379536718	G/A	SNP	EMS-induced mutation	-	-	synonymous variant	Α	789		<u>TraesCS3D01G273600.1</u>
Cadenza1165.chr3D.379536763	3D:379536763	C/T	SNP	EMS-induced mutation	-	-	synonymous variant	R	774		<u>TraesCS3D01G273600.1</u>
Cadenza1808.chr3D.379536786	3D:379536786	G/A	SNP	EMS-induced mutation	-	-	missense variant	P/S	767	0.15	<u>TraesCS3D01G273600.1</u>
Cadenza1494.chr3D.379536853	3D:379536853	G/A	SNP	EMS-induced mutation	-	-	splice region variant intron variant	-			<u>TraesCS3D01G273600.1</u>
Cadenza2068.chr3D.379536992	3D:379536992	C/T	SNP	EMS-induced mutation	-	-	missense variant	G/E	730	0.09	<u>TraesCS3D01G273600.1</u>
Cadenza1819.chr3D.379536995	3D:379536995	G/A	SNP	EMS-induced mutation	-	-	missense variant	A/V	729	0.07	<u>TraesCS3D01G273600.1</u>
Cadenza1793.chr3D.379537002	3D:379537002	G/A	SNP	EMS-induced mutation	-	-	missense variant	L/F	727	0.02	<u>TraesCS3D01G273600.1</u>
Cadenza0225.chr3D.379537010	3D:379537010	G/A	SNP	EMS-induced mutation	-	-	missense variant	A/V	724	0.01	<u>TraesCS3D01G273600.1</u>
Cadenza0844.chr3D.379537011	3D:379537011	C/T	SNP	EMS-induced mutation	-	-	missense variant	A/T	724	0.01	<u>TraesCS3D01G273600.1</u>
Cadenza0726.chr3D.379537030	3D:379537030	G/A	SNP	EMS-induced mutation	-	-	synonymous variant	1	717		<u>TraesCS3D01G273600.1</u>

All mutations for a given gene

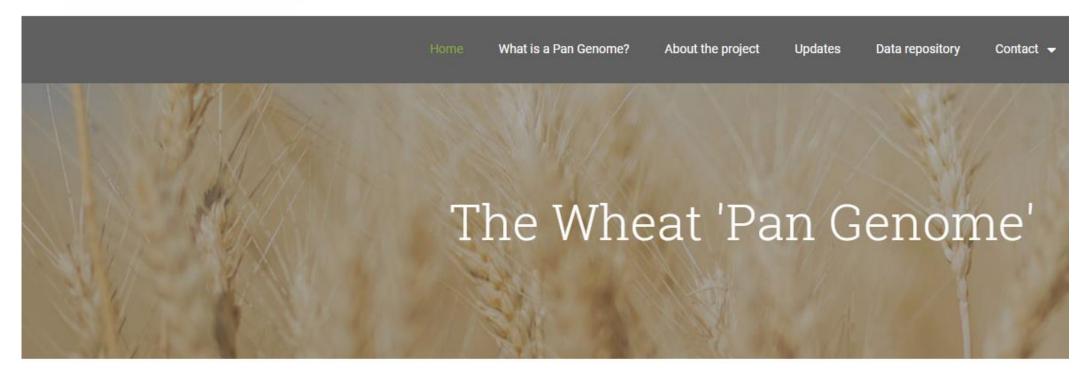
Variant image @



A look into the future







Would you like to know more?

- Our outreach team gives workshops and courses for organizations and institutions
- Minimum of 10 participants
- Workshops are free (just pay for travel and food)
- http://www.ensembl.org/info/about/outreach/index.html
- helpdesk@ensemblgenomes.org











Thanks!!!



































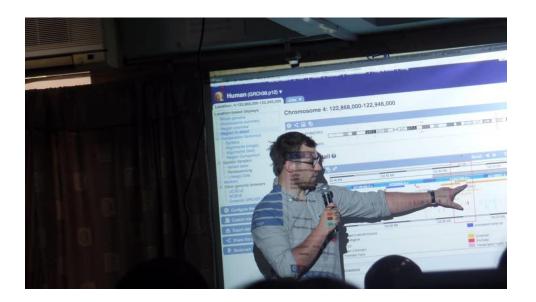


Thanks again!



Ensembl4Breeders Event Feb 22nd 2019





C Ensembl

Experience meeting with Ensembl Plants and the plant breeding community

European Bioinformatics Institute Hinxton, Cambridge

