

## GEPHE SUMMARY

HvbHLH1 ( <a href="https://www.gephebase.org/search-criteria/?and+GeneGephebase=%HvbHLH1">#gephebase-summary-title</a> )	Gephebase Gene	GP00000493	GephelD
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria/?and+TraitCategory=%Morphology">#gephebase-summary-title</a> )	Trait Category		
Coloration ( <a href="https://www.gephebase.org/search-criteria/?and+Trait=%Coloration">#gephebase-summary-title</a> )	Trait		
Hordeum vulgare - Saffron	Trait State in Taxon A		
Hordeum vulgare - Retriever	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria/?and+TaxonomicStatus=%Domesticated">#gephebase-summary-title</a> )	Taxonomic Status		
Hordeum vulgare ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Hordeum+vulgare">#gephebase-summary-title</a> )	Latin Name		Latin Name
-	Common Name		Common Name
barley; Hordeum vulgare L.; Horedum vulgare species	Synonyms		Synonyms
cellular organisms; Eukaryota; Viridiplantae; Streptophyta; Streptophytina; Embryophyta; Tracheophyta; Euphyllophyta; Spermatophyta; Magnoliophyta; Mesangiospermae; Liliopsida; Petrosaviidae; commelinids; Poales; Poaceae; BOP clade; Pooideae; Triticodae; Triticeae; Hordeinae; Hordeum	Rank		Rank
Hordeum () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4512">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4512</a> )	Lineage		Lineage
4513 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4513">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4513</a> )	Parent		Parent
Yes	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
Hordeum vulgare - Saffron	Taxon A Description		Taxon B Description
	Hordeum vulgare - Retriever		

## GENOTYPIC CHANGE

bHLH1	Generic Gene Name	UniProtKB Hordeum vulgare subsp. vulgare E5FCX3 ( <a href="http://www.uniprot.org/uniprot/E5FCX3">http://www.uniprot.org/uniprot/E5FCX3</a> )
-	Synonyms	GenebankID or UniProtKB ADR30773 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/ADR30773">https://www.ncbi.nlm.nih.gov/nuccore/ADR30773</a> )
-	String	
-	Sequence Similarities	
GO:0046983 : protein dimerization activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046983">https://www.ebi.ac.uk/QuickGO/term/GO:0046983</a> )	GO - Molecular Function	
-	GO - Biological Process	
-	GO - Cellular Component	
		Presumptive Null

Yes ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=%Yes))

Molecular Type

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

Aberration Type

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

Deletion Size

10-99 bp

Molecular Details of the Mutation

16bp deletion resulting in premature stop codon

Experimental Evidence

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

Main Reference

Genome-wide association mapping to candidate polymorphism resolution in the unsequenced barley genome. (2010) (<https://pubmed.ncbi.nlm.nih.gov/21115826/>)

Authors

Cockram J; White J; Zuluaga DL; Smith D; Comadran J; Macaulay M; Luo Z; Kearsey MJ; Werner P; Harrap D; Tapsell C; Liu H; Hedley PE; Stein N; Schulte D; Steuernagel B; Marshall DF; Thomas WT; Ramsay L; Mackay I; Balding DJ; Waugh R; O'Sullivan DM

Abstract

Although commonplace in human disease genetics, genome-wide association (GWA) studies have only relatively recently been applied to plants. Using 32 phenotypes in the inbreeding crop barley, we report GWA mapping of 15 morphological traits across  $\approx 1/4500$  cultivars genotyped with 1,536 SNPs. In contrast to the majority of human GWA studies, we observe high levels of linkage disequilibrium within and between chromosomes. Despite this, GWA analysis readily detected common alleles of high penetrance. To investigate the potential of combining GWA mapping with comparative analysis to resolve traits to candidate polymorphism level in unsequenced genomes, we fine-mapped a selected phenotype (anthocyanin pigmentation) within a 140-kb interval containing three genes. Of these, resequencing the putative anthocyanin pathway gene *HvbHLH1* identified a deletion resulting in a premature stop codon upstream of the basic helix-loop-helix domain, which was diagnostic for lack of anthocyanin in our association and biparental mapping populations. The methodology described here is transferable to species with limited genomic resources, providing a paradigm for reducing the threshold of map-based cloning in unsequenced crops.

Additional References

## RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS