Importance of polyploidy, inheritance patterns and manipulation.

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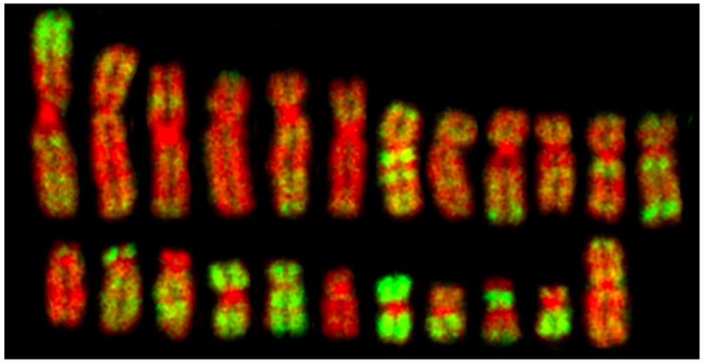
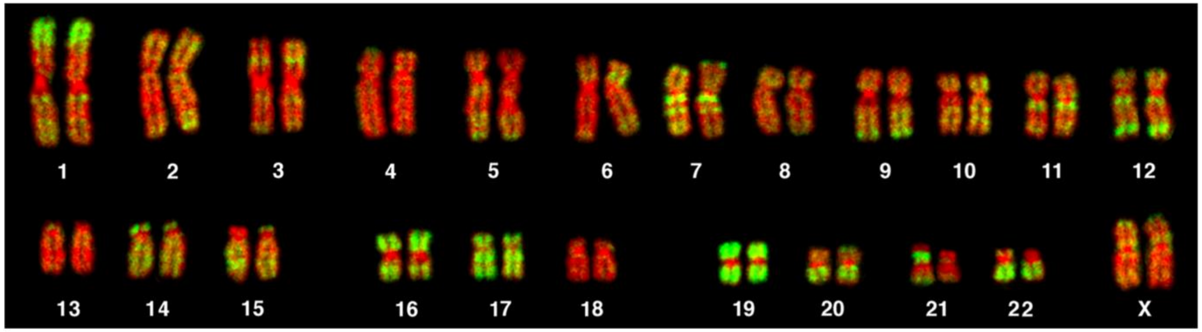
March 25, 2021 - HS 703 - Raleigh, North Carolina

Ploidy level

- **Basic chromosome number**: the number of different chromosomes that make up a single complete set. Humans, x = 23

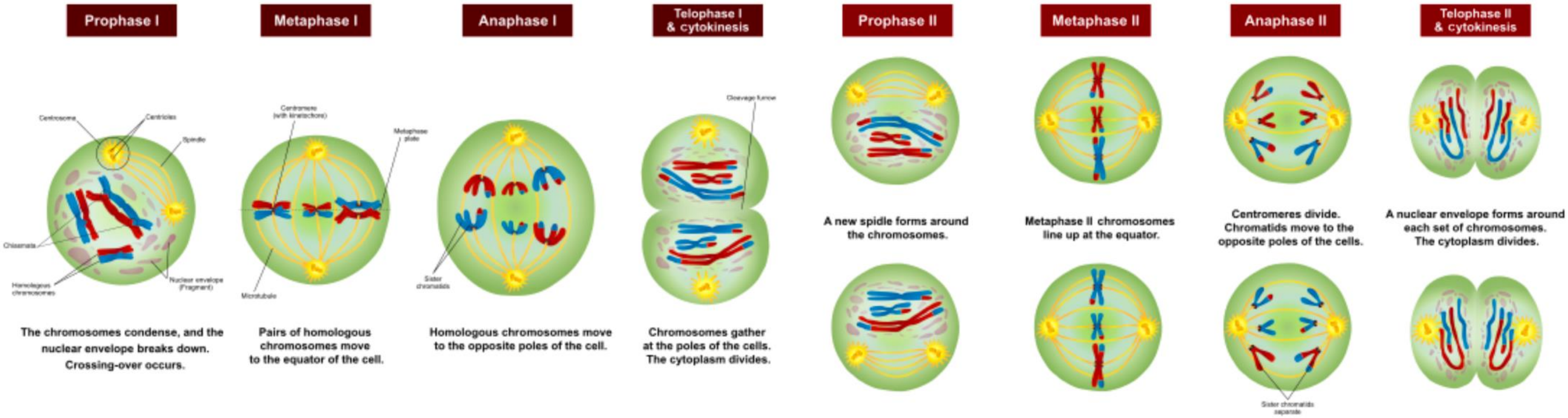


Ploidy level: Number of basic chromosome sets. Humans p = 2



Meiotic pairing and gamete formation in diploids

- Meiotic process is quite stable
- It resolves in four products with N = x = basic number



Polyploid: definition and characteristics

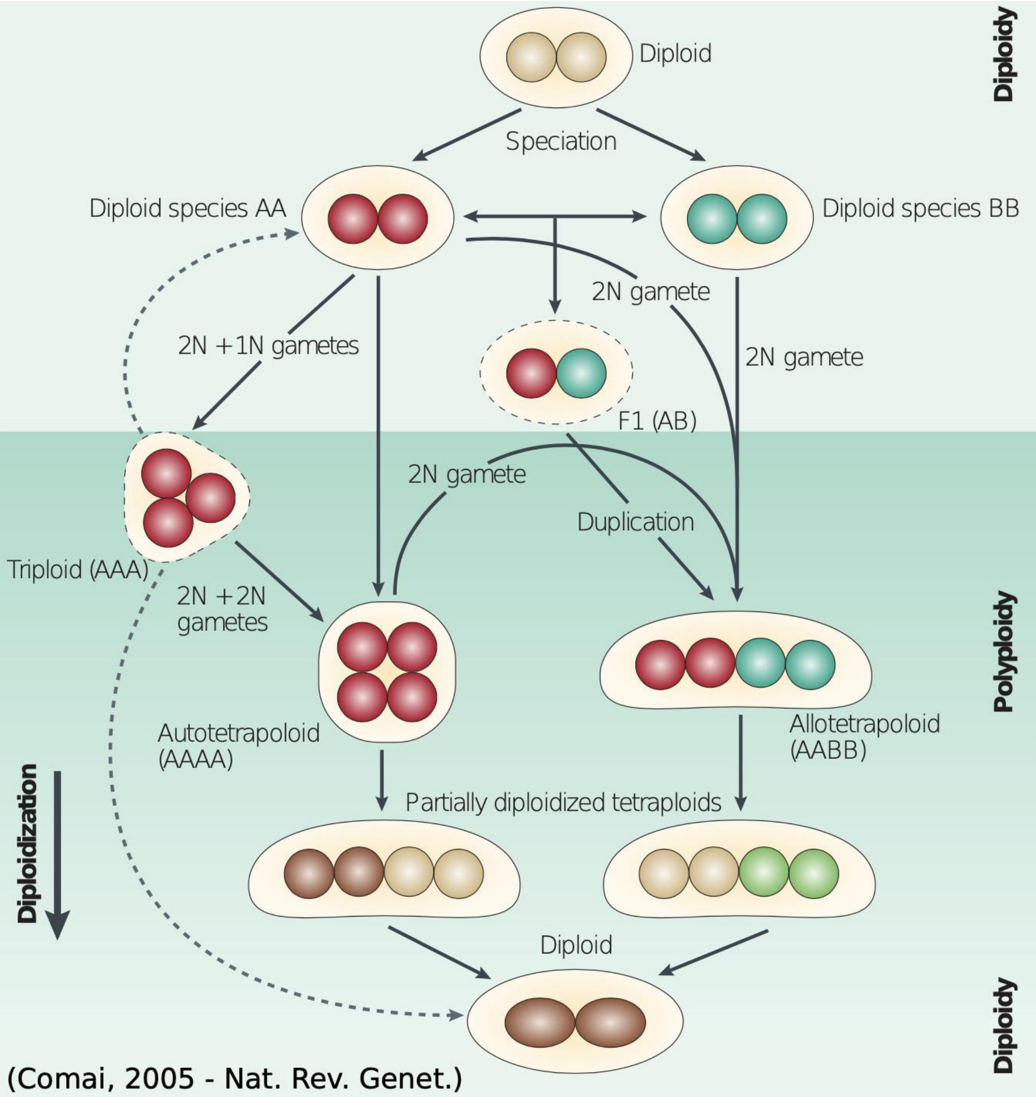
- Organisms with more than two complete sets of chromosomes per cell nucleus

- Incidence: estimations range from 30 to 35 % (Stebbins 1971) to 70 %(Masterson 1994) for flowering plants.

- Origin: somatic doubling and unreduced gametes.

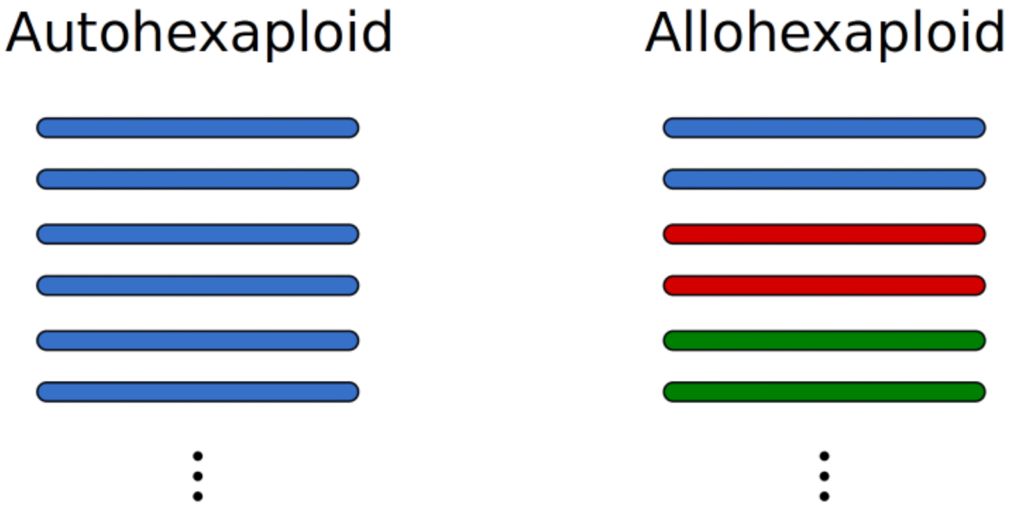
- Two process to form unreduced gametes (production of 2N gametes):
  - *first division restitution* (FDR): errors during meiosis I → same levels of heterozygosity as the parents
  - *second division restitution* (SDR): sister chromatids do not segregate in anaphase II → lower levels of heterozygosity when compared to the parents

How are polyploids formed?



(Comai, 2005 - Nat. Rev. Genet.)

Polyploid species: Allo vs. Autopolyploids



- Multiple sets have the same origin
- Multiple sets have different origins

- Homologous pairing and inheritance pattern play an important role in this definition.
- Segmental allopolyploids: concept proposed by Stebbins in 1950. Occurs when a polyploid is originated from related species leading to intermediate modes of inheritance.

Polyploidy consequences

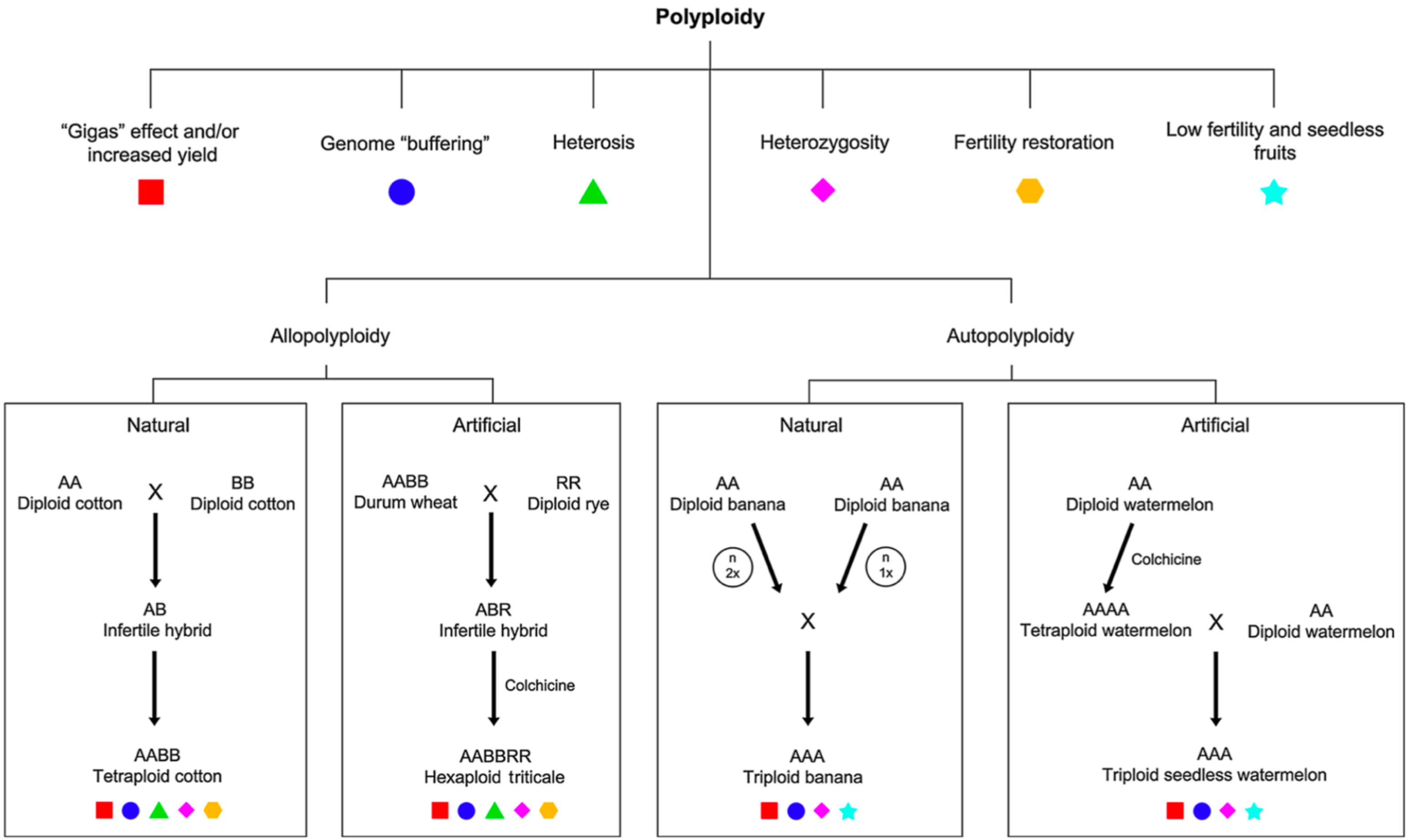
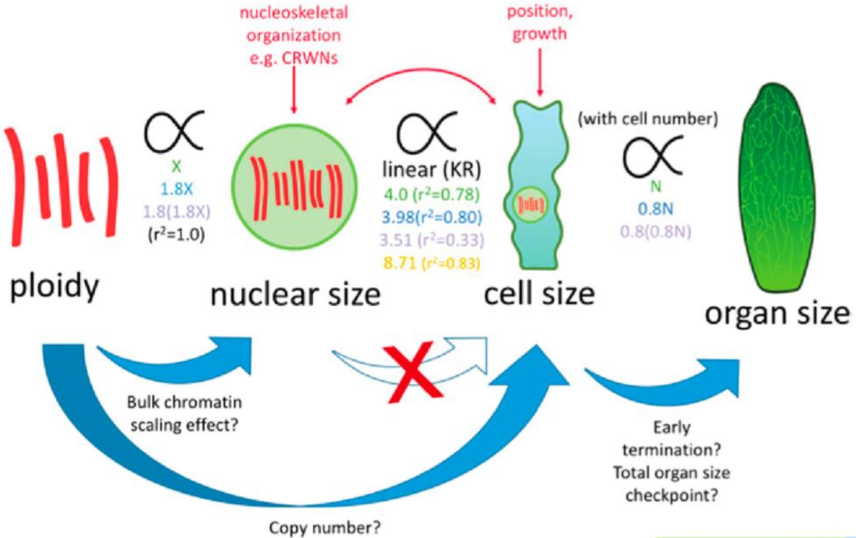
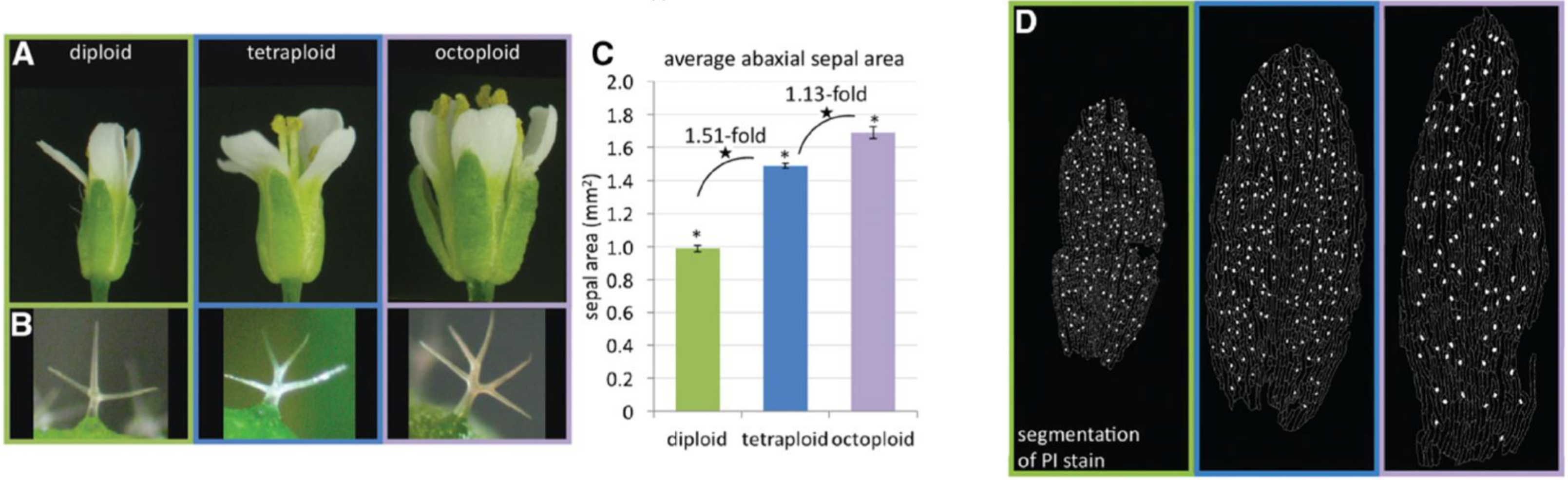


Fig. 1 Schematic representation of four cultivated species and some of the main polyploidy consequences for application in crop improvement. The *symbols* "n 1*x*" and "n 2*x*" refer to reduced and unreduced reproductive cells, respectively

(Sattler et al. 2015)

Ploidy and Size

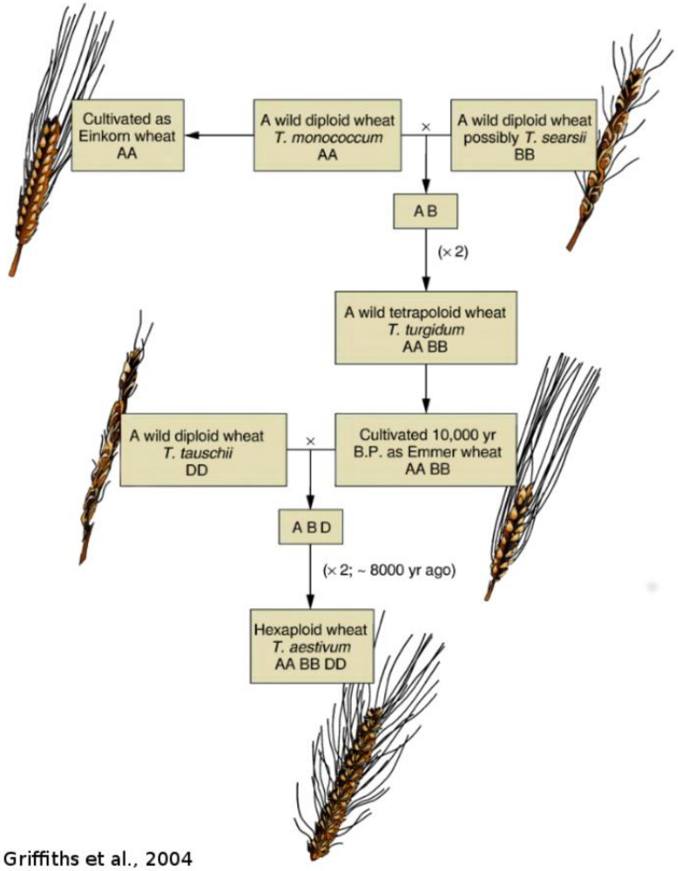




Robinson *et al.* (2018) - Plant Cell - `https://doi.org/10.1105/tpc.18.00344`

Allopolyploid example

Wheat - *Tritucum aestivum*



Cultivated as
Einkorn wheat
AA

A wild diploid wheat
T. monococcum
AA

×

A wild diploid wheat
possibly *T. searsii*
BB

AB

(× 2)

A wild tetrapoloid wheat
T. turgidum
AA BB

A wild diploid wheat
T. tauschii
DD

×

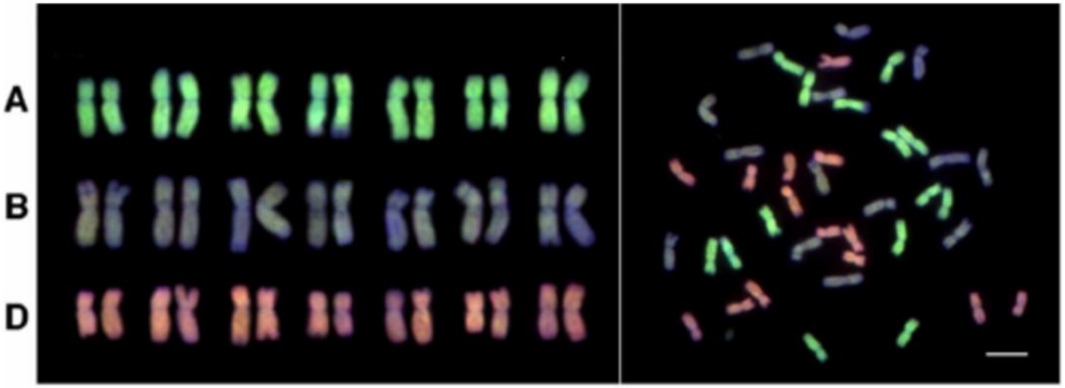
Cultivated 10,000 yr
B.P. as Emmer wheat
AA BB

ABD

(× 2; ~ 8000 yr ago)

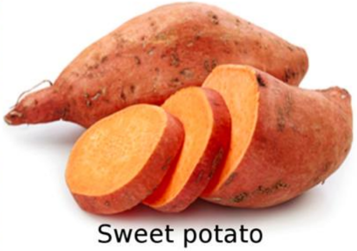
Hexaploid wheat
T. aestivum
AA BB DD

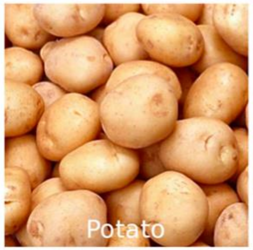
Griffiths et al., 2004

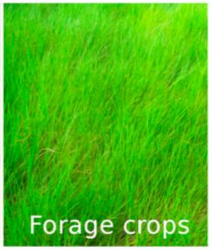


Zhang H et al. PNAS 2013;110:3447-3452

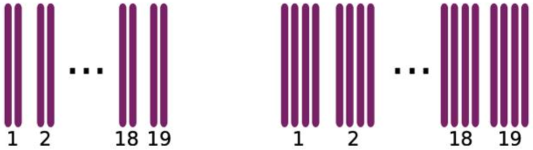
Autopolyploid examples





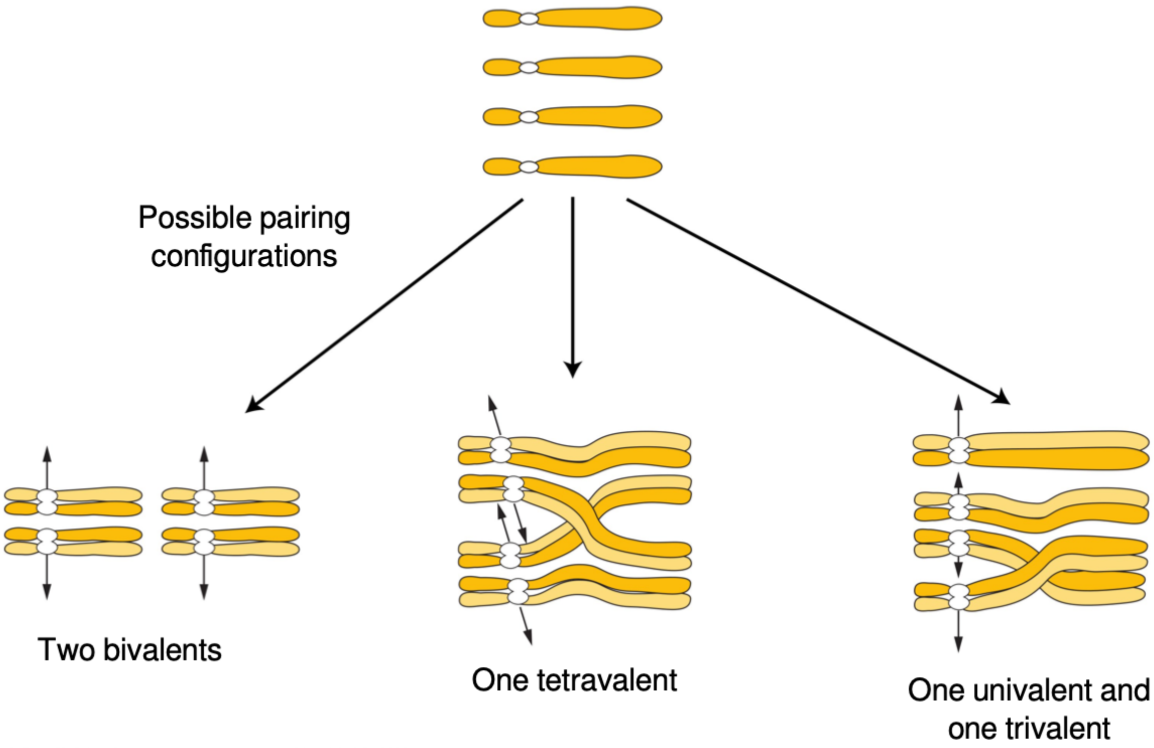


Diploid Autotetraploid



Meiotic pairing in autopolyploids

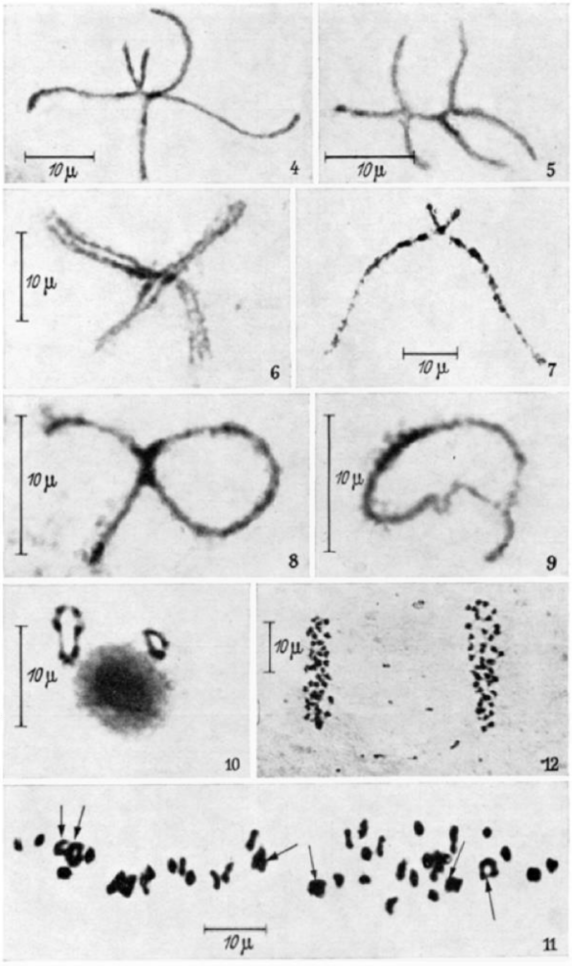
Tetraploid example



Griffiths et al. (2004)

Meiotic Pairing in polyploids

Sweetpotato



Hexavalents, quadrivalents and bivalents in sweetpotato (Magoon *et al.* 1970)

Sugarcane

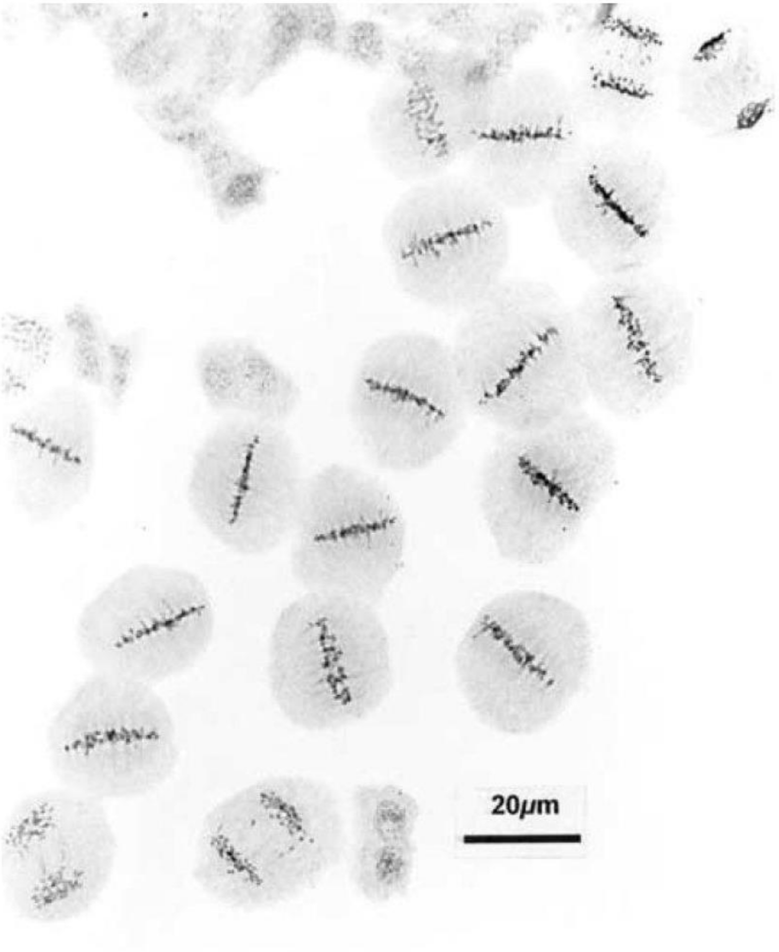
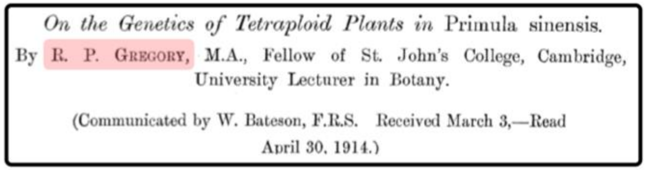


Figure 1. PMCs of *Saccharum* spp. hybrid clone 79N9059 at meiosis. As was the case in other clones, pairing was regular, bivalents generally formed. (Bielig *et al.* 2003)

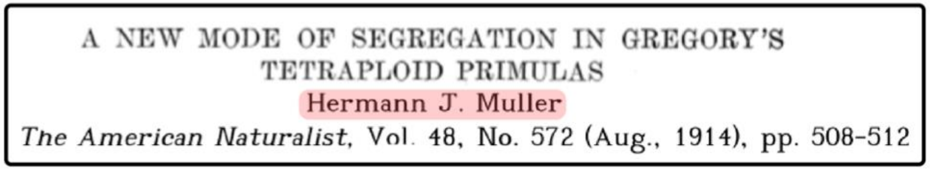
Autopolyploids: some important papers - 1914 to 1954



On the Genetics of Tetraploid Plants in Primula sinensis.

By R. P. GREGORY, M.A., Fellow of St. John's College, Cambridge, University Lecturer in Botany.

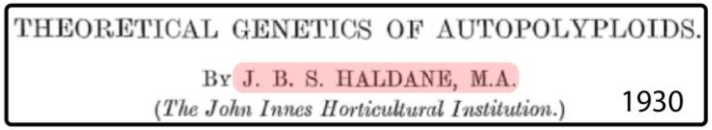
(Communicated by W. Bateson, F.R.S. Received March 3,—Read April 30, 1914.)



A NEW MODE OF SEGREGATION IN GREGORY'S TETRAPLOID PRIMULAS

Hermann J. Muller

The American Naturalist, Vol. 48, No. 572 (Aug., 1914), pp. 508-512

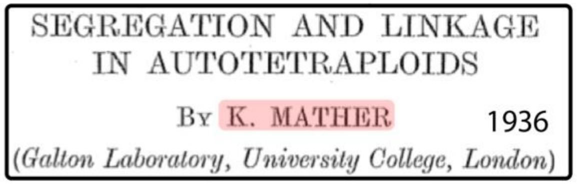


THEORETICAL GENETICS OF AUTOPOLYPLOIDS.

BY J. B. S. HALDANE, M.A.

(*The John Innes Horticultural Institution.*)

1930

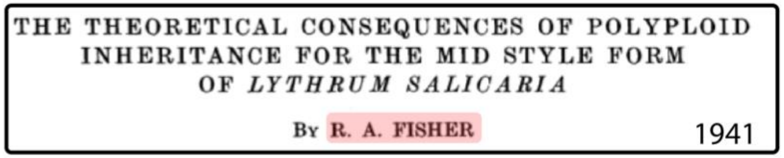


SEGREGATION AND LINKAGE IN AUTOTETRAPLOIDS

BY K. MATHER

(*Galton Laboratory, University College, London*)

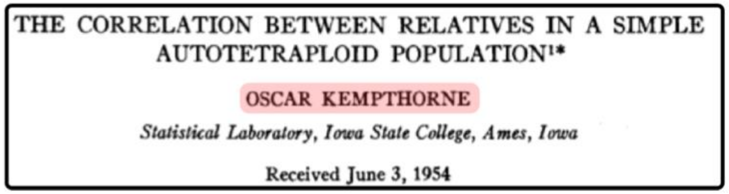
1936



THE THEORETICAL CONSEQUENCES OF POLYPLOID INHERITANCE FOR THE MID STYLE FORM OF *LYTHRUM SALICARIA*

BY R. A. FISHER

1941



THE CORRELATION BETWEEN RELATIVES IN A SIMPLE AUTOTETRAPLOID POPULATION[1]*

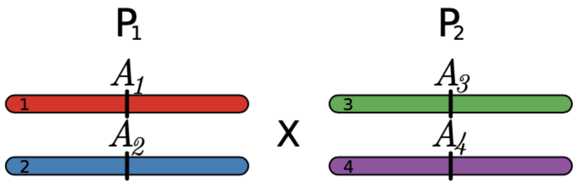
OSCAR KEMPTHORNE

Statistical Laboratory, Iowa State College, Ames, Iowa

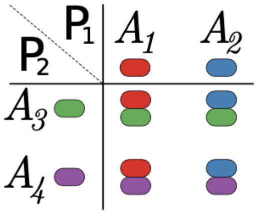
Received June 3, 1954

Assessing allelic variation in diploids

Multiallelic



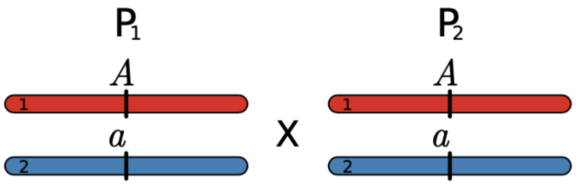
Gametes



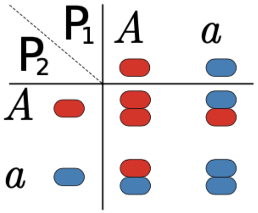
4 possible genotypes

1:1:1:1

Biallelic



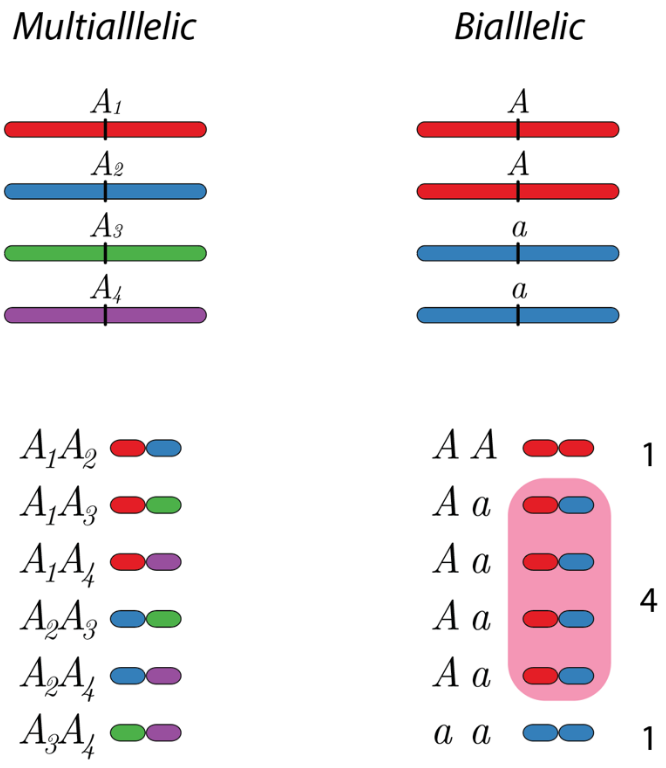
Gametes



3 possible genotypes

1:2:1

Gamete formation and allelic variation in polyploids*

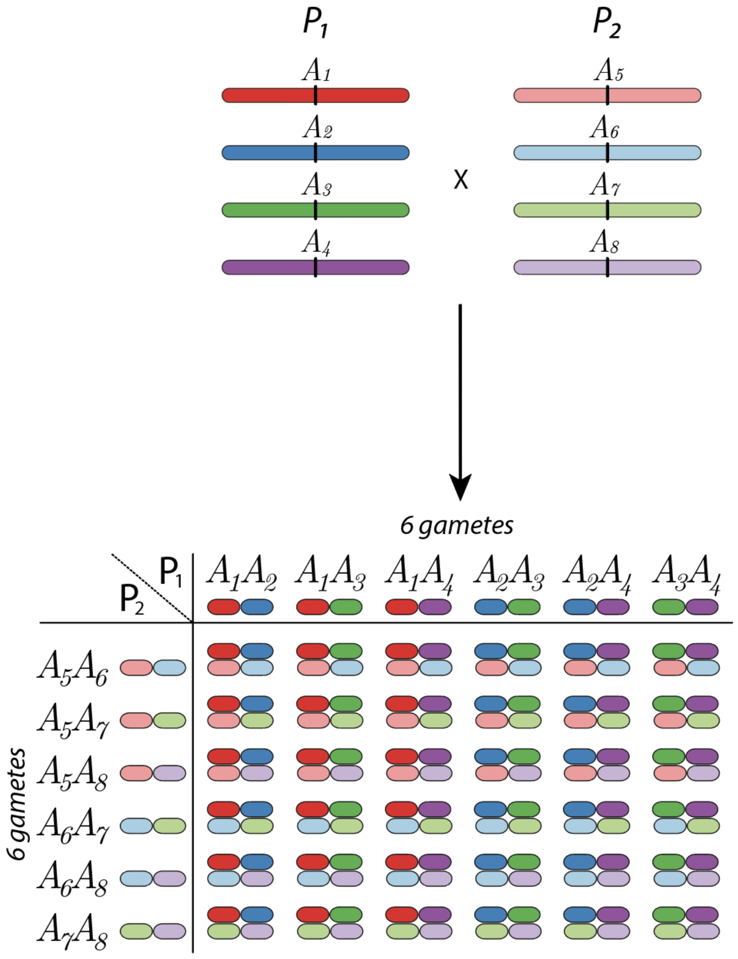


Number of possible gamtes considering one locus with no double-reduction in one and two parents

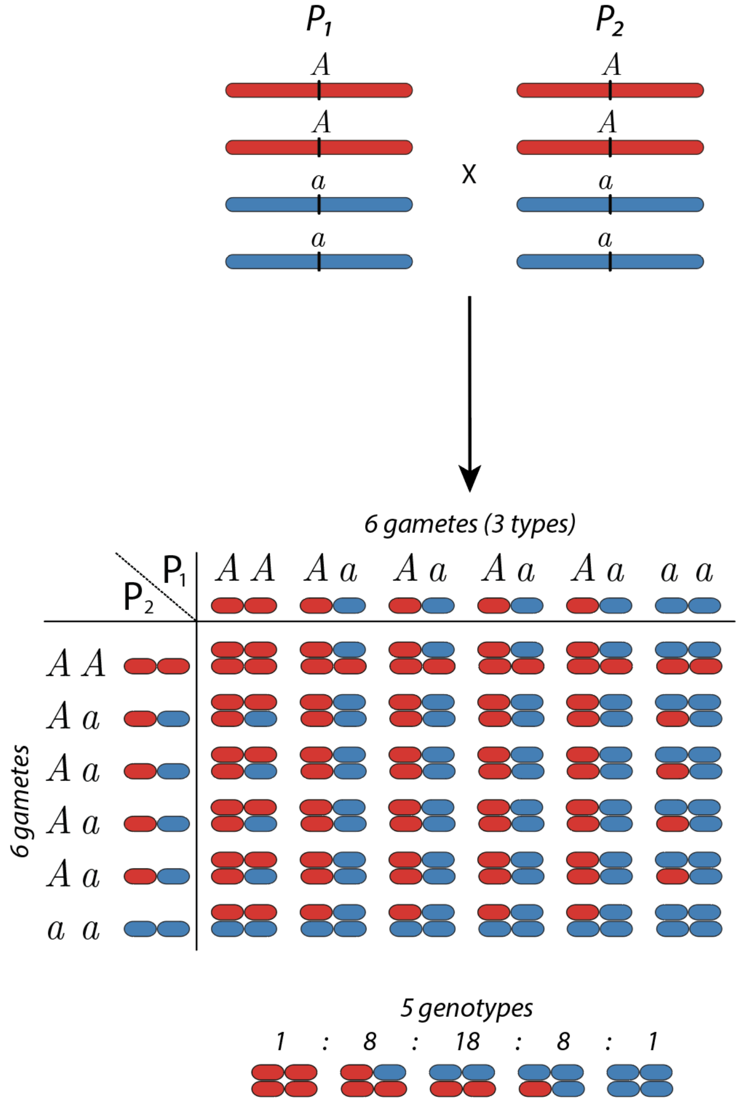
Ploidy	$\binom{p}{\frac{p}{2}}$	$\binom{p}{\frac{p}{2}}^2$
4	6	36
6	20	400
8	70	4900
10	252	63504
12	924	853776
14	3432	11778624
16	12870	165636900

*random pairing and no double reduction

Segregation in polyploids: autotetraploid example*



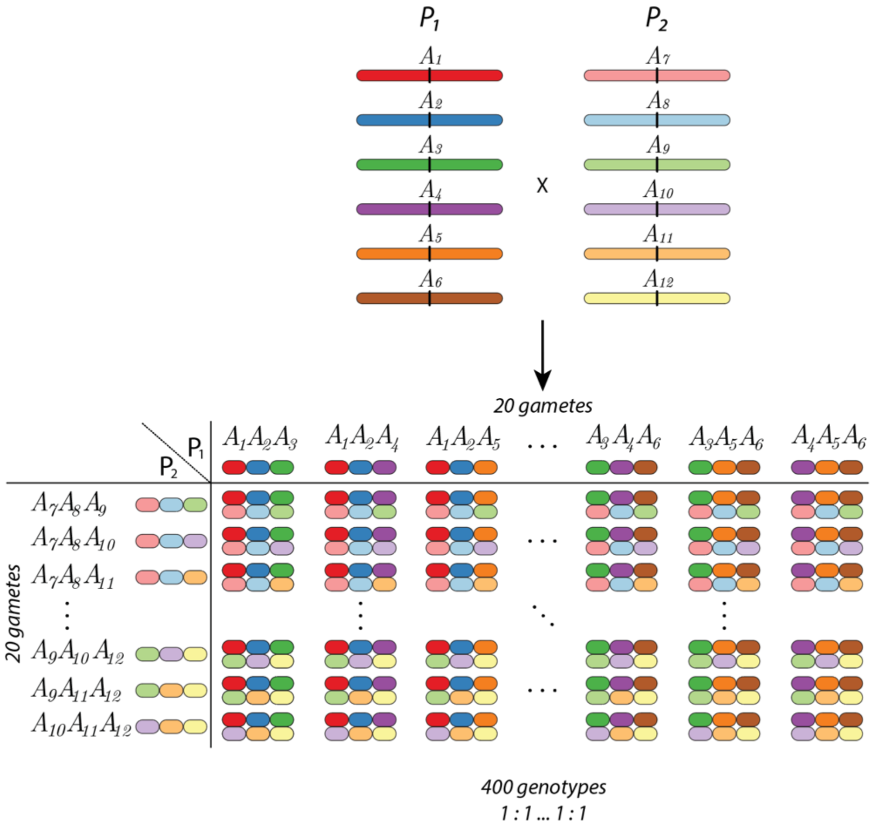
36 genotypes
1 : 1 … 1 : 1

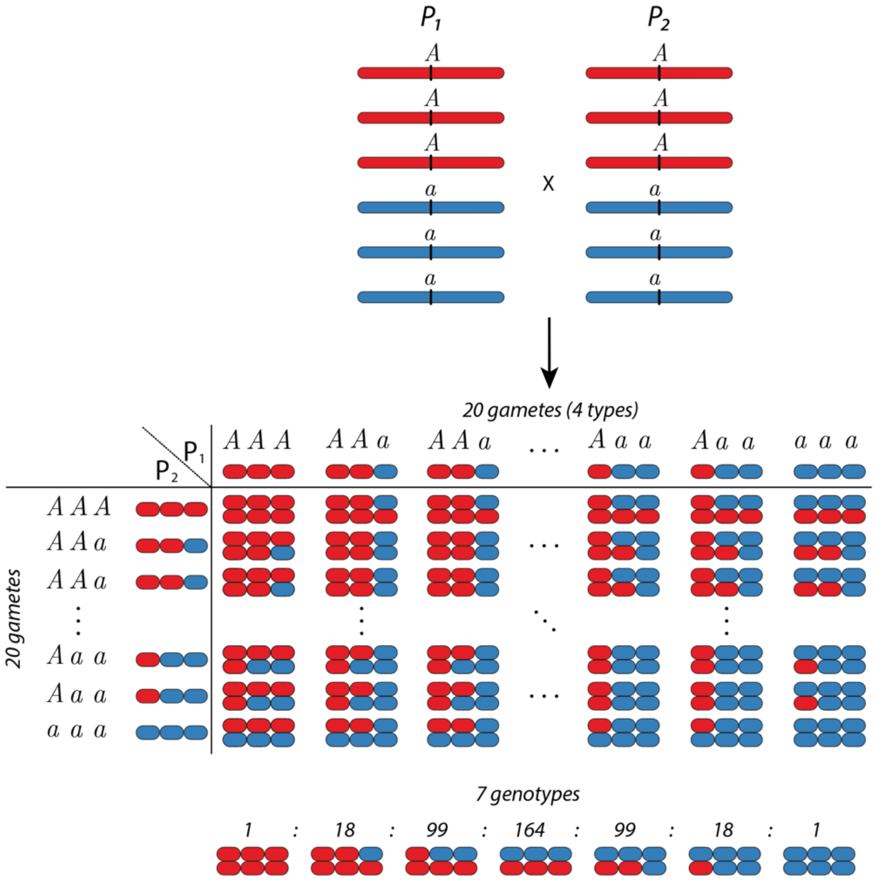


5 genotypes
1 : 8 : 18 : 8 : 1

*random pairing and no double reduction

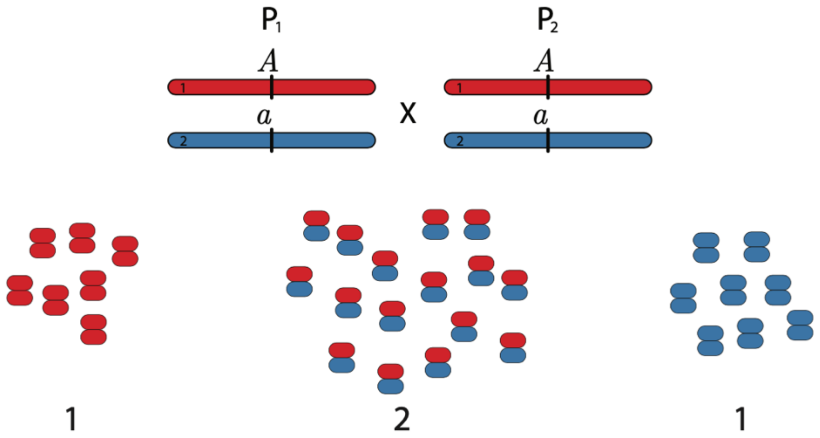
Segregation in polyploids: autohexaploid example*





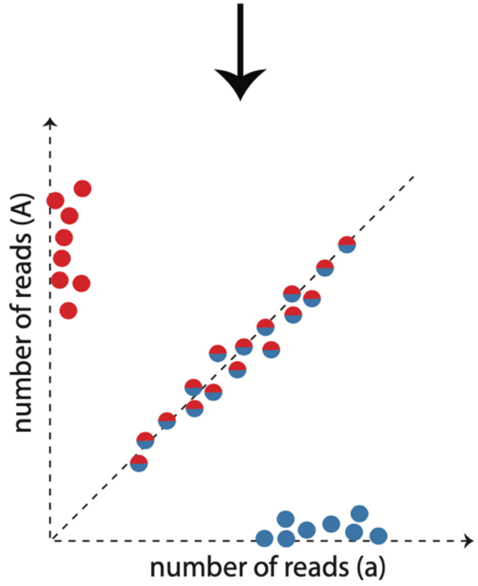
*random pairing and no double reduction

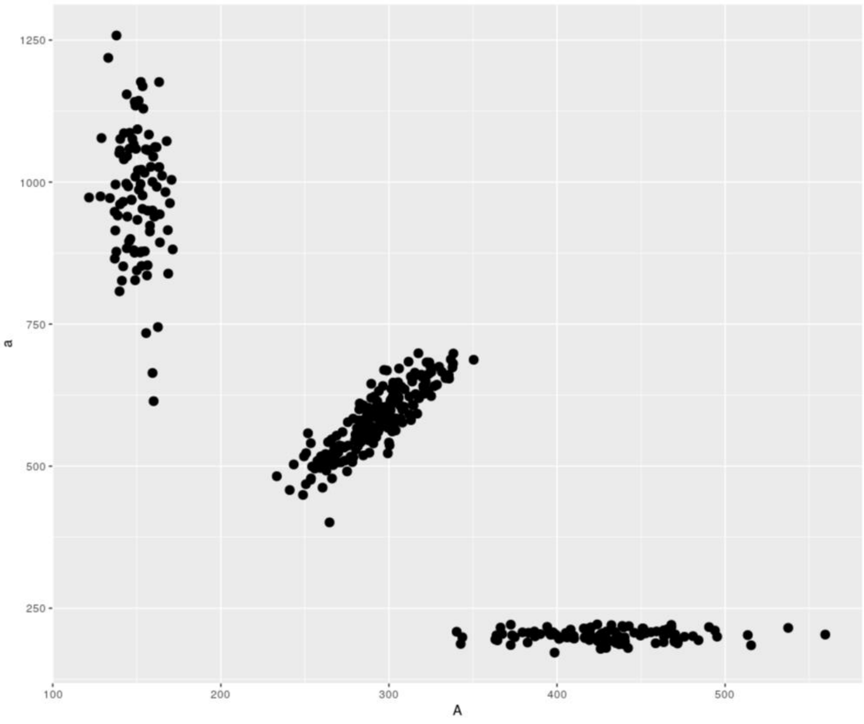
Assessing allelic variation in diploids



GBS

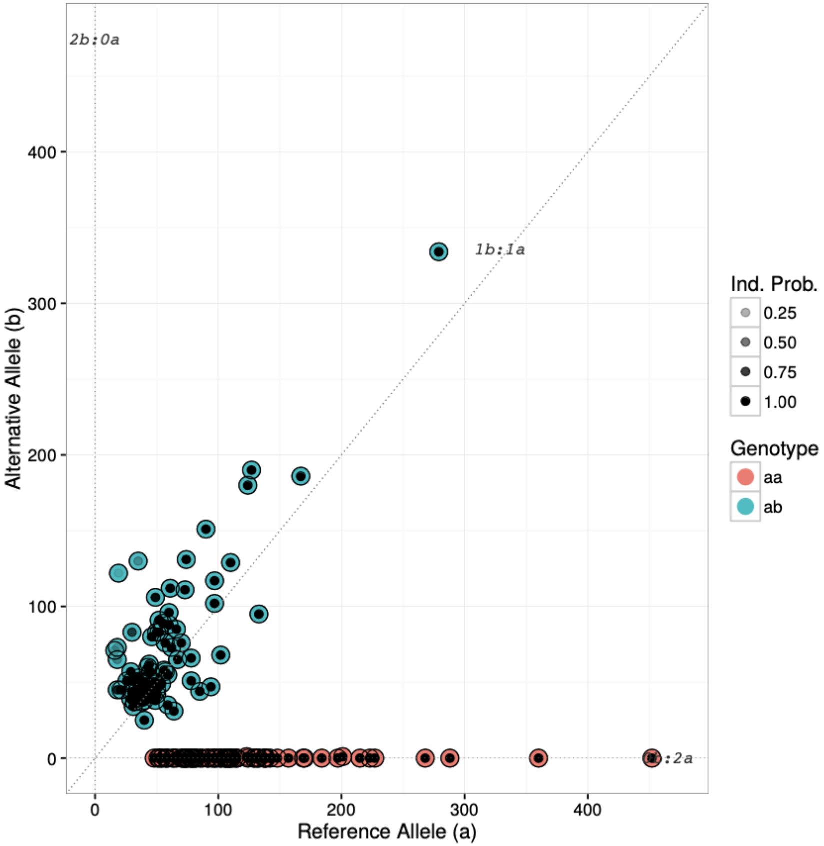
Read counts for each one of the allelic variants



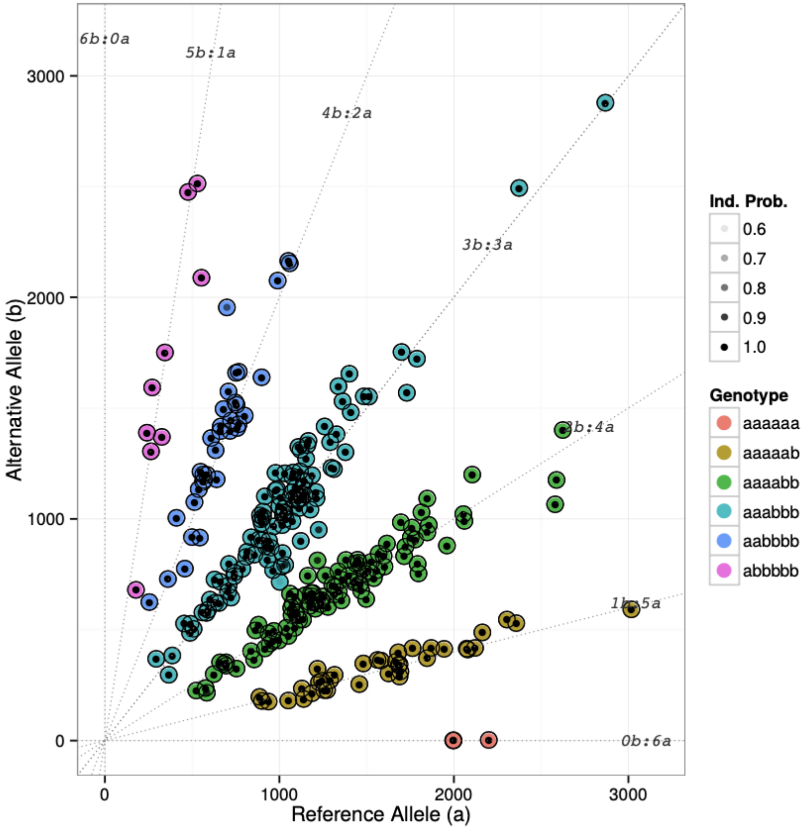


Assessing allelic variation in autopolyploids

Diploid

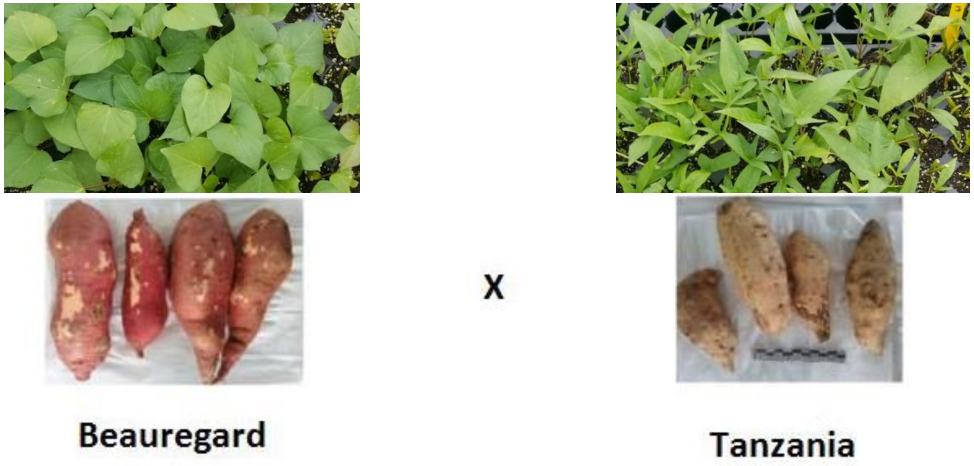


Hexaploid



Biparental Population - BT

- Beauregard x Tanzania
- 315 individuals
- GBS – GBSpoly protocol (Bode Olukolu – U Tennessee)
- Two reference genomes *I. trifida* and *I. triloba* (Zhangjun Fei's group – BTI Cornell)

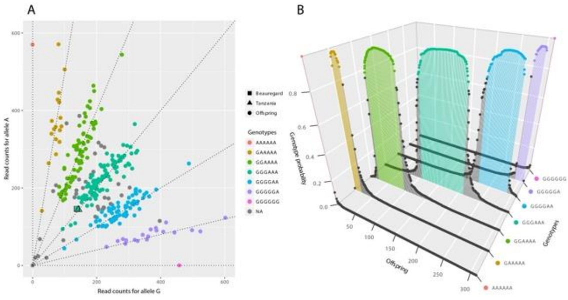


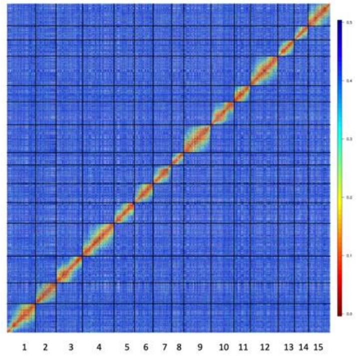

Phenotyping Beauregard x Tanzania Mapping Population

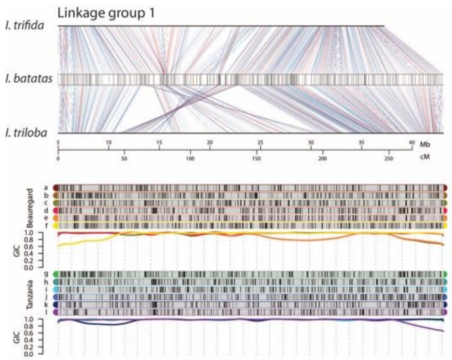
Design: Alpha lattice (80 x 4)
Genotypes: 320
Replications: 3
Plot: 8 plants/row, 2 rows
Site: Namulonge
Planted: 08/03/2017

Linkage analysis in BT population

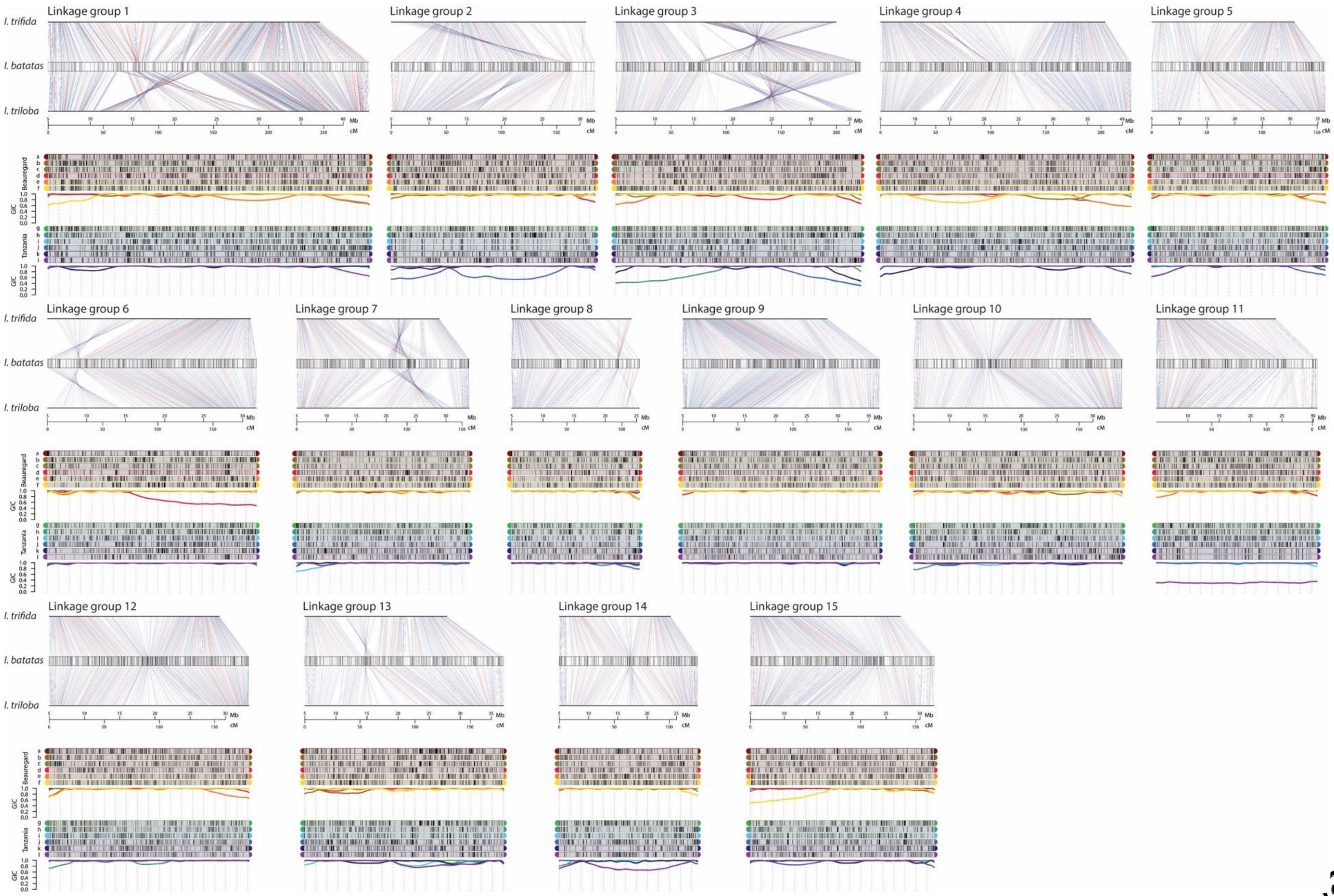
- Genotype calling and filter
- Pairwise estimation of recombination fractions
- Filtering based on pairwise analysis
- Clustering SNPs in linkage groups
- Ordering markers within each linkage group
- Phasing and map re-estimation
- Haplotype analysis
- Meiotic analysis



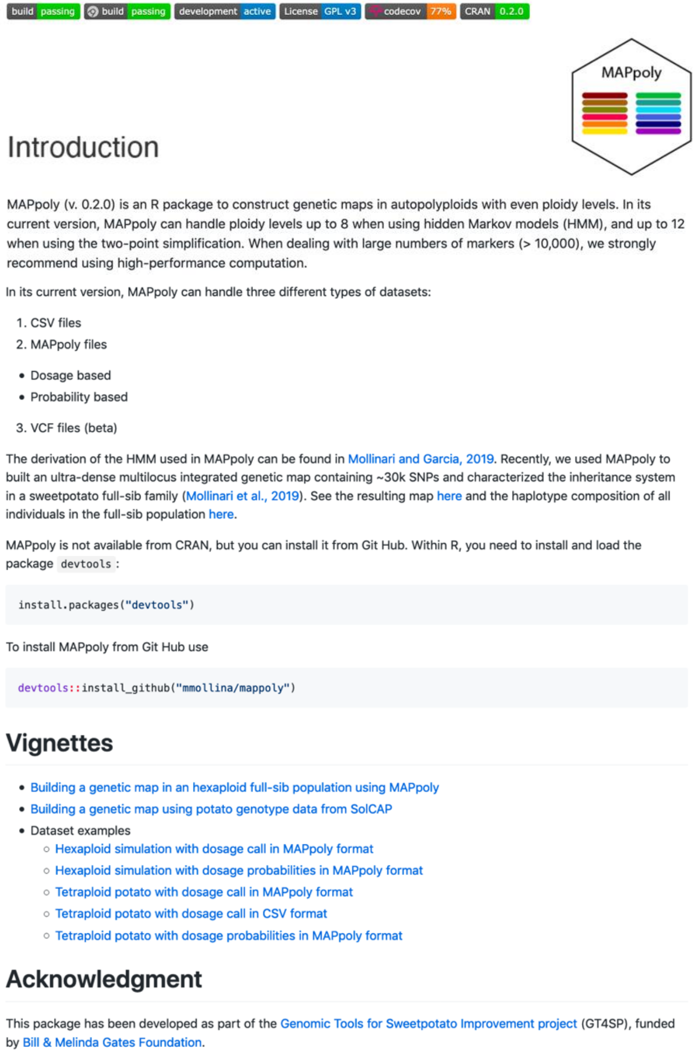




Sweetpotato genetic map



MAPpoly – R package to construct multipoint genetic maps in polyploids



Introduction

MAPpoly (v. 0.2.0) is an R package to construct genetic maps in autopolyploids with even ploidy levels. In its current version, MAPpoly can handle ploidy levels up to 8 when using hidden Markov models (HMM), and up to 12 when using the two-point simplification. When dealing with large numbers of markers (> 10,000), we strongly recommend using high-performance computation.

In its current version, MAPpoly can handle three different types of datasets:

1. CSV files
2. MAPpoly files

- Dosage based
- Probability based

3. VCF files (beta)

The derivation of the HMM used in MAPpoly can be found in Mollinari and Garcia, 2019. Recently, we used MAPpoly to built an ultra-dense multilocus integrated genetic map containing ~30k SNPs and characterized the inheritance system in a sweetpotato full-sib family (Mollinari et al., 2019). See the resulting map here and the haplotype composition of all individuals in the full-sib population here.

MAPpoly is not available from CRAN, but you can install it from Git Hub. Within R, you need to install and load the package `devtools`:

```
install.packages("devtools")
```

To install MAPpoly from Git Hub use

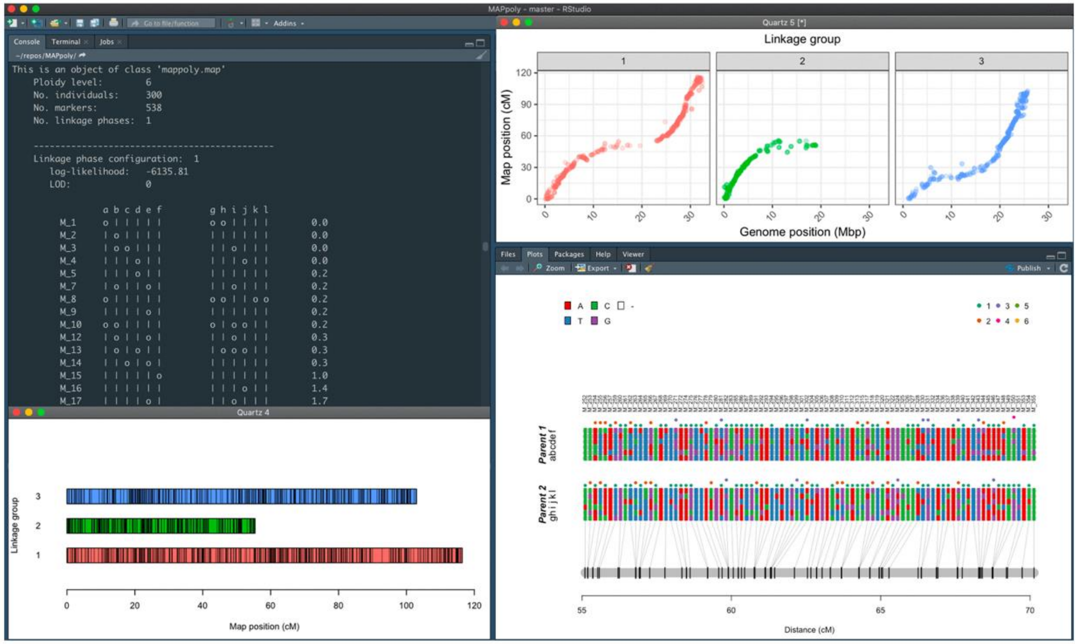
```
devtools::install_github("mmollina/mappoly")
```

Vignettes

- Building a genetic map in an hexaploid full-sib population using MAPpoly
- Building a genetic map using potato genotype data from SolCAP
- Dataset examples
  - Hexaploid simulation with dosage call in MAPpoly format
  - Hexaploid simulation with dosage probabilities in MAPpoly format
  - Tetraploid potato with dosage call in MAPpoly format
  - Tetraploid potato with dosage call in CSV format
  - Tetraploid potato with dosage probabilities in MAPpoly format

Acknowledgment

This package has been developed as part of the Genomic Tools for Sweetpotato Improvement project (GT4SP), funded by Bill & Melinda Gates Foundation.

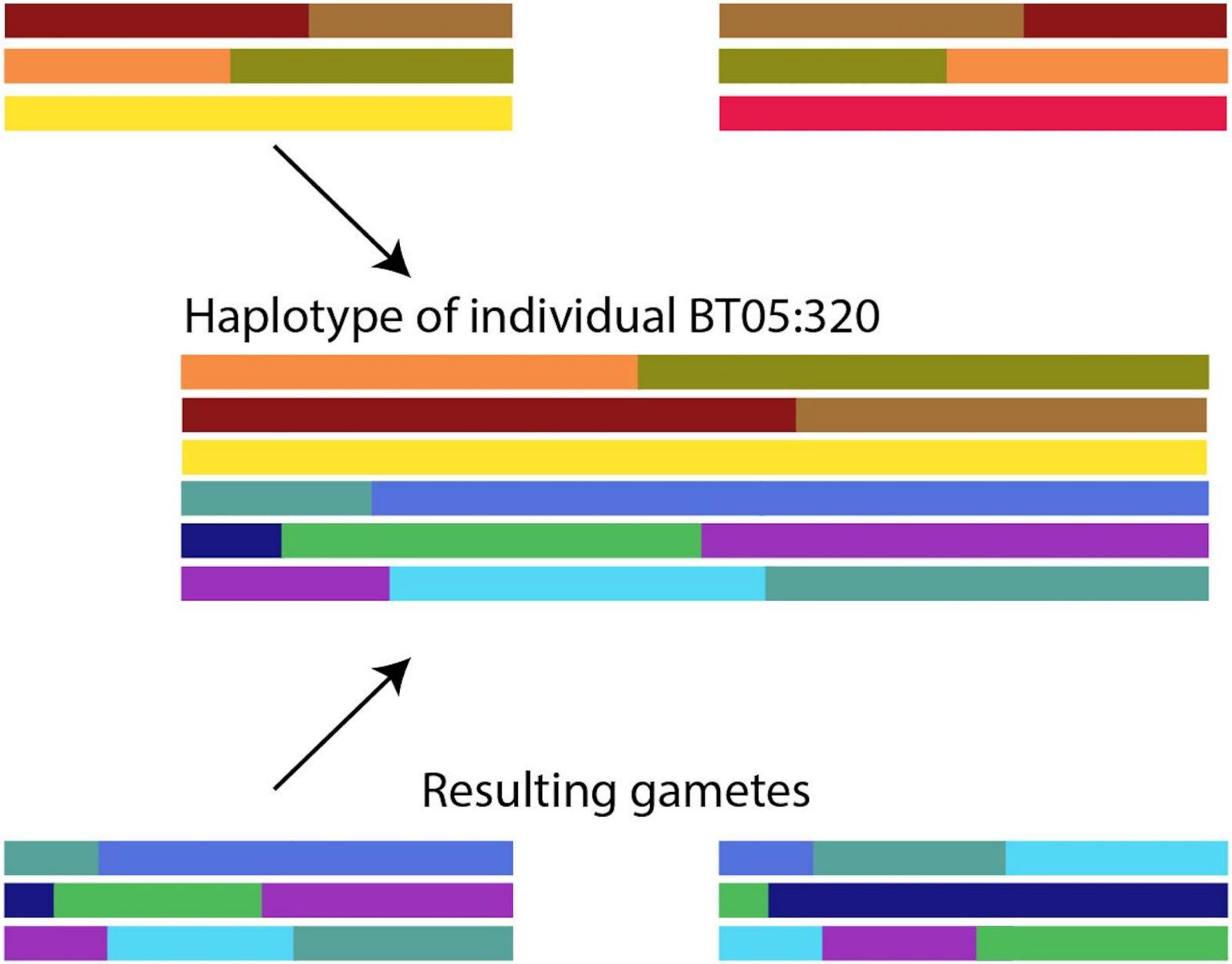


```
This is an object of class 'mappoly.map'
    Ploidy level:          6
    No. individuals:       300
    No. markers:           538
    No. linkage phases:    1

    ---------------------------------------------

    Linkage phase configuration:  1
       log-likelihood:    -6135.81
       LOD:               0

                a b c d e f        g h i j k l
       M_1      o | | | | |        o o | | | |        0.0
       M_2      | o | | | |        | | | | | |        0.0
       M_3      | o o | | |        | | o | | |        0.0
       M_4      | | | o | |        | | | o | |        0.0
       M_5      | | | o | |        | | | | | |        0.2
       M_7      | o | | o |        | | o | | |        0.2
       M_8      o | | | | |        o o | | o o        0.2
       M_9      | | | | o |        | | | | | |        0.2
       M_10     o o | | | |        o | o o | |        0.2
       M_12     | o | | o |        | | o | | |        0.3
       M_13     | o | o | |        | o o o | |        0.3
       M_14     | | o | o |        | | | | | |        0.3
       M_15     | | | | | o        | | | | | |        1.0
       M_16     | | | | | |        | | | o | |        1.4
       M_17     | | | | o |        | | o | | |        1.7
```

stable: https://CRAN.R-project.org/package=mappoly

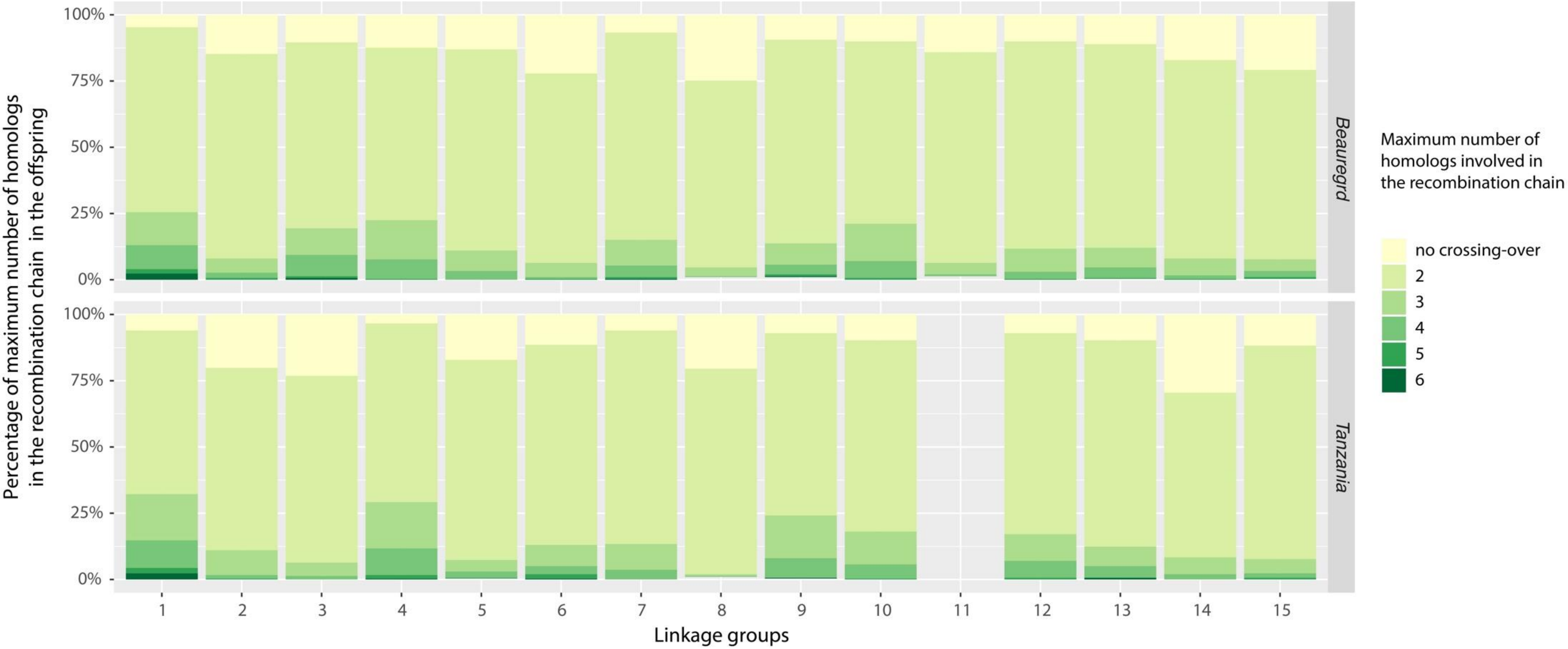
development: https://github.com/mmollina/MAPpoly

Haplotype reconstruction in the offspring



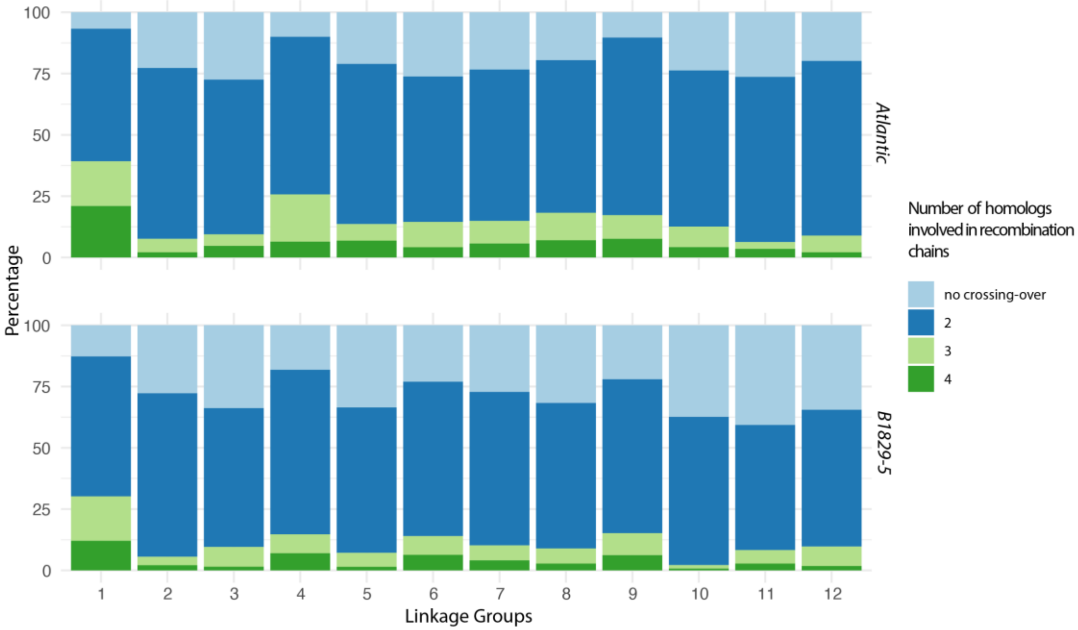
https://gt4sp-genetic-map.shinyapps.io/offspring_haplotype_BT_population/

Number of homologs involved in recombination chains



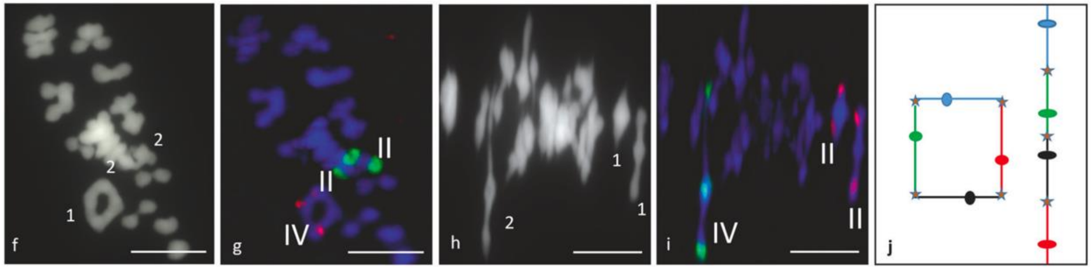
Homologs in recombination chains – potato

Pereira *et al.* (2020) - Recombination landscape in a *Solanum tuberosum* cv.

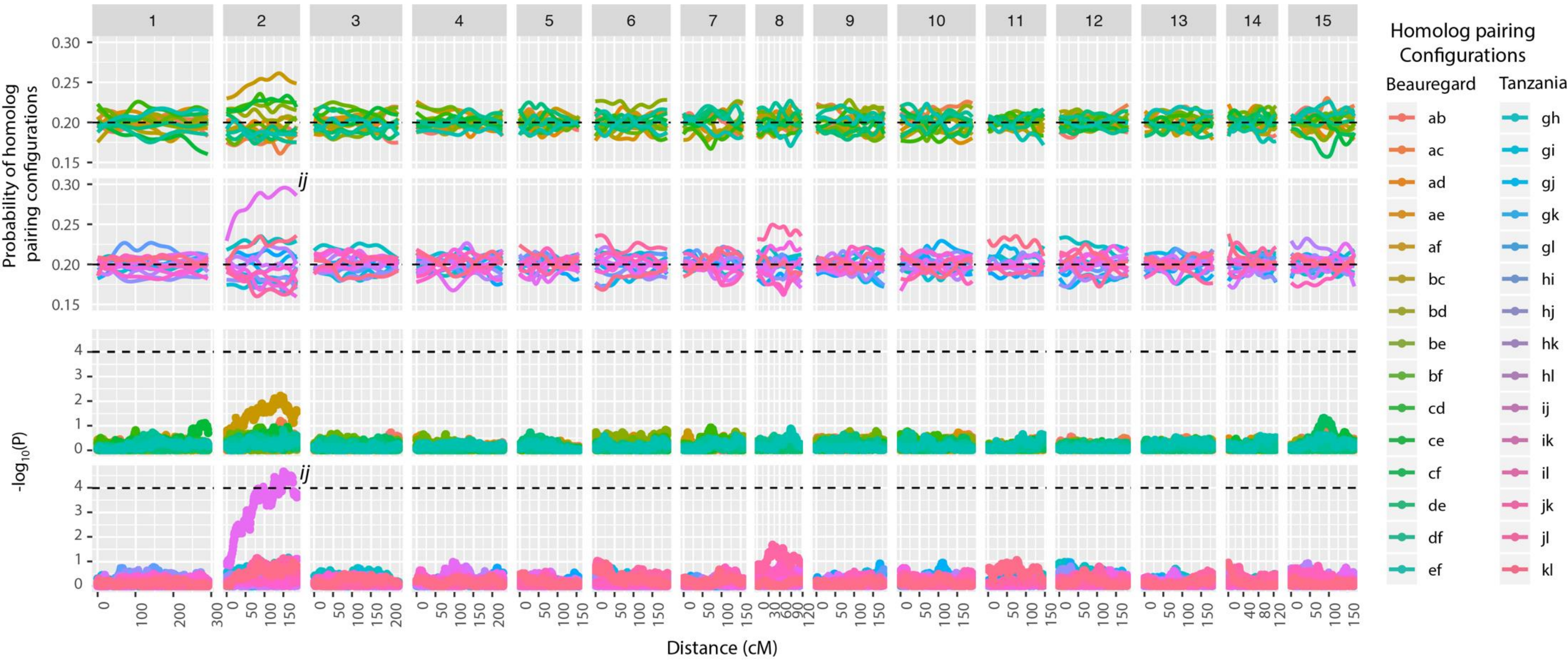


	Choudhary *et al.* (2020)	**Pereira *et al.* (2020)**
bivalents	predominant	62.3 %
multivalents	7~48%	2.2~39.2%

Choudhary *et al.* (2020) – Using fluorescence *in situ* hybridization (FISH) 5S rDNA probe (red) and 45S rDNA probe (green)



Preferential pairing profiles: Sweetpotato is vastly autohexaploid



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