Adaptation to genome duplication in autotetraploid *Arabidopsis arenosa*
The age of genomes

Curiouser and curiouser!

It’s a great time for evolutionary genetics!
• Genetics of adaptation to habitat
• Adaptation to genome change
• Evolution of gene interaction networks
Arabidopsis arenosa – an outcrossing relative of A. thaliana
Arabidopsis arenosa – part of a user-friendly genus

**Arabidopsis arenosa**
Sequenced
(Illumina by us; JGI in progress)

**Arabidopsis halleri**

**Arabidopsis lyrata**
Sequenced

**Arabidopsis thaliana**
Sequenced

5-15 MYA

0.5-2 MYA

?
I. Genetics of adaptation to whole genome duplication

II. Selection on gene networks in A. arenosa?
Historic genome duplication events in eukaryotes

Polyploidy events are thought to have contributed to speciation and plant and animal diversity.
Whole genome duplication in animals

Polyploidy is also a normal part of development

But cancer cells are also often polyploid
Whole genome duplication – a common perturbation in plants
e.g. many crop species are polyploid

Many wild species are polyploid as well
There are two major kinds of polyploids:

- **Autopolyploid**
  - "within species"

- **Allopolyploid**
  - "hybrid"
Formation

Early

Extensive genome change
Epigenetic changes
Instability
Aneuploidy

Established

Increased meiotic stability
Cytologically diploid
Evolutionary potential

Stabilization

There has not been any study (prior to ours) of genetic adaptation to genome duplication.
Arabidopsis arenosa tetraploids are widely distributed

Ploidy tests in our material: Elizabeth Svedin, Brian Dilkes, Purdue Univ.

Jesse Hollister
Brian Arnold
Katherine Xue
April Dobbs
Meiosis can be challenging for neo-autotetraploids

?? Can lead to Aneuploidy, breakage, etc..
Established autotetraploids rarely form multivalents

New or synthetic polyploids

Established polyploids

Bivalents can be ensured by:

1. Pairing partner preferences
2. Limiting crossovers
What about *A. arenosa*?

Consistent with findings from Carvalho et al. 2010 *Sex Plant Rep*.

We have population genetic evidence that *pairing partner choice* remains random.
A. arenosa Population Genetics & Genomics

Illumina short read sequences for:
12 tetraploids (future plan – also Carpathian tetraploids)
Newly also 8 Carpathian diploids (Levi Yant (Kramer lab), Ben Hunter)

Hollister et al. PLoS Genetics, 2012
A “genome scanning” approach

ALL (48 out of 48) *arenosa* seq differ from *lyrata*

Look for regions with:
Excess derived SNPs
Low polymorphism

1 out of 48 *arenosa* seq diff from *lyrata*
Looking for sweep signatures - Two lists

1. Composite likelihood ratio for unusual SFS (top 1%) (Nielsen et al., Genome Res 2005)

2. Top 5% of SFS test AND bottom 5% of diversity within A. arenosa

Over-represented functional categories in “candidate gene lists”

Tests for selection in tetraploids vs *A. lyrata* (347 genes total)

- Chromosome organization and structure
- Chromosome cohesion and segregation (meiosis)
- DNA repair and Homologous recombination
- Regulation of basal transcription
- Intracellular transport
- Cell division, growth and morphogenesis
- Metabolism
- Development and flowering time
- Epigenetic regulation

Top 0.5% differentiated by ploidy within *A. arenosa* (100 genes total)

- Chiasma assembly
- M phase (cell cycle)

Parallels in other kingdoms


• Examined >3000 mutants in diploid and tetraploid yeast
• Identified 39 tetraploid-specific lethals
• Implicate genome stability –
  —homologous recombination, sister chromatid cohesion, spindle function.

Functional classes and homologous genes, overlap with our data

Genes identified in yeast that we also found in A. arenosa are implicated in human disease:

• Oncogenes / instability of polyploid cancers
• Genome instability syndromes (e.g. Nijmegen Break, Bloom & Cornelia de Lange)
• Other (e.g. Alzheimers and neurodegeneration diseases)
**ASY1** – SNP differentiation from *A. lyrata*

ALL (48 out of 48) *arenosa* seq differ from *lyrata*

1 out of 48 *arenosa* seq diff from *lyrata*

ASY in other plant species

• **ASY1 in Arabidopsis thaliana** (diploid)
  – essential for homologous chromosome synapsis.

• **PAIR2 in rice** (diploid)
  – essential for homologous chromosome synapsis.

• **TaASY1 in wheat** (polyploid)
  – quantitatively affects pairing of homologous chromosomes in 4N wheat.
ASY1 is differentiated by cytotype in *A. arenosa*

Derived ASY1 may have been selected from diploid standing variation

ZYP1 (also required for SC formation) is also differentiated by cytotype (Levi Yant, Jesse Hollister)

K = Lysine
E = Glutamic Acid

Katherine Xue
Within HORMA domain

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Conclusions:

A. *arenosa* seems to have genetically adapted to its polyploid state

Chromosome synapsis, recombination and segregation show evidence of selection

Adjustment of metabolism, cell cycle, cell growth likely also important

Selection seems to have acted on connected members of gene networks

Co-evolution? Selection operating via small independent changes in functionally related genes?

One of our sites:
The “Zinnen” (Battlements)
Upper Danube Valley, Germany
Also many thanks to:
Peg Richards (Awesome admin!)
Levi Yant (Kramer lab, Harvard)
Brian Dilkes & Elisabeth Svedin (Purdue Univ)
Luca Comai (UC Davis)
Nancy Kleckner (Harvard)
John Wakeley (Harvard)
Chris Franklin (Univ Birmingham)

Also: Kristin Tsuo, Yanniv Dorone, Grace Daher
Polyploids persist
But require more changes
Compensatory

-Jesse Hollister