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Detecting directional and epistatic selection from candidate genes: methodological improvements and a case study of European beech

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SUMMARY

SIGNATURE OF SELECTION AT SINGLE- AND MULTILOCUS LEVELS

- accounting for the uncertainty of haplotype inference in F_{ST} outlier tests
- re-discovering Ohta's test of epistatic selection for candidate genes

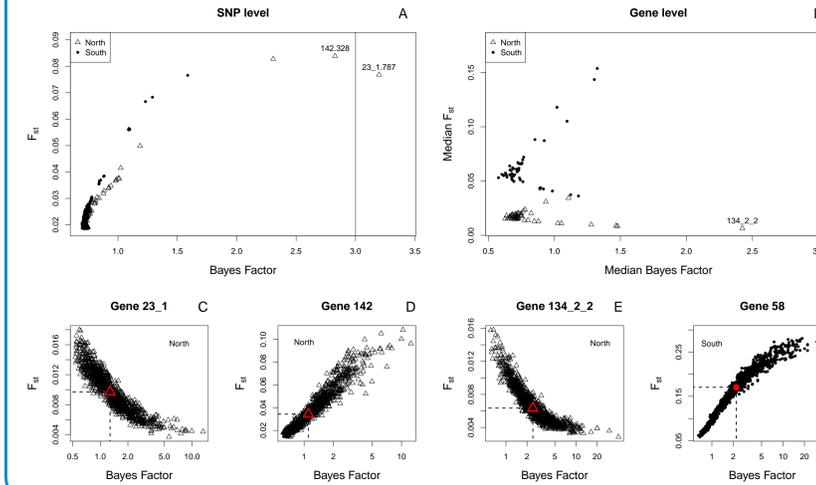
A CASE STUDY OF *Fagus sylvatica*

- sampling at a **short spatial scale** with sharp environmental differences
- SNPs from candidate genes potentially involved in **climate response**



See more details in: K Csilléry, H Lalagüe, GG Vendramin, SC González-Martínez, B Fady and S Oddou-Muratorio 2014 Detecting local adaptation and epistatic selection in climate related candidate genes at a short spatial scale in European beech (*Fagus sylvatica* L.) populations. *Molecular Ecology* 23: 4696-4708

RESULTS: DIRECTIONAL SELECTION

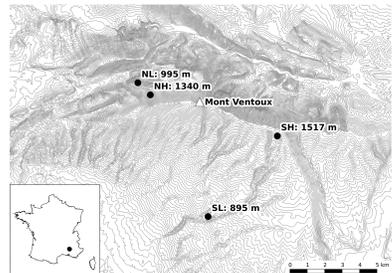


CONCLUSIONS:

- outlier detection at the SNP (A) vs gene level (B) revealed **different loci** under selection
- gene level outlier detection was strongly influenced by **uncertainty in haplotype reconstruction** (C-F)
- uncertainty of haplotype inference can be accounted for by **averaging Bayes factors** over many possible phase reconstructions (B)

MATERIALS & METHODS

SAMPLING SITES



NL: North Low, NH: North High, SL: South Low, SH: South High

DETECTING DIRECTIONAL SELECTION:

F_{ST} outlier test at the SNP and candidate gene levels with Bayescan¹
 For gene level tests, haplotype phase was estimated using PHASE²

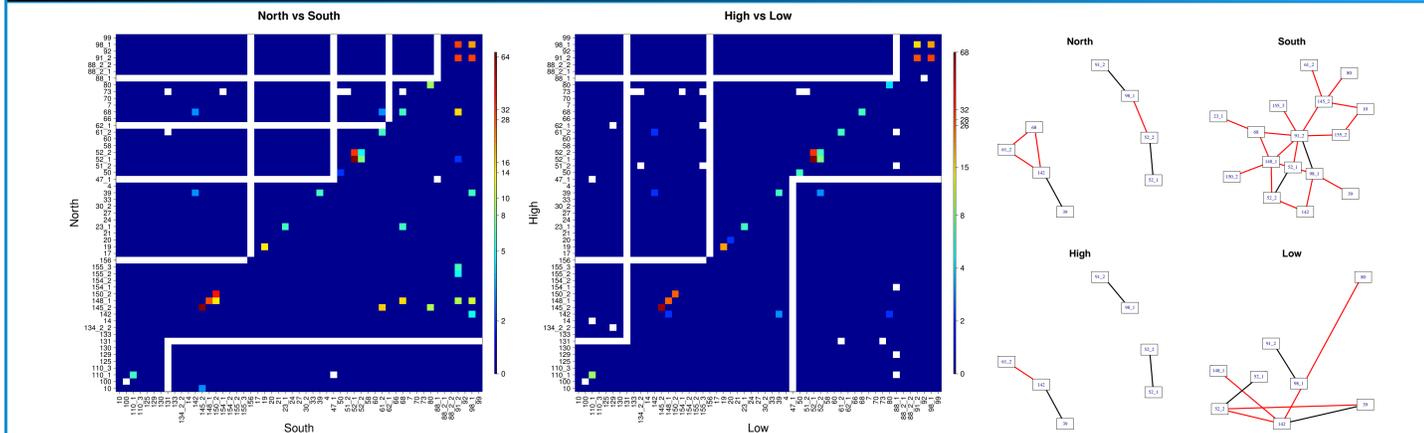
DETECTING EPISTATIC SELECTION:

Following Ohta³, we decomposed the variance of linkage disequilibrium within a subdivided population into between and within population components:

- D_{IS}^2 : within subpopulations
- D_{ST}^2 : two loci of different gametes in a subpopulation relative to the total population
- D_{IS}^2 : two loci of one gamete in a subpopulation relative to the total population
- D_{ST}^2 : total population

Ohta's test: epistatic selection is more likely than drift if $D_{ST}^2 < D_{IS}^2$ and $D_{IS}^2 < D_{ST}^2$

RESULTS: EPISTATIC SELECTION



Gene pairs "light-up" in red if they contain at least an SNP that show a signal of epistatic selection between them. Genes in the diagonal show within-gene epistatic selection signal. Networks are drawn between genes that show a unique between-gene epistatic selection signal.

WHAT ARE EPISTATIC NETWORK GENES CODE FOR?

Key genes with unique between gene epistatic selection signal in North/High (N/H) and South/Low (S/L) population pairs:

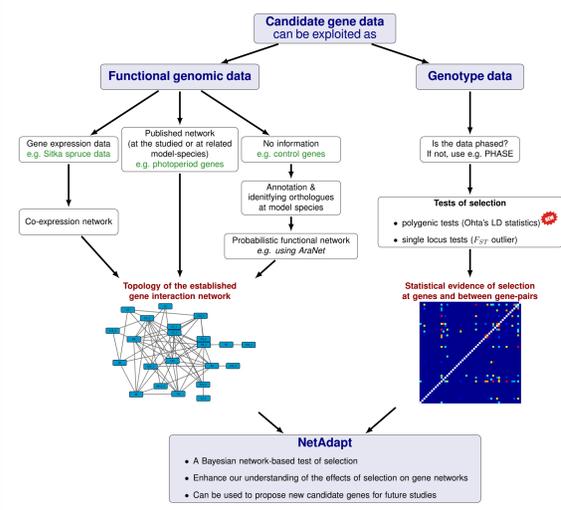
- (N/H) **Gene 68** is connected **61_2** and **142** via two non-synonymous SNPs. **Gene 61_2** is a member of the heat shock protein 70 family and **68** catalyzes glycolysis, both play a key role in stress response.
- (S/L) **Gene 50**'s SNP was situated in a 3'UTR region and the gene codes a major transcription factor in response to abiotic stress and has been shown to respond to cold temperatures.
- (S/L) **Gene 80** regulates stomatal closure (key importance in response to drought) and has been suggested to play a role in dormancy.
- (S/L) **Genes 148_1** and **145_2** are well-known budburst candidate genes.

WHY DID IT WORK?

Ohta's test has been relatively little used and most studies found no signal of epistatic selection, so why did it work here?

- **Recent selection:** *F. sylvatica* populations re-colonized Mont Ventoux about five generations ago⁶ and since have been exposed to sharp environmental differences
- **Functionally related genes** favored the build-up and maintenance of LD due to epistatic selection
- **Sampling from sharply different environments:** 0.23% of the SNP pairs showed evidence of epistatic selection, with nearly 80% of them being within genes. However, most epistatic interactions unique to a population pair (N, S, H, or L) were observed between different genes. Indeed, most systematically mapped **epistatic interactions between different genes bring new functionality that may only be advantageous in a particular environment**⁷

NETADAPT: A FUTURE TEST FOR CANDIDATE GENES



POLYGENIC ADAPTATION

- Most traits that potentially play a role in adaptation are controlled by many genes
- Genes do not act independently, but interact through developmental, metabolic and biochemical networks⁴

THE ROLE OF EPISTASIS IN ADAPTATION

- **Negligible?** The elevated frequency of co-occurrence of beneficial allele combinations at different genes is expected to be continuously broken down by recombination.
- **Not necessarily!** However, if genes carrying beneficial allele combinations are also functionally connected via networks, the statistical signal of epistatic selection may be maintained⁵

KEY REFERENCES

¹Foll and Gaggiotti. *Genetics* 180.2 (2008): 977-993; ²Stephens and Scheet. *AJHG* 76 (2005):449-462; ³Ohta. *PNAS* 79.6 (1982): 1940-1944; ⁴Mackay. *Nat. Rev. Genet.* (2013); ⁵Hansen. *Evolution* 67.12 (2013): 3501-3511; ⁶Lander et al. *Mol. Ecol.* 20.24 (2011): 5182-5196; ⁷Lehner. *Trends in Genet.* 27.8 (2011): 323-331.

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