Polygenic Adaptation: Introduction and Case Study (maritime pine)

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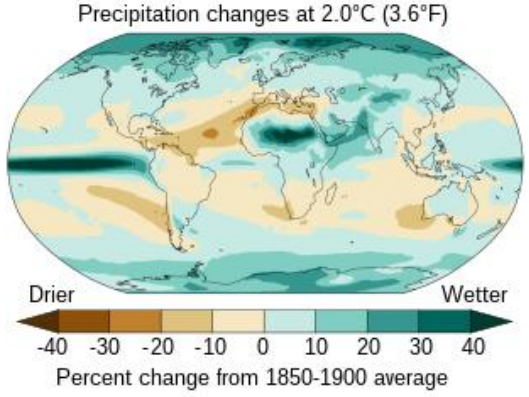
Adaptation to new environmental conditions



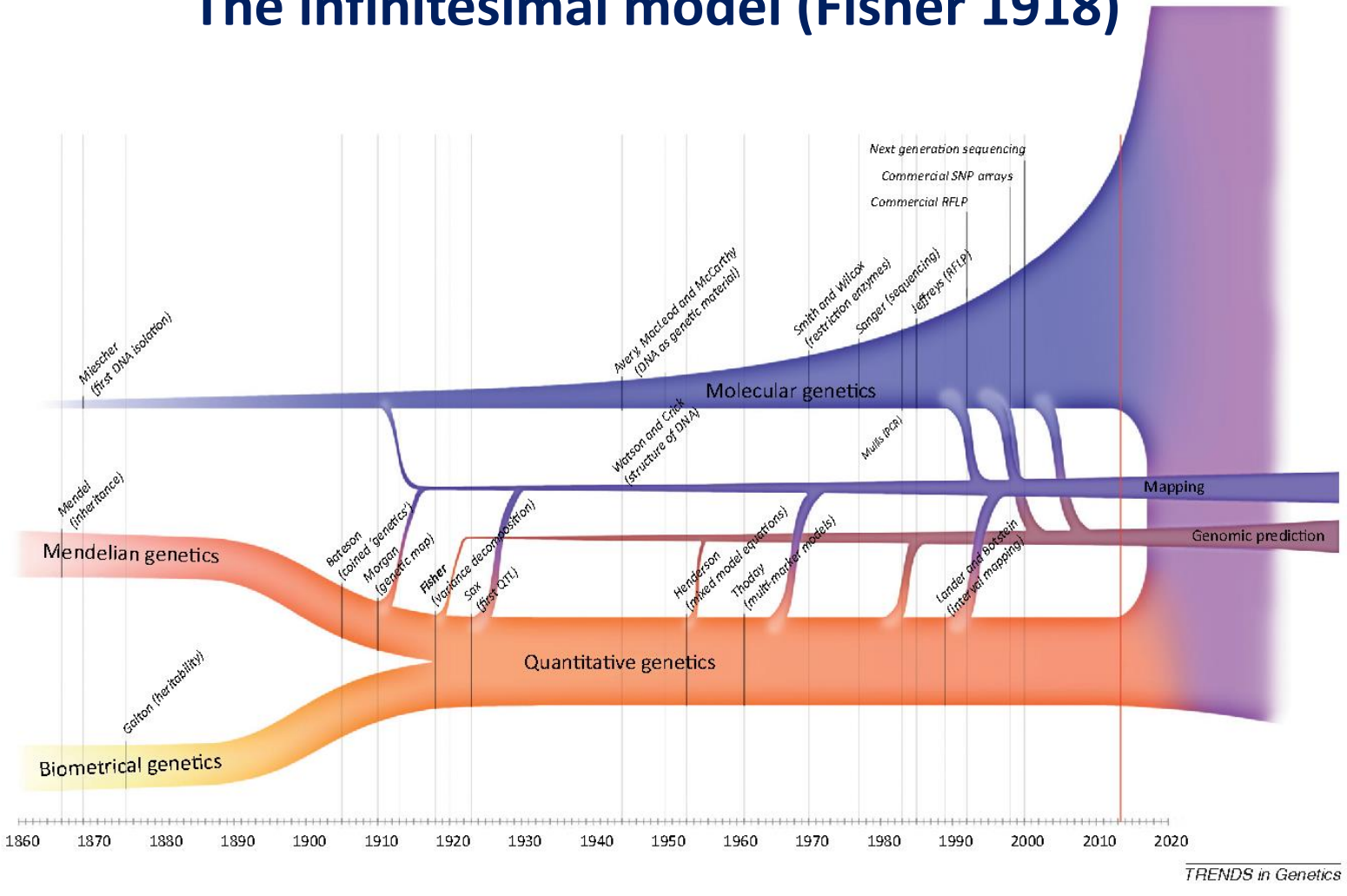
Past **Present** **Future**

Understanding how species have adapted

Knowledge on how species will adapt

genetic architecture of fitness-related traits

The infinitesimal model (Fisher 1918)



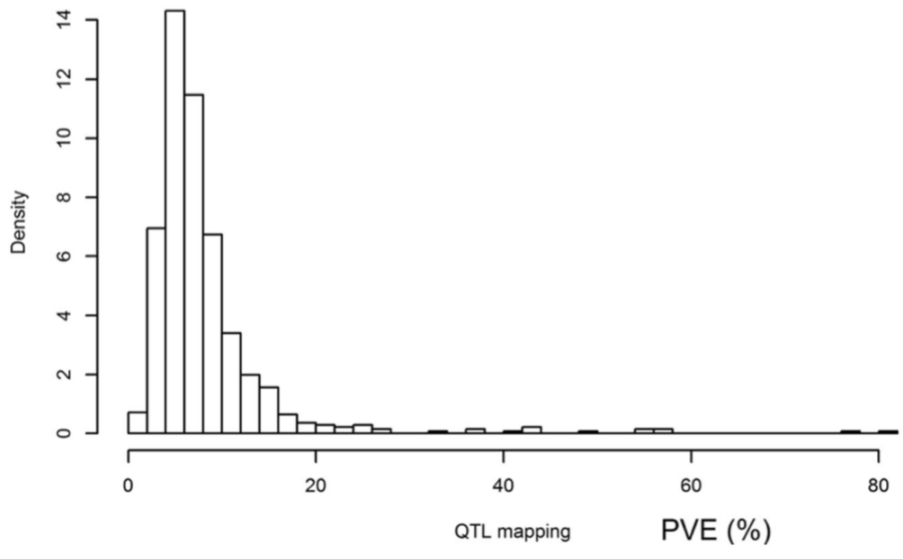
Nelson et al. 2013 *TIG*

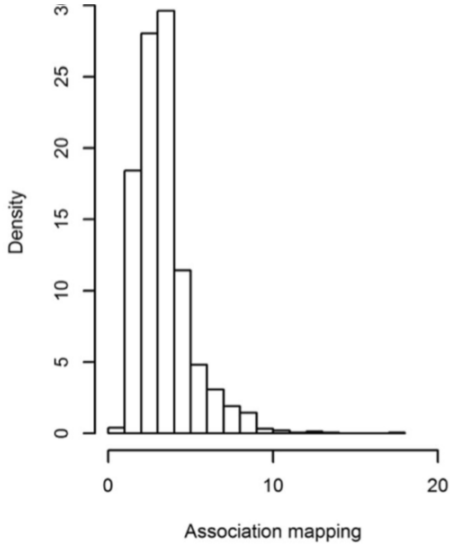
Knowledge from genotype-phenotype association studies

GWAS and QTL analysis ⇨ most traits are polygenic

Genetic architecture of complex traits:

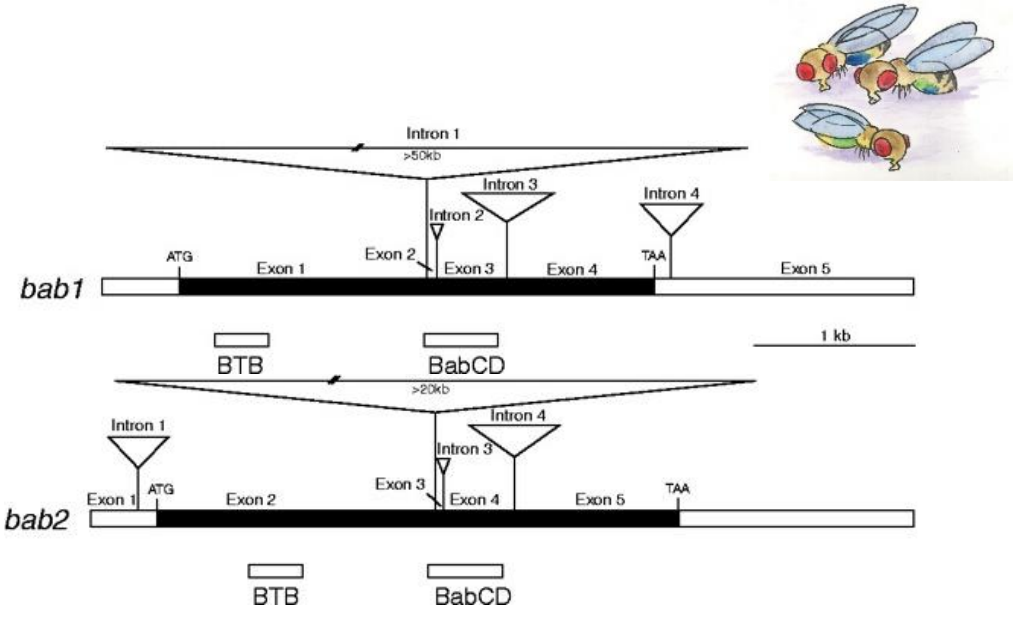
↑ genes ↓ effect size ↓ PEV





Hall et al. 2016 *TGG*

Case study from Remington 2015 *Evolution*



The ***bab*** locus is responsible for a major effect QTL involving 60% of the variation in female abdominal pigmentation in *D. melanogaster.*

The ***bab*** locus is responsible for a major effect QTL involving 60% of the variation in female abdominal pigmentation in *D. melanogaster*, but single SNPs explain each <1% of variation, making them basically undetectable by standard GWAS

Table 1. Estimation of polymorphism and region effect size.

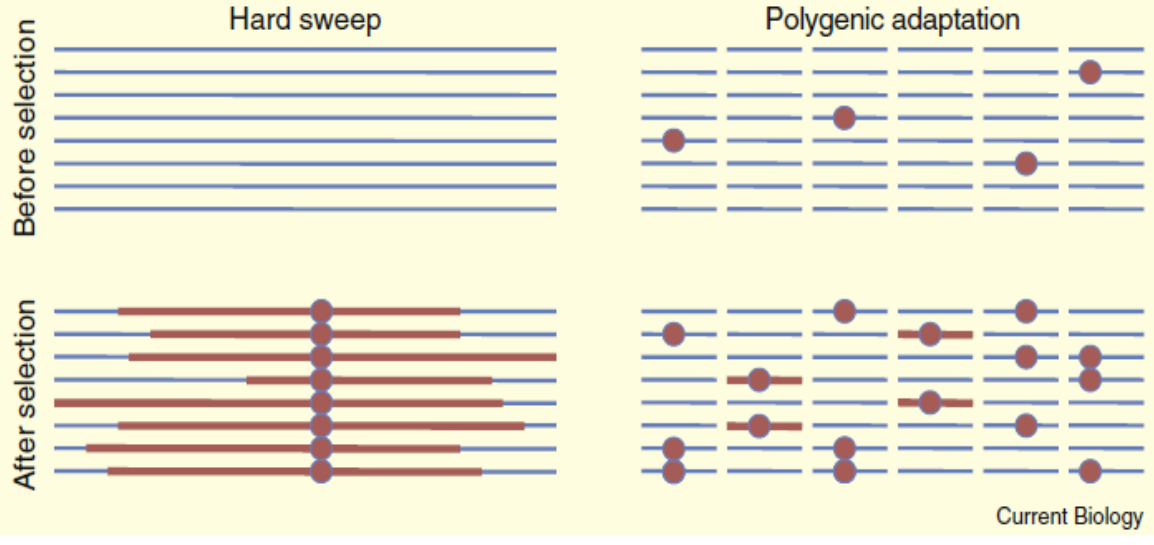
	Number of sites	Average effect size of each polymorphism	Proportion of total variation explained by average polymorphism	Total estimated effect of region	Proportion of total variation explained
Region 1	55	0.80 (.19)	0.009	44	0.512
Region 2	21	0.97 (.28)	0.011	20.37	0.237
Region 3	29	1.02 (.30)	0.012	29.58	0.344
All 3 regions	105	0.69 (.10)	0.008	72.45	0.842

doi:10.1371/journal.pgen.1001275.t001

Bickel et al. 2011 *PLoS Genetics*

Polygenic adaptation model

From hard sweeps to soft sweeps: **Polygenic adaptation model**



Pritchard et al. 2010 *Current Biology*

Evidences of polygenic adaptation

Evidence of widespread selection on standing variation in Europe at height-associated SNPs

Michael C Turchin[1–5,8], Charleston WK Chiang[1–6,8], Cameron D Palmer[1–5], Sriram Sankararaman[5,6], David Reich[5,6], Genetic Investigation of ANthropometric Traits (GIANT) Consortium[7] & Joel N Hirschhorn[1–6]

Genetic redundancy fuels polygenic adaptation in *Drosophila*

Neda Barghi[1], Raymond Tobler[1,2¤a], Viola Nolte[1], Ana Marija Jakšić[1,2¤b], François Mallard[1¤c], Kathrin Anna Otte[1], Marlies Dolezal[1,3], Thomas Taus[1,2], Robert Kofler[1], Christian Schlötterer[1]*

Evolutionary genomics of gypsy moth populations sampled along a latitudinal gradient

Christopher J. Friedline, Trevor M. Faske, Brandon M. Lind, Erin M. Hobson, Dylan Parry, Rodney J. Dyer, Derek M. Johnson, Lily M. Thompson, Kristine L. Grayson, Andrew J. Eckert
... See fewer authors

Human adaptations to diet, subsistence, and ecoregion are due to subtle shifts in allele frequency

Angela M. Hancock[a], David B. Witonsky[a], Edvard Ehler[a,b], Gorka Alkorta-Aranburu[a], Cynthia Beall[c], Amha Gebremedhin[d], Rem Sukernik[e], Gerd Utermann[f], Jonathan Pritchard[a,g], Graham Coop[a,h], and Anna Di Rienzo[a,1]

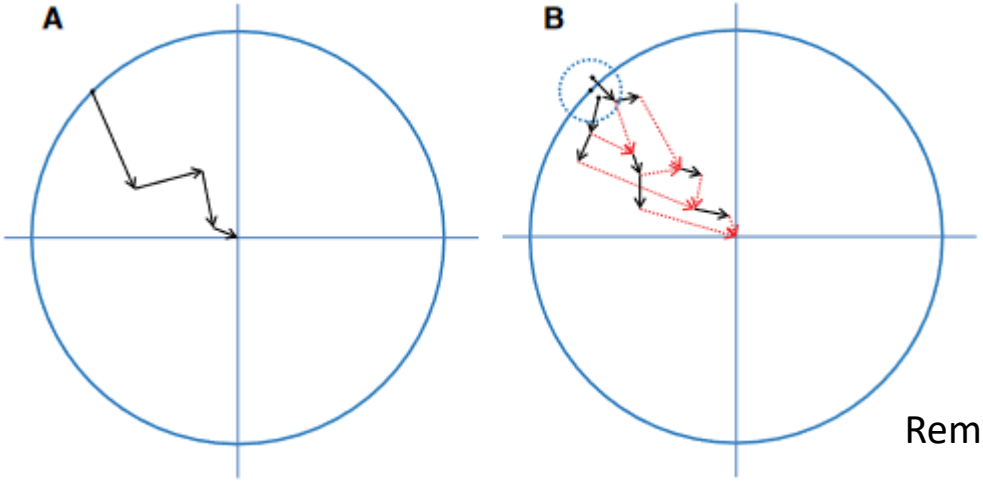
Seascape genomics of eastern oyster (*Crassostrea virginica*) along the Atlantic coast of Canada

Simon Bernatchez[1,2,3,4] | Amanda Xuereb[2] | Martin Laporte[1] | Laura Benestan[1] | Royce Steeves[3] | Mark Laflamme[3] | Louis Bernatchez[1*] | Martin A. Mallet[4*]

The Footprint of Polygenic Adaptation on Stress-Responsive *Cis*-Regulatory Divergence in the *Arabidopsis Genus*

Fei He,[1,2] Agustin L. Arce,[1] Gregor Schmitz,[1] Maarten Koornneef,[3] Polina Novikova,[4] Andreas Beyer,[2] and Juliette de Meaux*,[1]

Polygenic adaptation can be very fast



Interestingly, under a model of polygenic adaptation, phenotypic optima can be reached very fast

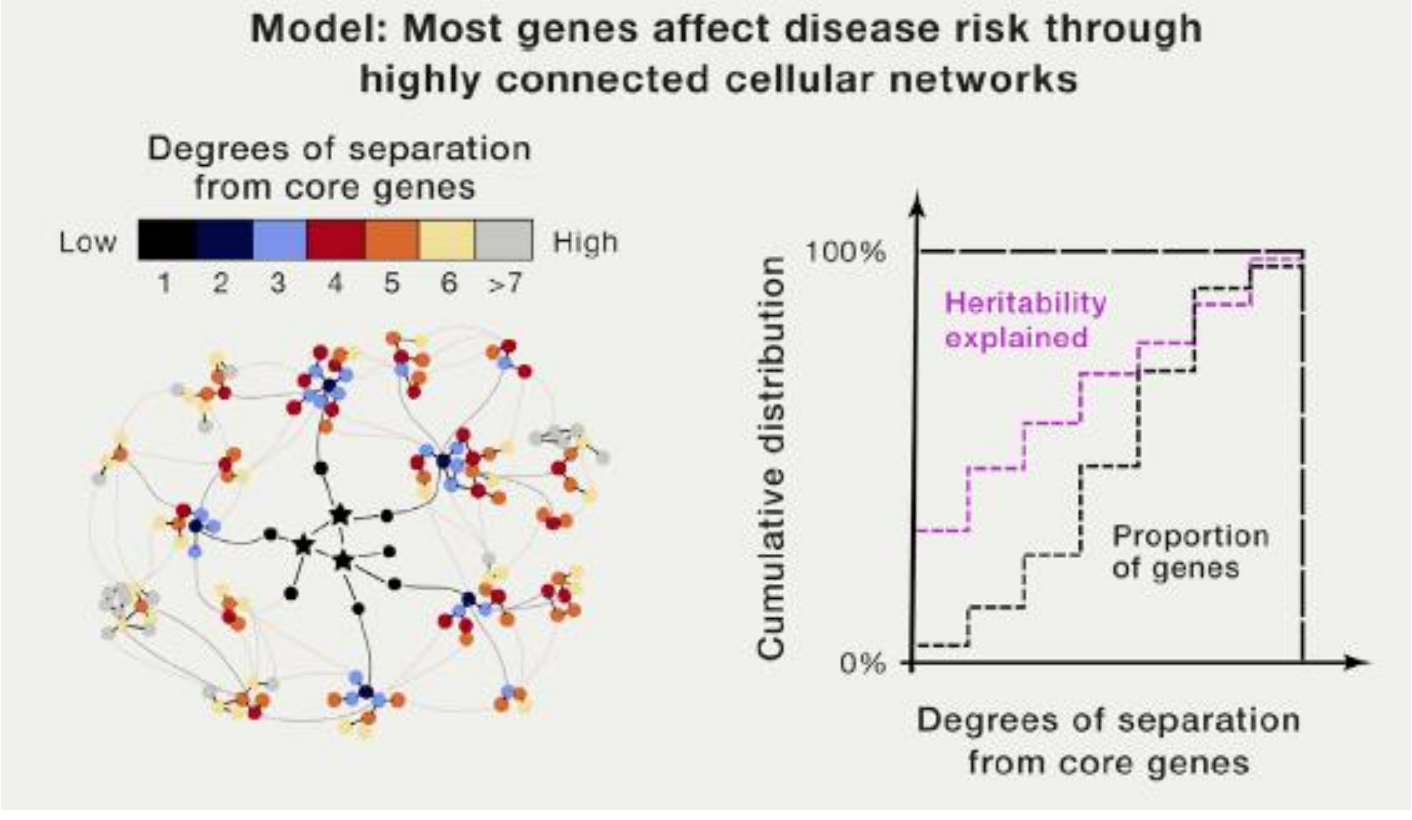
Remington 2015 *Evolution*

And many others!

Modes of Rapid Polygenic Adaptation

Kavita Jain*[,1] and Wolfgang Stephan*[,2,3]
[1]Theoretical Sciences Unit, Jawaharlal Nehru Centre for Advanced Scientific Research, Bangalore, Karnataka, India
[2]Natural History Museum Berlin, Berlin, Germany
[3]Biocenter, University of Munich, Planegg, Germany
***Corresponding authors:** E-mails: jain@jncasr.ac.in; stephan@bio.lmu.de.
Associate editor: Deepa Agashe

An expansion of the polygenic adaptation model: the omnigenic model



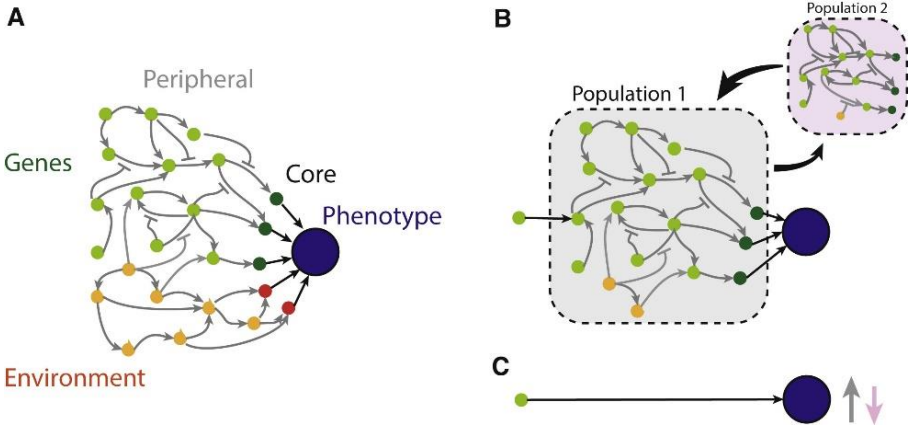
Boyle et al. 2017 *Cell*

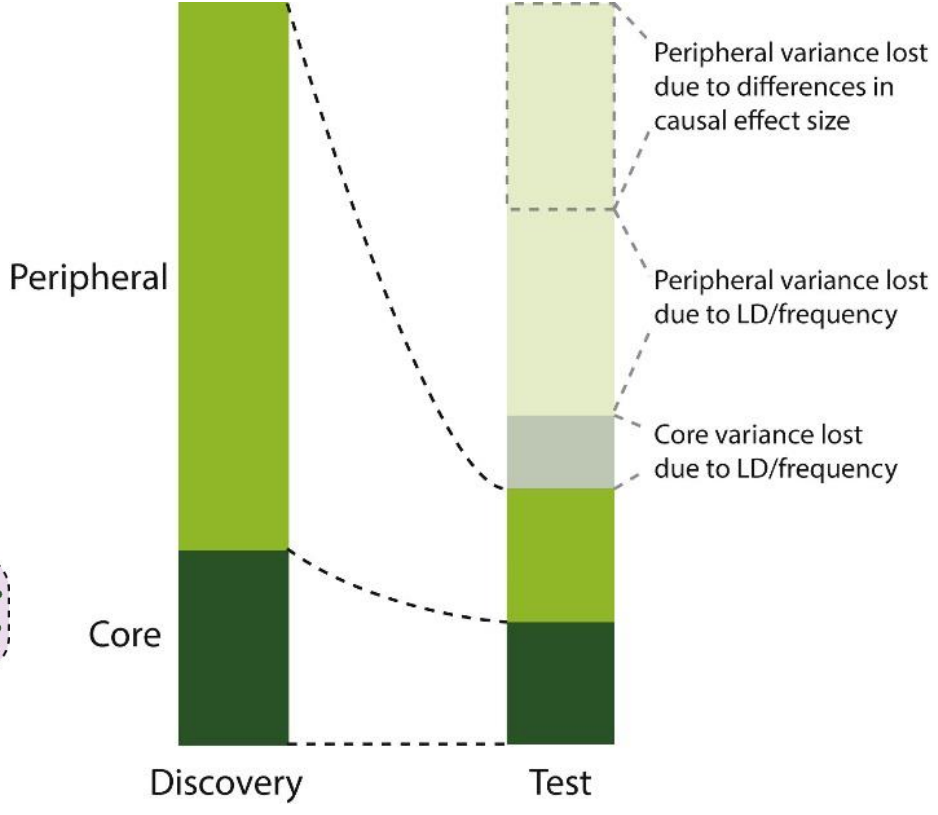
PERSPECTIVE

The omnigenic model and polygenic prediction of complex traits

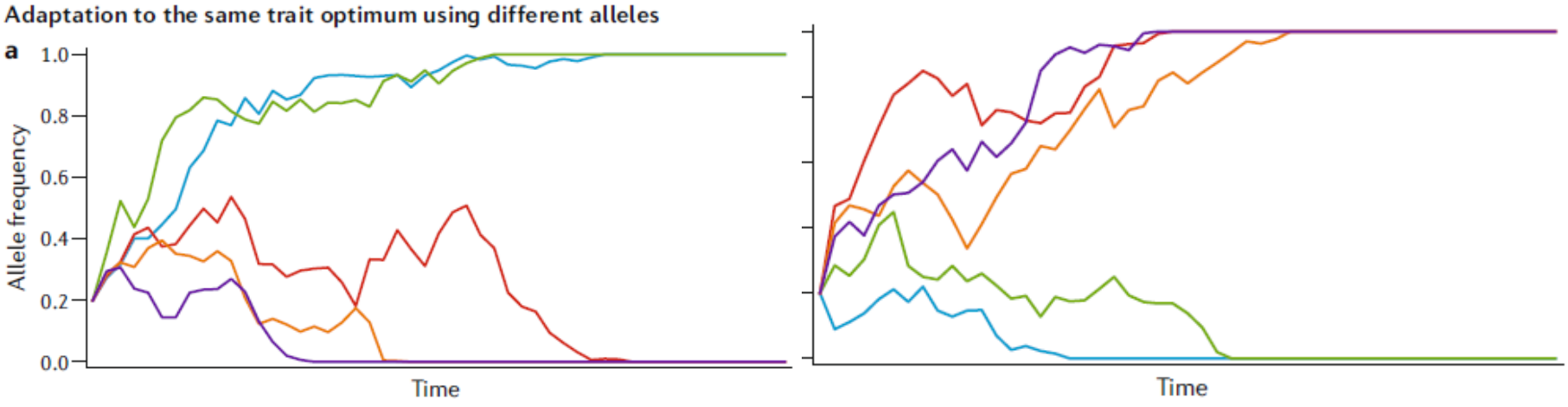
Iain Mathieson[1,*]

The omnigenic model also explains why the effects of most GWAS variants vary between populations



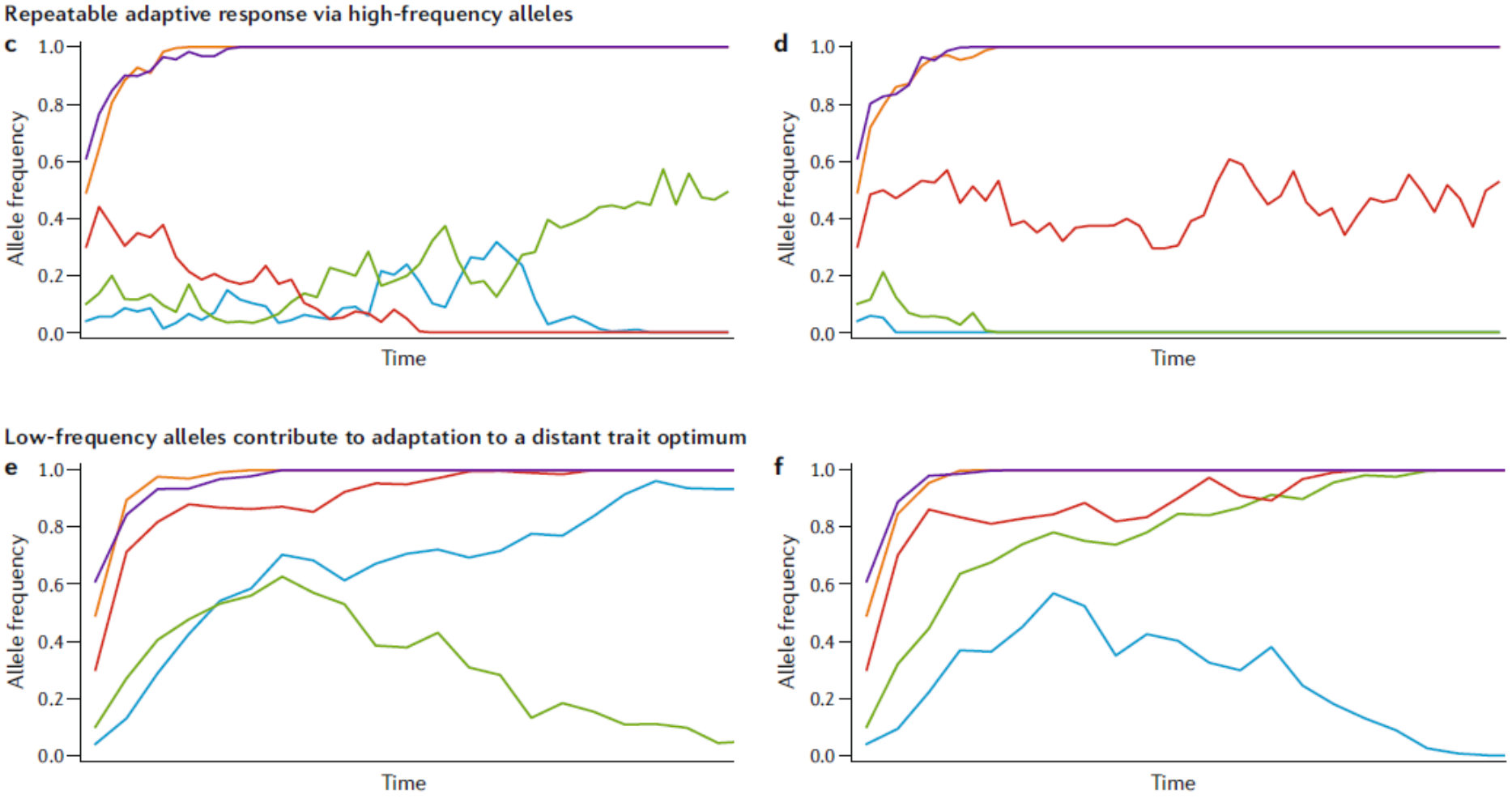


Two key features at the base of polygenic adaptation



Adaptation can be typically achieved by many different combinations of adaptive alleles (**redundancy**) → non-parallelism between replicated populations

Barghi et al. 2020 *Nature Reviews Genetics*



Heterogeneity in allele frequency among loci

Case study in maritime pine

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DOI: 10.1111/mec.16367

ORIGINAL ARTICLE

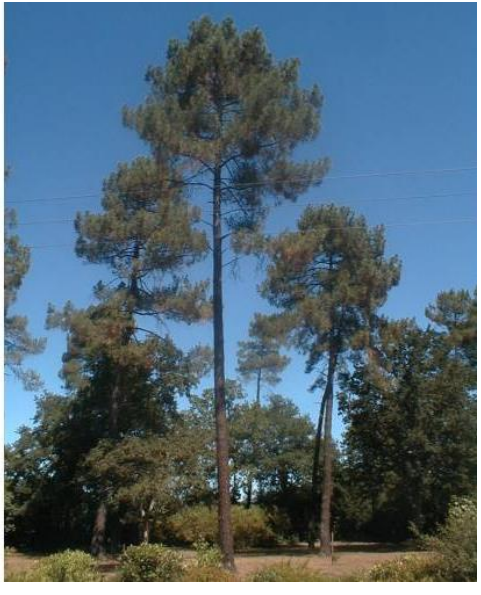
MOLECULAR ECOLOGY WILEY

Polygenic adaptation and negative selection across traits, years and environments in a long-lived plant species (*Pinus pinaster* Ait., Pinaceae)

Marina de Miguel[1,2] | Isabel Rodríguez-Quilón[3] | Myriam Heuertz[1] | Agathe Hurel[1] | Delphine Grivet[3] | Juan Pablo Jaramillo-Correa[4] | Giovanni G. Vendramin[5] | Christophe Plomion[1] | Juan Majada[6] | Ricardo Alía[3] | Andrew J. Eckert[7] | Santiago C. González-Martínez[1]

Maritime pine (*Pinus pinaster* Ait.)

- Long-lived conifer
- Ecologically and economically important in South-western Europe
- Isolated populations in Atlantic and Mediterranean environments.
- High differentiation in quantitative traits



CLONAPIN common garden network

4 identical trials + Bordeaux (smaller)

36 population

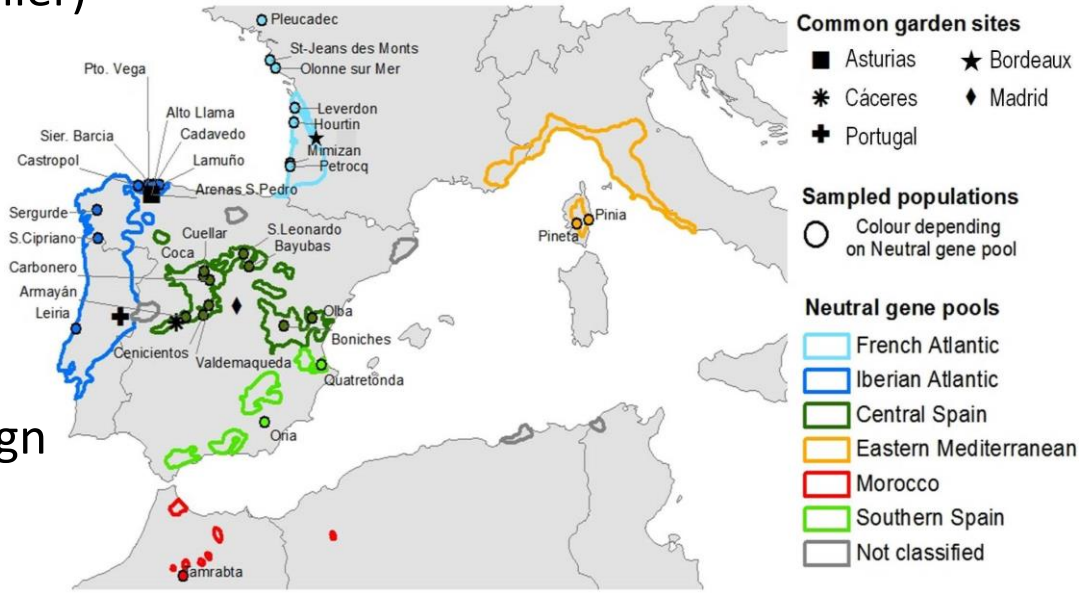
6 neutral gene pools

535 clones, 8 ramets/clone

Randomized complete block design

Plantation in 2010

2011 in Bordeaux



Data collected

Genotypes

9k Illumina Infinium SNP (Plomion et al. 2016)

Random EST sequences and candidate genes for adaptation to biotic and abiotic factors

5165 polymorfic SNPs , 523 clones

Phenotypes

Height

Survival

Phenology-related traits

Biotic stress response

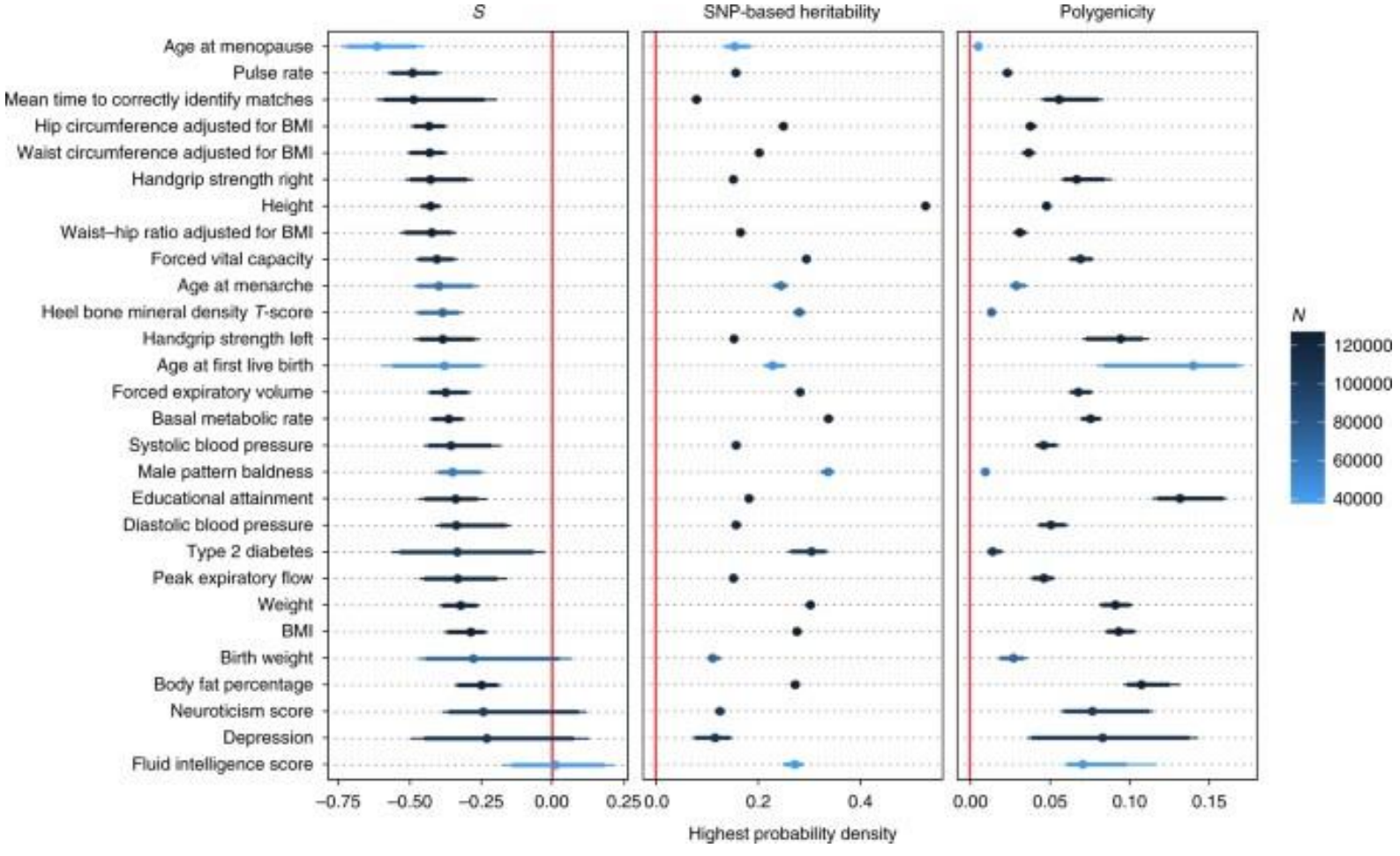
Diplodia sapinea

Armillaria ostoyae

Pine processionary moth

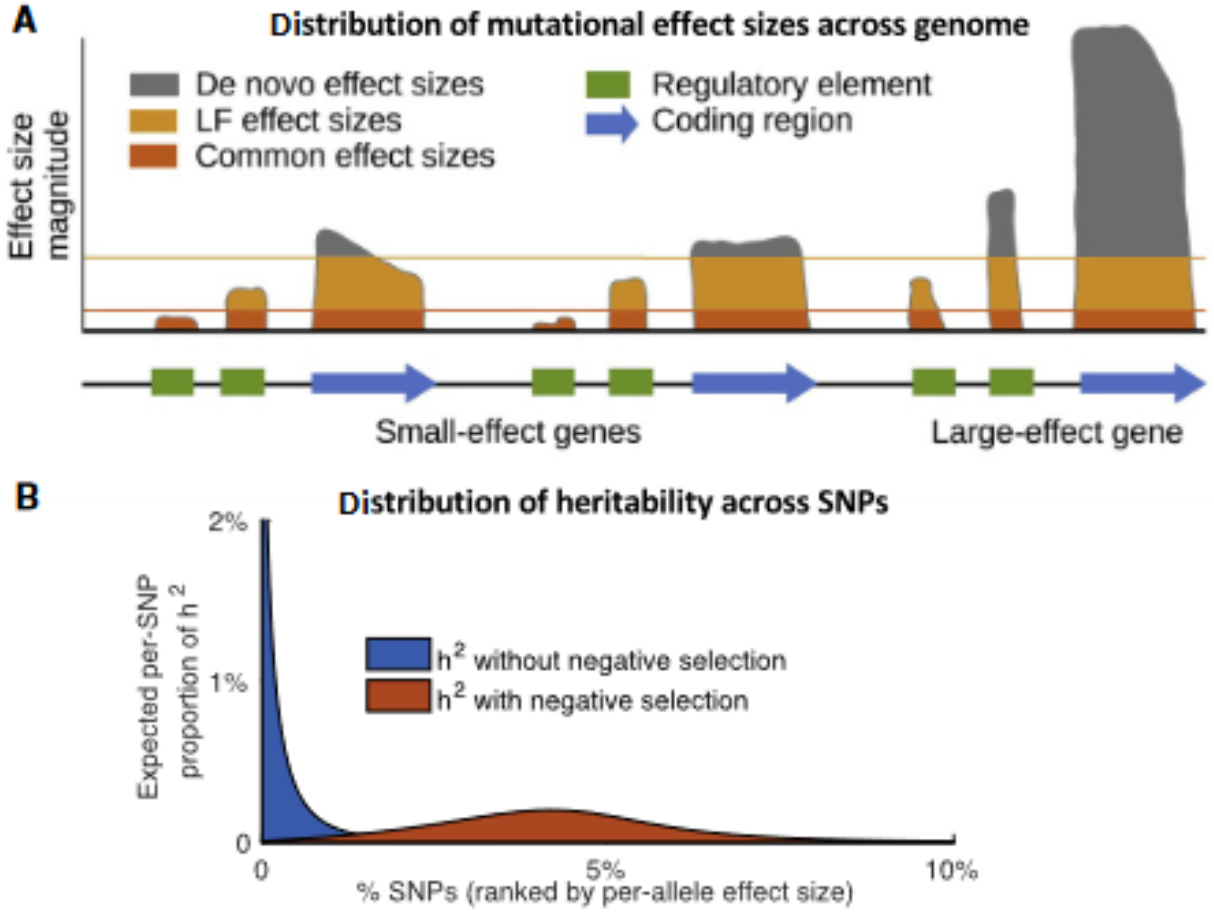
Functional traits

Polygenicity estimates using Bayesian MLMs in humans



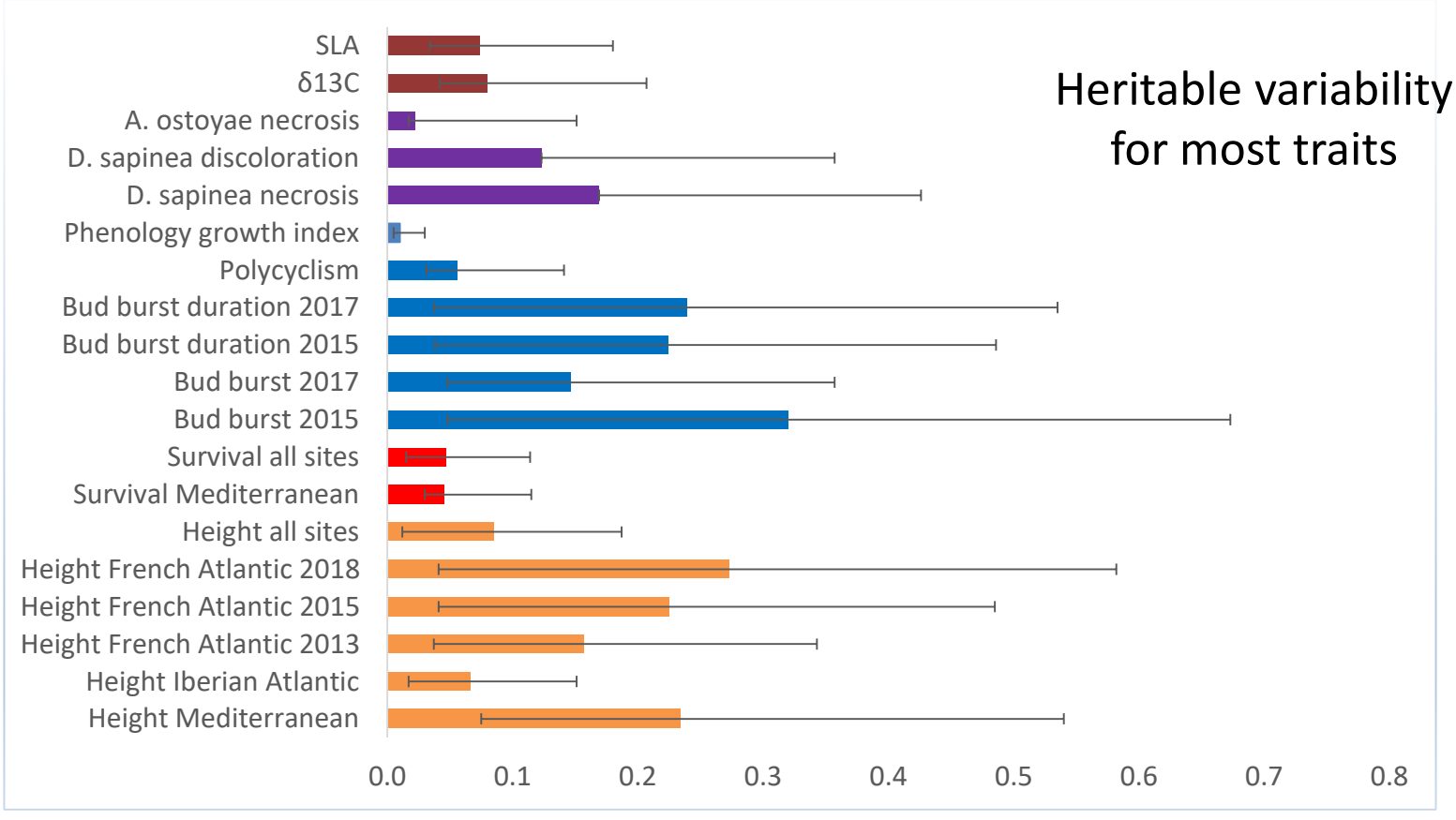
Zeng et al. 2018 *Nature genetics*

The role of negative selection

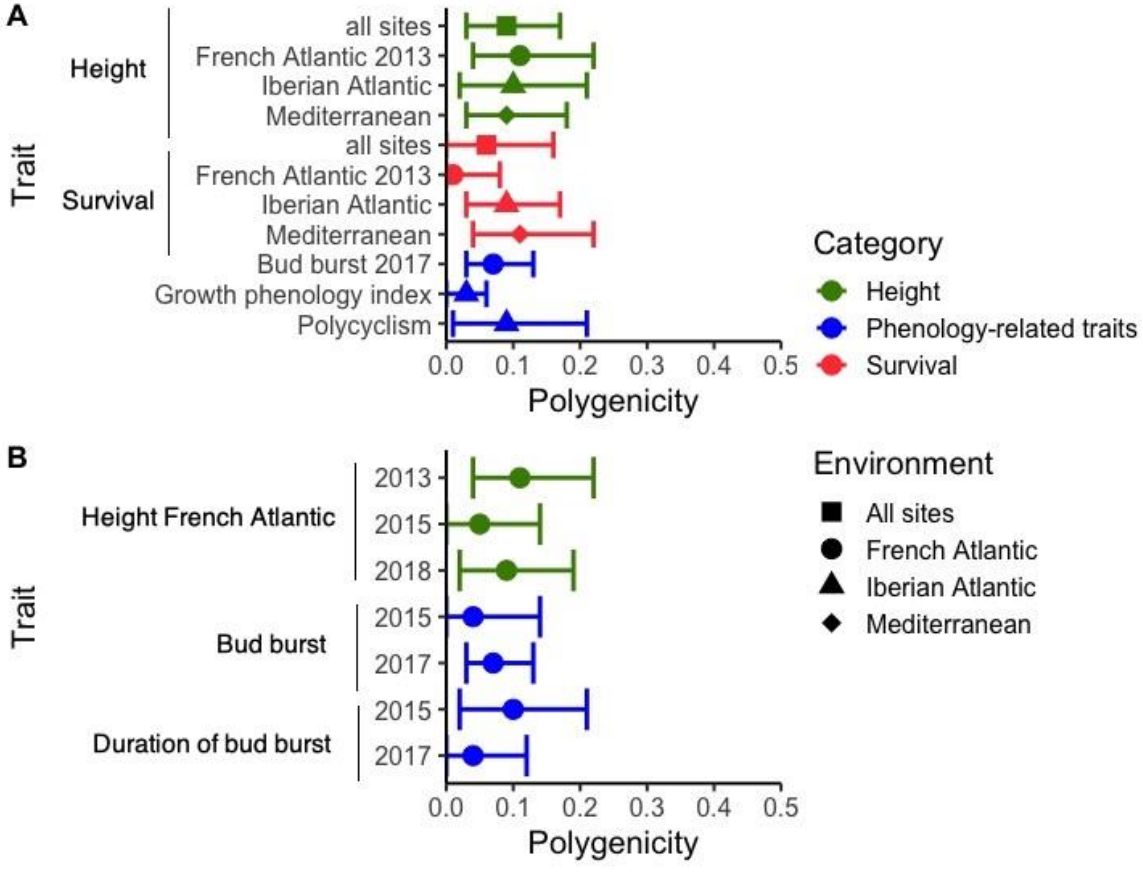


O'connor et al. 2019 *AJHG*

Heritability (H^2)

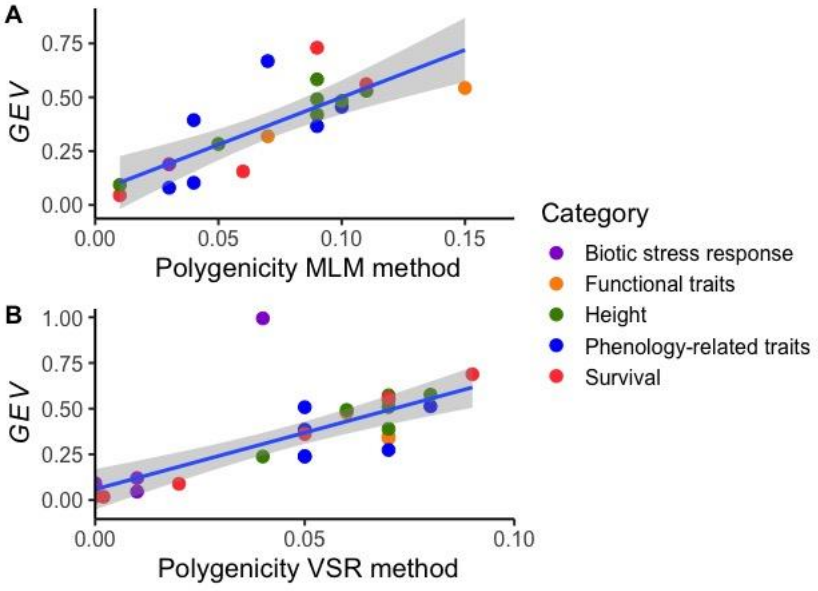


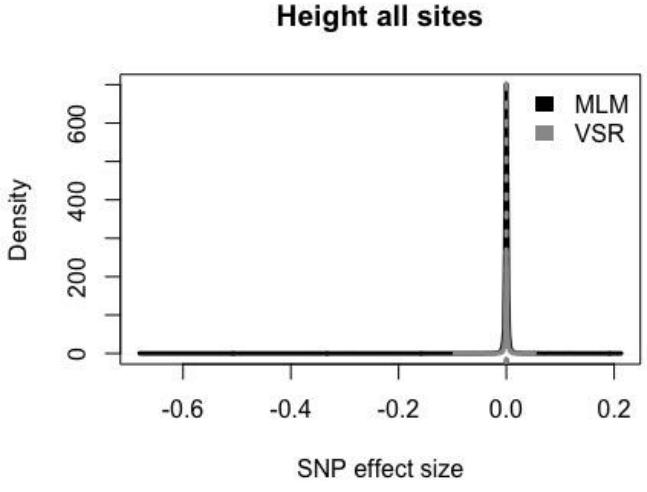
Polygenicity: Proportion of non-zero SNPs



- Average polygenicity 6% of genotyped SNPs (0-27%)
- Highly stable across traits, years and environments

Genetic explained variance (based on BLUPs)



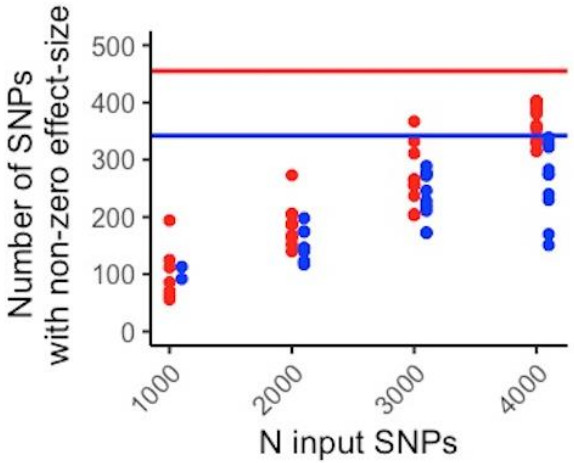


The higher the polygenicity the more genetic variability explained

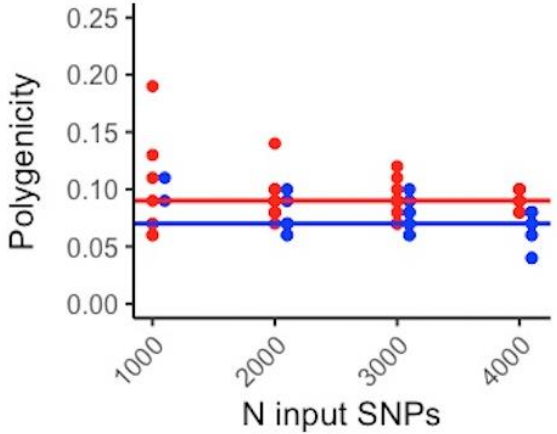
Small-effect SNPs are contributing to polygenicity

Consistency of the SNP dataset

a)

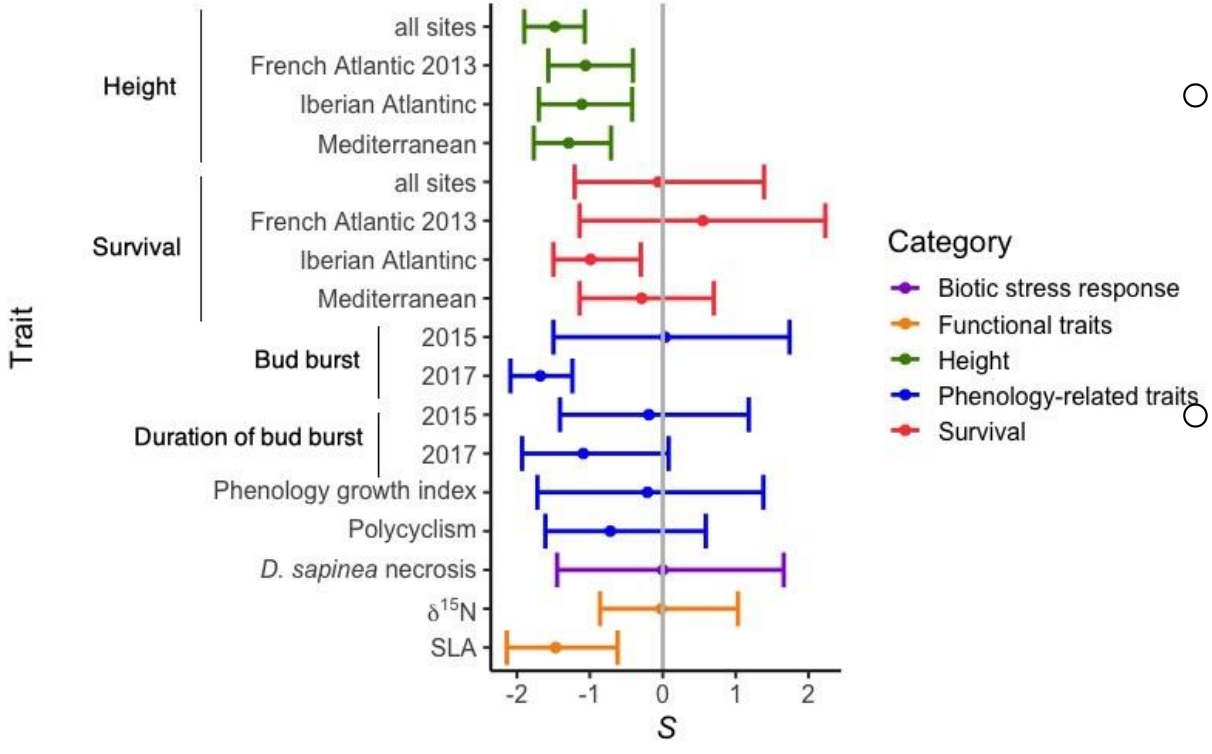


b)



- Consistent estimates of polygenicity even for smaller SNP datasets
- Precision of the estimated polygenicity increased with SNP dataset size

Some evidence of negative selection



- Signatures of negative selection did not mimic the stability observed for polygenicity

- Negative selection has been pointed as a driver of polygenicity (O'Connor et al. 2019)

Conclusions

- Most complex adaptive traits in a long-lived plant species were highly polygenic (average 6% of genotyped SNPs had non-zero effect-size)
- The degree of polygenicity was similar for traits spanning different functional categories, and this genetic architecture was considerably stable over time and environments
- We detected signatures of negative selection on SNPs related to height, survival, phenology-related traits and functional traits, but signatures of negative selection were not consistent across environments and years

Considerations

- Polygenic adaptation can take place rapidly because it only requires small allele frequency changes on standing genetic variation

- Current prediction models are usually based on outlier SNPs of large-effect

- Models for predicting population trajectories in forest trees under climate change could be underestimating the capacity of natural forest tree populations to adapt to new environments

Perspectives

- Estimate polygenicity in several forest tree species with different life-history traits, distributions and genome sizes (B4EST multispecies study – to be discussed tomorrow afternoon)

- Study polygenic adaptation using new models with an improved correction for population genetic structure (Josephs et al. 2019 *Genetics*)

- Study polygenic adaptation in unstructured populations (two case studies available in maritime pine)

Practical demo

GCTB

A tool for Genome-wide Complex Trait Bayesian analysis

Zeng et al. 2018 *Nature Genetics*

(20 min)