

Webinar Wheat Data Interoperability guidelines

research data sharing without barriers rd-alliance.org

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Introduction and contexte





The Wheat Initiative

- Created in 2011 following endorsement by G20 Agriculture Ministries to improve food security
- A framework to identify synergies and facilitate collaborations for wheat improvement at the international level
- The Wheat Initiative members
 - Countries: Argentina, Australia, Brazil, Canada, China, France, Germany, Hungary, India, Ireland, Italy, Japan, Spain, Turkey, UK, USA
 - International organizations: CIMMYT, ICARDA
 - Private companies: Arvalis, Bayer CropScience, Florimond Desprez V&F, KWS UK, Limagrain, Monsanto Company, RAGT 2n Saateen Union Research, Syngenta Crop Protection



The Wheat Data Interoperability WG

- Aims: contribute to the improvement of Wheat related data interoperability by
 - Building a common interoperability framework (metadata, data formats and vocabularies)
 - Providing guidelines for describing, representing and linking Wheat related data

Contributors

















Sponsors







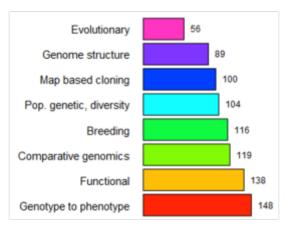




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State of the art

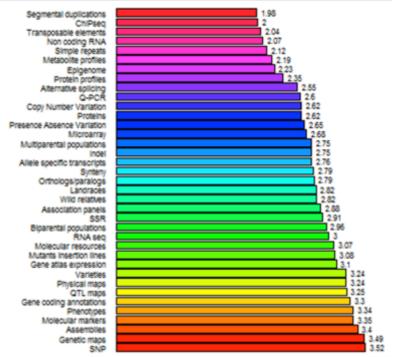
Studies



Repositories



Data types





The methodology

Surveys

- Landscape of Wheat related standards and their use by the community
- Comprehensive overview of Wheat related ontologies and vocabularies

Workshops

- Recommendations
- Mappings between different data formats
- Actions to conduct in order to improve the current level of Wheat related data interoperability
- Interoperability use cases

Implementation

- Interactive cookbook: recommendations + guidelines
- A repository of Wheat related linked vocabularies (Bioportal)



The outputs of the WDI working group



The deliverables

- Guidelines (http://wheatis.org/DataStandards.php)
 - Data exchange formats
 - Example: VCF (Variant Call Format) for sequence variation data, GFF3 for genome annotation data, etc.
 - Data description best practices
 - Consistent use of ontologies, consistent use of external database cross references
 - Data sharing best practices
 - Share data matrices along with relevant metadata (example: trait along with method, units and scales or environmental ones)
 - Useful tools and use cases that highlight data formats and vocabularies issues
- A repository of wheat related ontologies and vocabularies (http://wheat.agroportal.lirmm.fr/ontologies)
 - Allows the access to the ontologies and vocabularies through APIs.
- A prototype
 - Implementation of use cases of wheat data integration within the AgroLD (Agronomic Linked Data) tool: http://www.agrold.org



http://wheatis.org

About

Collaborators

Search

Data Standards

Submit Data

Tools Links

WheatIS Nodes

WheatIS



About

This project aims at building an International Wheat Information System. called hereafter WheatIS, to support the wheat research community. The main objective is to provide a single-access well base system to access to the available data resources and bioinformatics tools.

This project is based on the principles listed below:

- Collective building of the WheatIS to better respond to the needs of the international wheat community;
- Incremental implementation to offer rapidly an operational information system;
- Emphasis on Quality Assurance to serve as a framework for an approach with incremental implementation;
- Promotion of an open-access model for data exchange;
- Reliance on a distributed system;
- Use of Virtual Machine and Cloud Computing technologies to facilitate sharing data and tools:
- Promotion of the visibility of each participating platform to contribute to their sustainability.

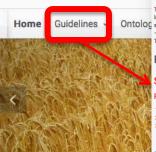


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Wheat Data Interoperability Guidelines

Sequence variations



The sequence variations are the nucleotides differences between two (or several) sequences at the same locus (usually between a reference sequence and another sequence). Three types of sequence variations-Ontolog single-nucleotide polymorphisms (SNPs), insertions and deletions (indels), and short tandem repeats (STRs) have been mainly reported in plant genomes.

The most currently available sequence variations for wheat are SNPs

Recommendations

For Variant (e.g. SNP) calling performed by bioinformaticians:

- 1. Use a reference wheat genome sequence
- 2. Data format: Use the VCF
- Provide associated metadata

1. Reference sequence

The currently most commonly used reference bread wheat sequence is the IWGSC survey sequence (cv Chinese Spring), available at the IWGSC Sequence Repository and EBI.

When available, we encourage the use of the chromosomes reference sequence.

Data format

We recommend to use the latest VCF file format

Description

Welcome

These recommendations has The Variant Call Format (VCF) is a text file used in bioinformatics for storing gene sequence variations. The format has been developed with the advent of large-scale genotyping and DNA sequencing projects, such a Group (WG), one of the WGs the 1000 Genomes Project. VCF format specifications can be found here

Warning: The VCF files generated for exome capture need to be labeled as such and can not been merge Interoperability Interest Grou with those from IWGSC context.

initiative that aims to reinfor 3. Metadata

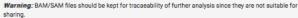
societal demands for sustair Data sharing

research programmes to inc | We recommend to provide a minimal set of metadata to contextualize the provenance of the SNPs and to rovide information about the SNP quality analysis.

For data sharing, the following information should be provided in the header section of the VCF file (header lines have to be preceded by "##" characters) or as a separate tabulated file.

	and have to be preceded by with challened of a dispersion and a departure laborated me.		
	Name	Description	
PROMOTE the adoption of commo standards, vocabularies a best practices for Wheat of management	RUN NAME	Name of the sequencing run that produced the data we are interested in.	
	RUN DESCRIPTION	Description of this run.	
	SUB RUN NAME	Part of a sequencing run that produced the data we are interested in. According to the sequencing technology involved, the sub run can be a lane (for 454 sequencers), a flowcell for (Ilumina sequencers)	
	ANALYSIS NAME	Name of the SNP calling analysis	
	ANALYSIS SOFTWARE NAME	Software used for the SNP calling analysis	
	ANALYSISCONTACT NAME	Person who performed the analysis	
	PROTOCOL NAME	Name of the sequencing protocol	
	MAPPING GENOME NAME	Name and version of the reference genome used to call the variations	
	MAPPING GENOME TAXON NAME	Taxon of the reference genome used to call the variations	
	MAPPING_GENOME DESCRIPTION	Description of the reference genome used to call the variations	
	GENOTYPE NAME	Name of the sample/individual that has been sequenced.	
	GENOTYPE TAXON	Taxon of the sample/individual that has been sequenced.	
Outstal	PROJECT NAME	Name of the project that funded the sequencing	

Filters applied to call SNPs (ex: DP > 10)



Data submission

For data submission in international repositories (EBI, NCBI), we advise to fill the dedicated XML format (http://www.ebi.ac.uk/ena/submit/preparing-xmls#vcf).

Most popular Tools

Identification of sequence variations includes 3 steps:

- 1. Mapping of the reads on the reference genome
- 2. Calling the sequence variations
- 3. Filtering out unrelevant results regarding mainly depth and sequence quality and mapping quality.

Mapping tools

- Bowtie 2

SNP calling tools

- SAM tools

Filter tools

- VCF tools
- VCF utils
- SAM tools

Example

Example of a VCF file dedicated to wheat data:

##fileformat=VCFv4.1

#CHROM POS ID REF ALT QUAL FILTER INFO FORMAT 102 403 407-IV 60 93 ACBarrie A labasskaja CS Estacao M6 Marquis Neepawa PI153785 PI166180 PI166333 PI177943 297 PI349512 PI366716 PI366905 PI382150 PI406517 PI445736 PI470817 PI477870 P I481718 PI481923 PI565213 PI82469 PI8813 PR267 Roemer Taxi Utmost acc1 acc2 a cc3 acc4 acc5 berkut chakwal86 cham6 clear_white dharwar_dry hidhab klein_cha

3929455_1al 1623 . T C 245.53 . AC=18; AF=0.196; AN=92; BaseQRankSum=0.079; DP=48 ;Dels=0.00;FS=0.000;HaplotypeScore=0.1087;InbreedingCoeff=0.2057;MLEAC=18;MLE AF=0.196;MQ=100.00;MQ0=0;MQRankSum=-1.426;QD=27.28;ReadPosRankSum=-0.158 GT:A D:DP:GQ:PL 0/0:1,0:1:3:0,3,41 0/0:1,0:1:3:0,3,41 1/1:0,1:1:3:41,3,0 1/1:0,1:1 :3:41,3,0 ./. 0/0:1,0:1:3:0,3,41 0/0:1,0:1:3:0,3,39 ./. 0/0:1,0:1:3:0,3,39 ./ . ./. 1/1:0,1:1:3:39,3,0 0/0:1,0:1:3:0,3,39 ./. 1/1:0,1:1:3:38,3,0 1/1:0,1:1:









Wheat Data Interoperability Guidelines

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Use cases (

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Ontologies & Vocabularies

In the context of Research Data, the use of vocabularies play a key role in managing, sharing and publishing data. Vocabularies enhance the quality of the interoperability and effectiveness of data exchange, thus facilitating the re-usage of data by others and in the process adding value to the local researcher.

This section focuses on vocabularies, their benefits and current situation in the context of Wheat Research Data. The aim is to provide a tool to support researchers in the selection of vocabularies to adopt according to the Wheat Data Interoperability Guidelines.

What type of vocabularies exist in the context of the Wheat Initiative?

There are different types of vocabularies like ontologies, thesauri, classification systems, controlled lists, syntax encoding standards, authority data, controlled vocabularies, taxonomies, glossaries, etc.

Why are vocabularies important for the Wheat Initiative?

What benefits can vocabularies bring to your daily work as a researcher?

They are many, including:

- > research visibility
- > research usage
- research uptake

What are currently the most used and relevant vocabularies in the context of Wheat Initiative?

From December to 2014 to January 2015 the editorial team conducted a survey "Towards a Comprehensive Overview of Ontologies and Vocabularies for Research on Wheat". The objective was to collect information about the visibility, interoperability, domain, content and other technical aspects of relevant ontologies and vocabularies. As a result, in February 2015 a report (link) was published, and also a list of vocabularies listed as follows:



Why adopt the WDI guidelines?



Benefits for 3 main target users

As a data producer or manager

- Easily conform to the well-recognized data repositories and facilitate the deposit of your data within these repositories;
- Share common meanings of the words you utilize to describe your data and make your data more machine-readable and computable
- Contribute to foster the development of smarter search tools and make your data more visible and discoverable

As a wheat related information system or tool developer

 Basing your tool or information system on the recommended data formats and vocabularies will make it easier to integrate data from various data sources, deliver smarter outputs for a wider audience

As a wheat related ontology developer

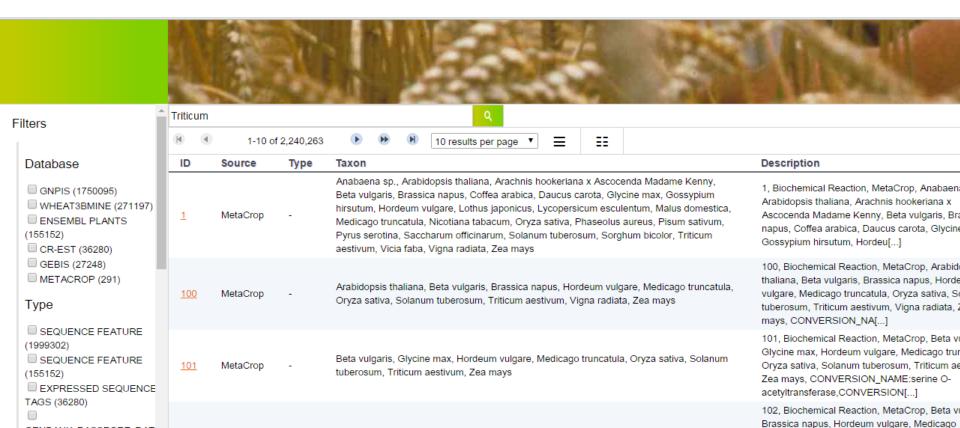
- Share your ontologies through the WDI wheat ontologies portal and make them more visible to the community
- Reuse or link your ontologies to existing concepts and terms in wheat related ontologies to enrich them, make them more visible and in some cases save you time.



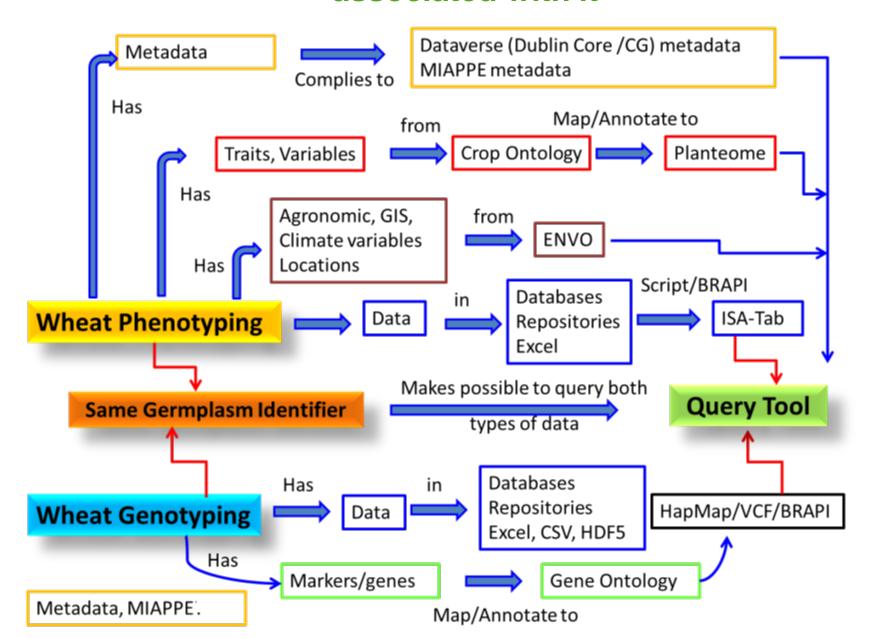
The international wheat information system

(WheatIS: https://urgi.versailles.inra.fr/wheatis/)

- Provide a single-access web based system to access to the available data resources and bioinformatics → work in progress
- 6 nodes already connected to the WheatIS search. Work in progress to connect more nodes. More information in http://wheatis.org/WheatIS%20nodes.php



Search for stem rust resistant Germplasm and genes associated with it



Benefits of data integration

- Enriches data and enables data analysis and visualization
- Demo with QTLNetMiner (https://ondex.rothamsted.ac.uk/QTLNetMiner/)
 - QTLNetMiner is one of the nodes of the WheatIS
 - Use case: search for candidate genes of "drought tolerance"







 Use case: explore relationships between the following concepts: "root development", "triticum aestivum" and "triticum urartu"





Use the query suggestor to find alternative search queries to improve your results



	drough	ht tolerance		Search	?
± Query Suggestor 1464 documents and 23845 genes will be found with this query ?					
∃ Genome or QTL Sea	arch				?
Gene List					?
				Define a QT	L region
otal 23845 genes were ry was found in 1464 d				you are inte	rested in
Map View Ge	ene View	Evidence View	Network View		
ownload as TAB delimite			Onday natwork		2
elect gene(s) and click "S	Show Networ	rk Dutton to see the	Ondex network.		

Include a list of gene names and see if they are related to your keyword

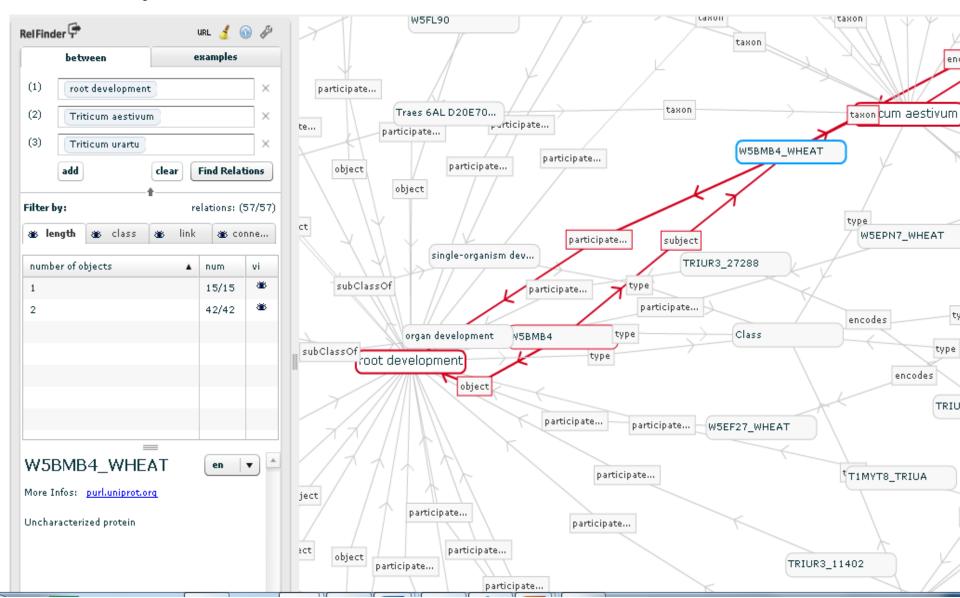


In total 23845 genes were found. Top 100 genes are displayed in Map view. Query was found in 1464 documents related with genes (3012 documents in total) Gene View Evidence View Network View 18 Map View Download as TAB delimited file ? Select gene(s) and click "Show Network" button to see the Ondex network. Max number of genes to show: 100 ▼ Known targets: Novel targets: **ACCESSION** ♦ Select ♦ TRAES_4DL_6063F821A 4D 8.36 2.20 no 0 <u>A</u> 1 4 2 1 TRAES_4BL_46BE39DD3 4B 74.63 2.20 no 0 <u>A</u>1421 Gene 4 2 1 TRAES_4AS_13CE9F8B7 4A 19.89 2.20 no 0 <u>△</u> 10 € TRAES_7BL_57F31555F 0 7B 167.18 1.83 no 2.57 1.74 TRAES_5AL_5D65F5235 5A TRAES_3B_2A5C09288 3B 1.6 68.30 2D TRAES_2DL_574F25C6E 76.53 1.63 66.45 TRAES 7BS 9F5A66A59 7B 1.63 1.5 TRAES 6AS C1F384744 6A 0.00 1.5 TRAES 6BS 04D7C6A64 65.07 1.56 0.00 TRAES_7DS_32CBC58AD | 7D **Network view** research data sharing without barriers rd-alliance.org

Agronomic Linked Data (AgroLD)

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Search > Explore



The more the wheat research community harmonize its practices in terms of data management, the more IS and tools like the WheatIS, QTLNetMiner and AgroLD can integrate data and provide valuable knowledge



How to adopt the guidelines?



Data formats

- For legacy data
 - Please provide your data in at least one of the recommended data formats even if, for some reasons, you need to also keep them in other non-recommended formats
- For future developments
 - Please consider using the recommended data formats from the beginning.
- Example: provide your sequence variation data in the latest VCF file format
 - Please refer to the <u>WDI guidelines</u> for precise recommendations on each data type



Best practices on data description and sharing

- Describe your data with the recommended metadata standards and annotate your data with the recommended vocabularies.
 - Examples:
 - For genome annotation data in GFF3 format, use of ontologies for functional annotation in column 9, such as, Gene Ontology and Sequence Ontology.
 - For observation Variables (including trait and environment variables), use existing variables, listed in the following vocabularies and ontologies:
 - Wheat crop ontology
 - Wheat INRA Phenotype Ontology (previously INRA Wheat Ontology)
 - Biorefinery ontology
 - XEO, XEML Environment Ontology
- Deposit your data in the WheatIS data repository or well established data repositories



Adopting Agroportal

- Share your wheat related ontologies within the <u>WDI</u> slice in Agroportal
- Before developing a new ontology
 - Make sure there is not an existing one within the WDI slice in Agroportal that covers your needs
- When developing a new ontology
 - Please reuse or link to exiting concepts and terms in the ontologies within the <u>WDI slice in Agroportal</u> whenever possible.
 - Please align your ontologies to the existing ones within the <u>WDI slice in</u> <u>Agroportal</u> and share the mapping results



Endorsements/Adopters

Laboratory	Contact		
NIAB,	Professor Mario Caccamo		
	Head of Crop Bioinformatics		
USDA ARS and Cold Spring Harbor Laboratory,	Doreen Ware		
	Adjunct Associate Professor		
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Australian Center for Plant Functional Genomics,	Dr Baumann, Ute		
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The Genome Analysis Center,	Robert Davey		
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	Rosemary Shrestha, Data Coordinator		

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And

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Thank you!

