













Single genes vs. polygenic adaptation



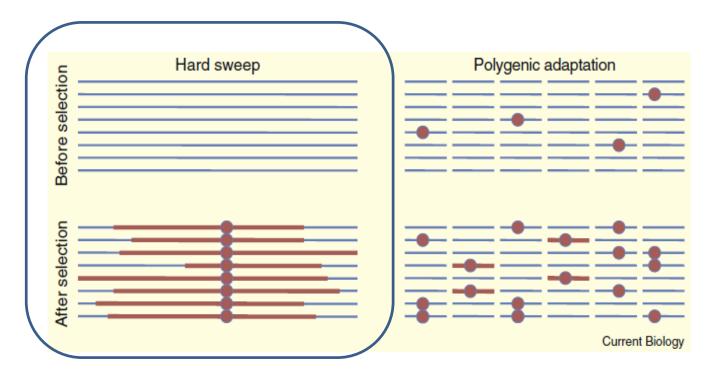
A Kremer^{1,2} and V Le Corre³

underlying genes in response to divergent selection

UNITED

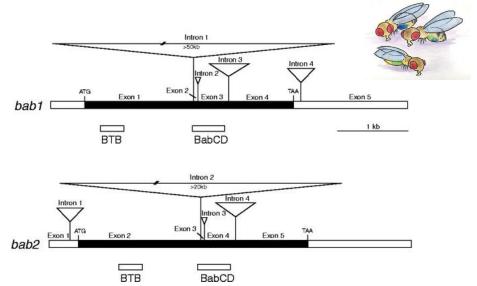
Polygenic traits controlled by small effect SNPs

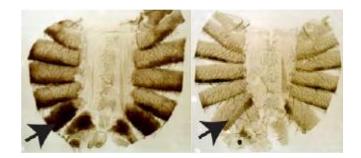
Most methods are designed to detect hard sweeps



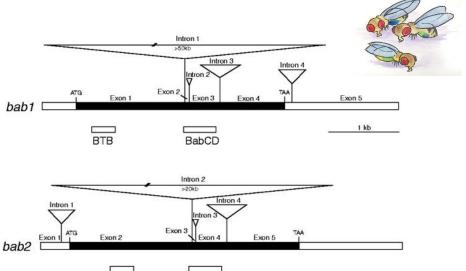
Pritchard et al. (2010) Current Biology

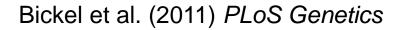
Case study from Remington 2015 Evolution

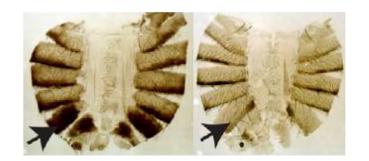




The *bab* locus is responsible for a major effect QTL involving 60% of the variation in female abdominal pigmentation in *D. melanogaster*





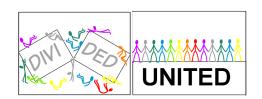


The *bab* locus is responsible for a major effect QTL involving 60% of the variation in female abdominal pigmentation in *D. melanogaster*, but single SNPs explain each <1% of variation, making them basically undetectable by standard GWAS

Table 1. Estimation of polymorphism and region effect size.

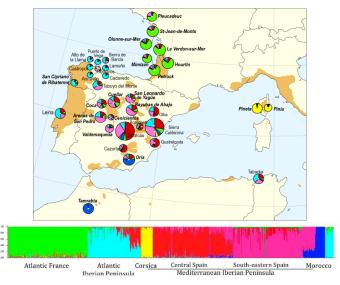
| | Number of sites | Average effect size of each polymorphism | Proportion of total variation explained by average polymorphism | Total estimated effect of region | Proportion of total variation explained |
|---------------|-----------------|--|---|----------------------------------|---|
| Region 1 | 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



Maritime pine (Pinus pinaster Aiton)

Strong population genetic structure with six large gene pools and complex patterns of gene flow and local adaptation

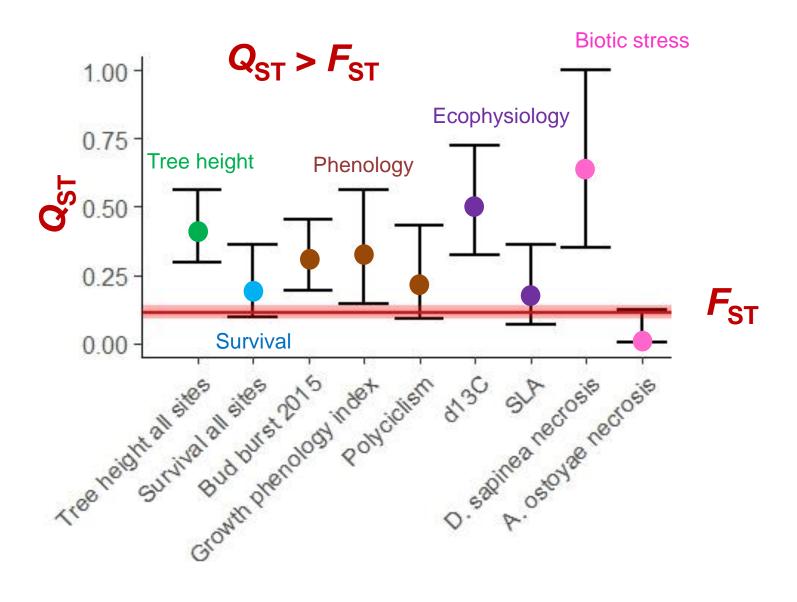


Jaramillo-Correa et al. (2015) Genetics

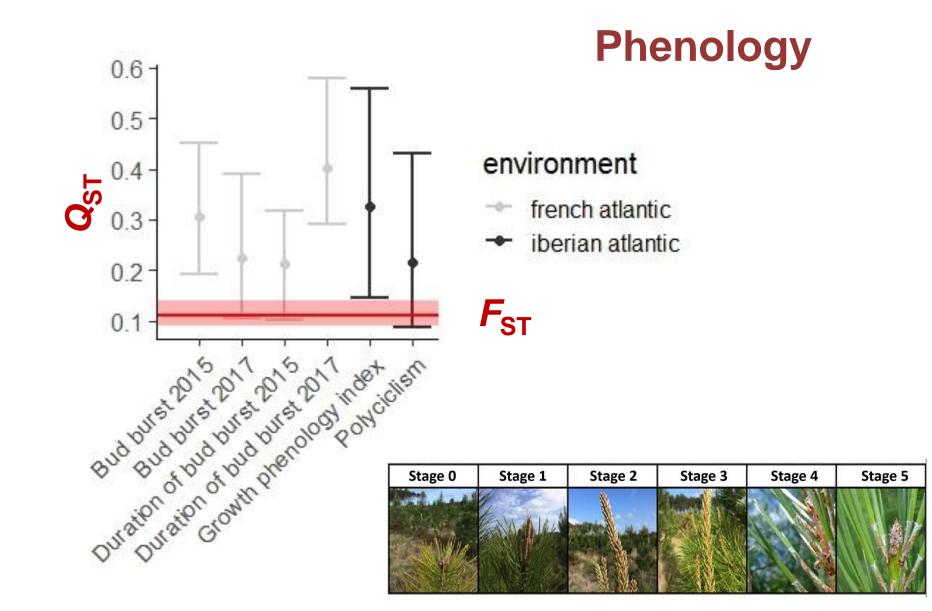
CLONAPIN trial network

- 26,000 trees phenotyped (650 clones) in 5 common gardens in contrasted environmental conditions
- 6,100 SNPs (*Infinium assay*)
- Several phenotypic traits measured, including height, survival, phenology and response to pathogens and pests

Evidence for local adaptation from CLONAPIN common gardens



But as always in biology, it is not simple...



How to look to polygenic adaptation?

The Annals of Applied Statistics 2011, Vol. 5, No. 3, 1780–1815 DOI: 10.1214/11-AOAS455 © Institute of Mathematical Statistics, 2011

Bayesian Variable Selection Regression (BVSR)

BAYESIAN VARIABLE SELECTION REGRESSION FOR GENOME-WIDE ASSOCIATION STUDIES AND OTHER LARGE-SCALE PROBLEMS

By Yongtao Guan and Matthew Stephens¹

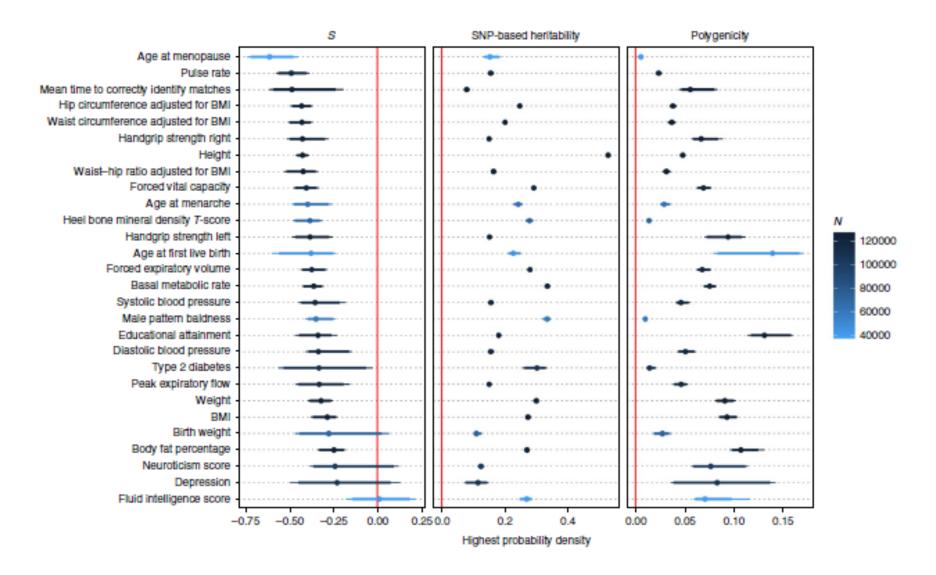


Signatures of negative selection in the genetic architecture of human complex traits

Jian Zeng¹, Ronald de Vlaming ^{2,3}, Yang Wu¹, Matthew R. Robinson¹, Luke R. Lloyd-Jones¹, Loic Yengo¹, Chloe X. Yap ¹, Angli Xue¹, Julia Sidorenko¹, Allan F. McRae¹, Joseph E. Powell¹, Grant W. Montgomery¹, Andres Metspalu⁵, Tonu Esko⁵, Greg Gibson⁶, Naomi R. Wray ^{1,7}, Peter M. Visscher ^{1,7} and Jian Yang ^{1,7}*

Bayesian mixed linear model (BLM), including also the relationship between SNP effect size and minor allele frequency (S)

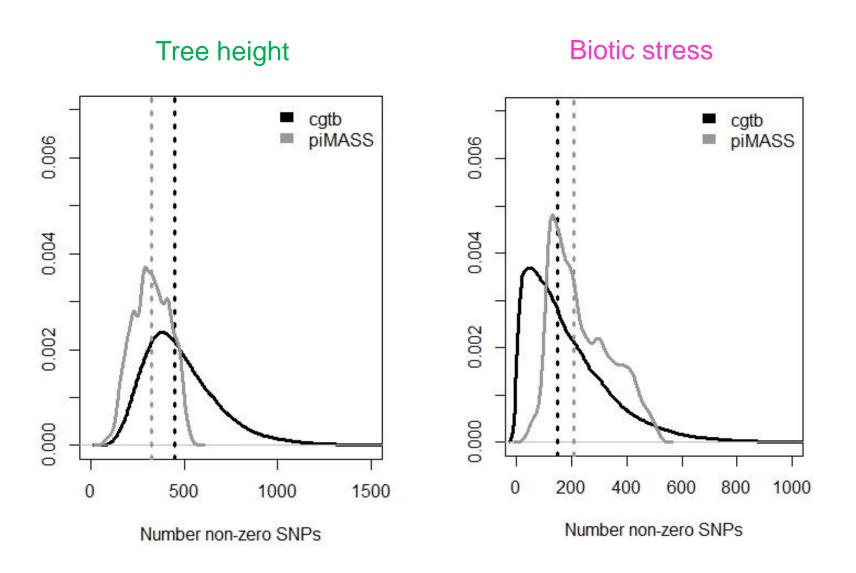
How does it look in humans?



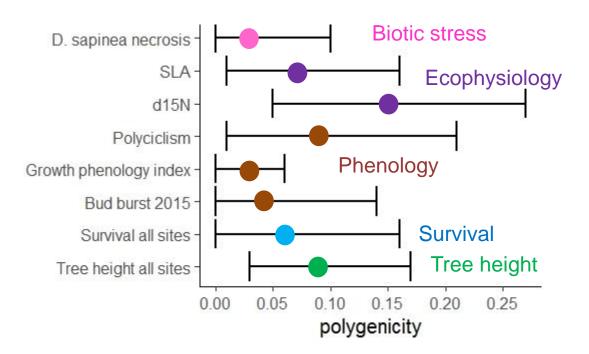
Zeng et al. (2018) Nature genetics

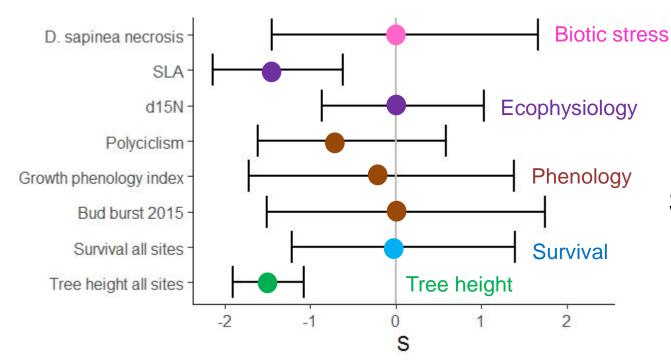
And in maritime pine?

BVSR and BLM methods agree for most traits!

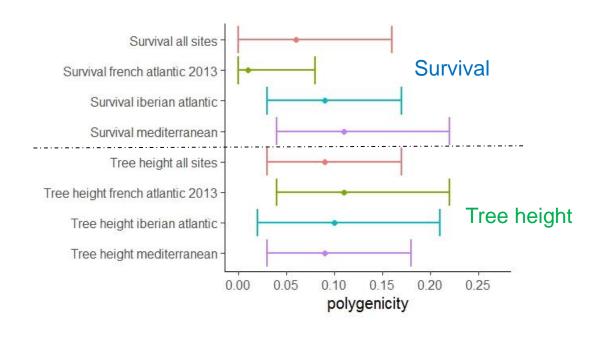


Polygenicity values around 5-10%



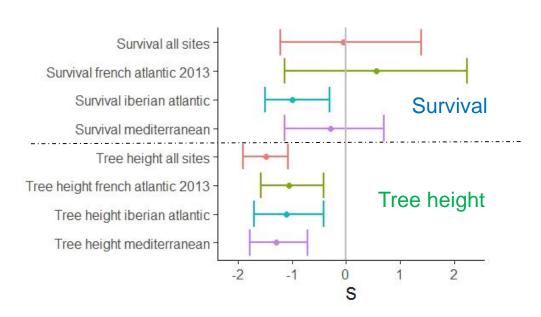


Negative selection is important for SLA and tree height



environment

- all sites
- french atlantic
- iberian atlantic
- mediterranean



Polygenicity and negative selection are variable across traits, but remarkably not across environments

Conclusions and perspectives

- Levels of poligenicity are high, with over 5-10% of SNPs having non-zero effects, and vary across traits, but not across environments (indicating similar genetic architecture)
- New methods to study polygenic adaptation at gene and pathway level can potentially increase our understanding on how trees adapt to a changing environment (but we need more data for GWAS!)
- Negative selection is relevant for some main adaptive traits

What's next?

Large common garden for Corsican populations of maritime pine (900 families from 30 populations) that has been genotyped for over 100k SNPs using sequence capture methods

Tree height, survival, phenology and carbon discrimination data already available



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www.evoltree.org

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