Importance of polyploidy, inheritance patterns and manipulation.

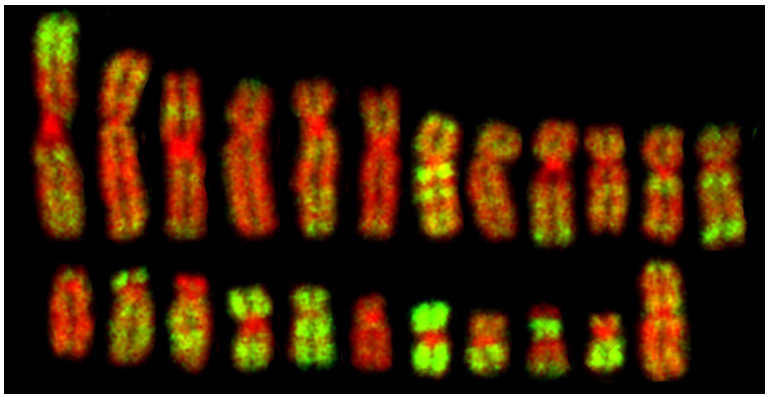
Marcelo Mollinari
mmollin@ncsu.edu

Bioinformatics Research Center - Hicks Hall 332
Department of Horticultural Science
North Carolina State University

March 19, 2019 - HS 703 - Raleigh, North Carolina

Ploidy level

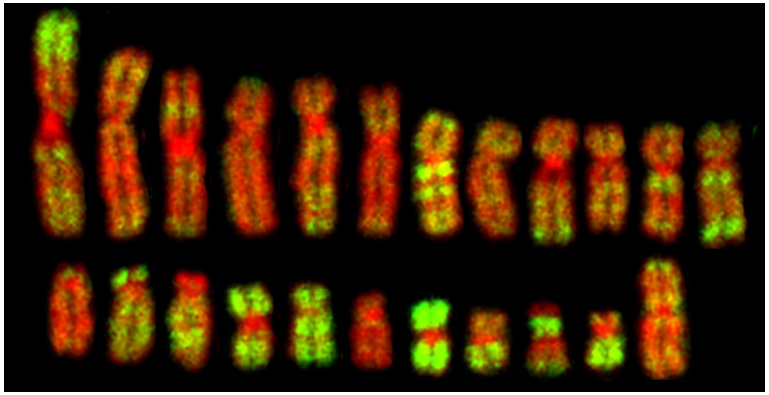
- **Basic chromosome number**: the number of different chromosomes that make up a single complete set. Humans, $x = 23$



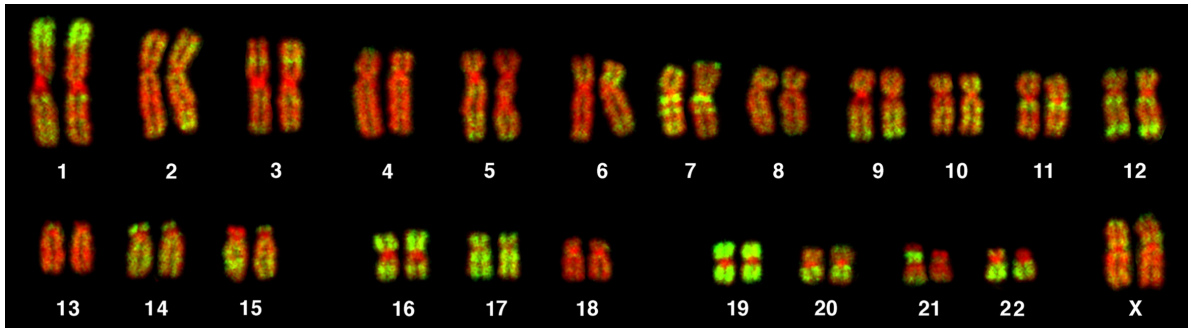
- **Ploidy level**: Number of basic chromosome sets. Humans $p = 2$

Ploidy level

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

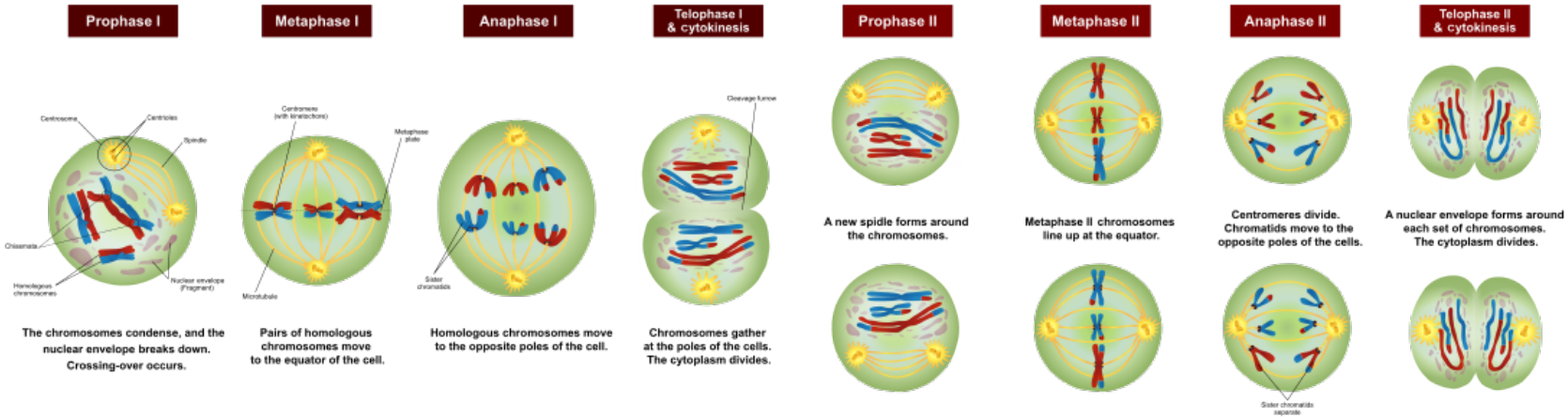


- **Ploidy level**: Number of basic chromosome sets. Humans $p = 2$



Meiotic pairing and gamete formation in diploids - Review

- 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 $\rightarrow$ same levels of heterozygosity as the parents
  - second division restitution (SDR): sister chromatids do not segregate in anaphase II $\rightarrow$ lower levels of heterozygosity when compared to the parents

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 $\rightarrow$ same levels of heterozygosity as the parents
  - second division restitution (SDR): sister chromatids do not segregate in anaphase II $\rightarrow$ lower levels of heterozygosity when compared to the parents

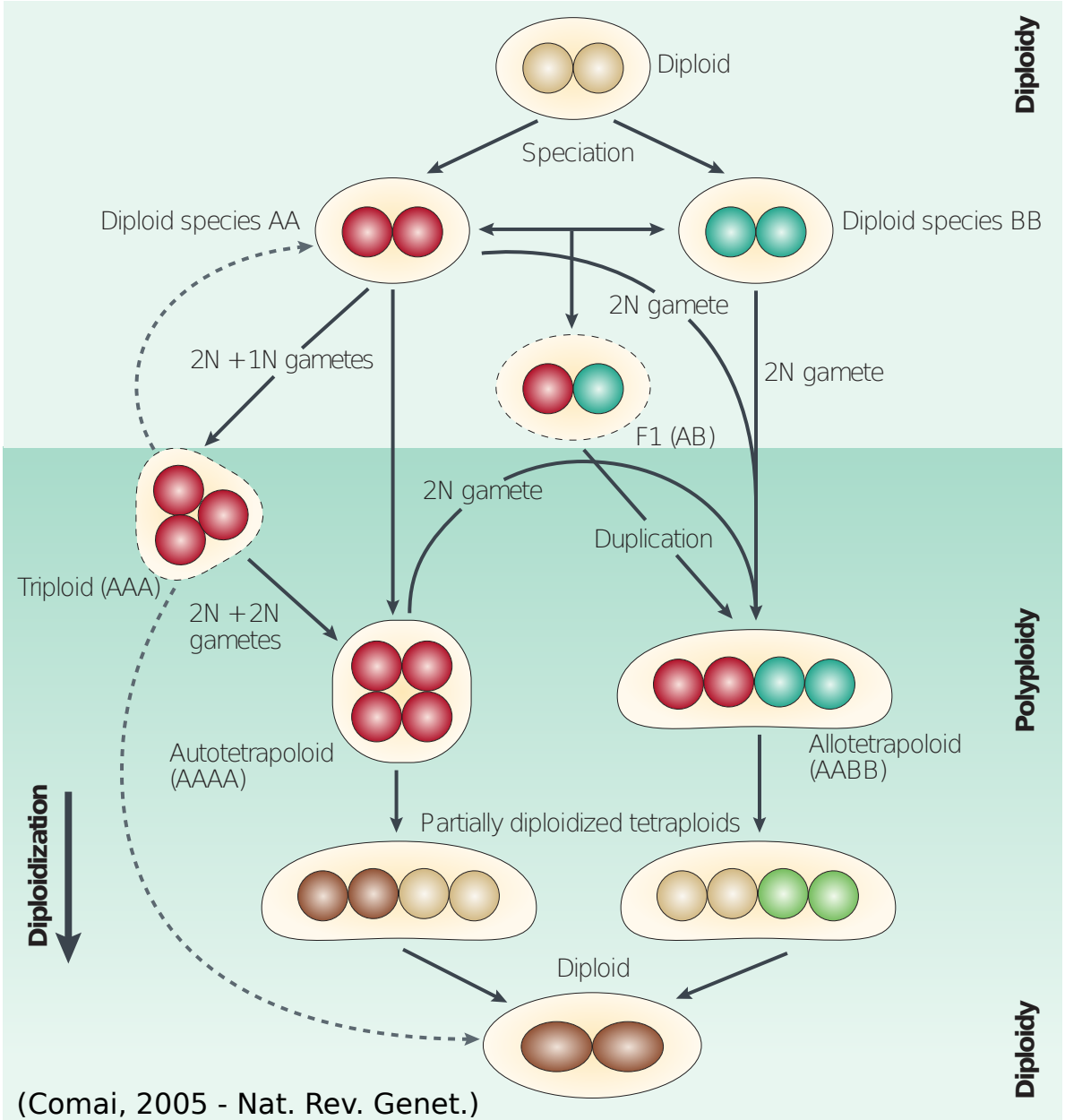
Polyploid - Definition and characteristics

- 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 $\rightarrow$ same levels of heterozygosity as the parents
  - second division restitution (SDR): sister chromatids do not segregate in anaphase II $\rightarrow$ lower levels of heterozygosity when compared to the parents

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 $\rightarrow$ same levels of heterozygosity as the parents
  - second division restitution (SDR): sister chromatids do not segregate in anaphase II $\rightarrow$ lower levels of heterozygosity when compared to the parents

How are polyploids formed?

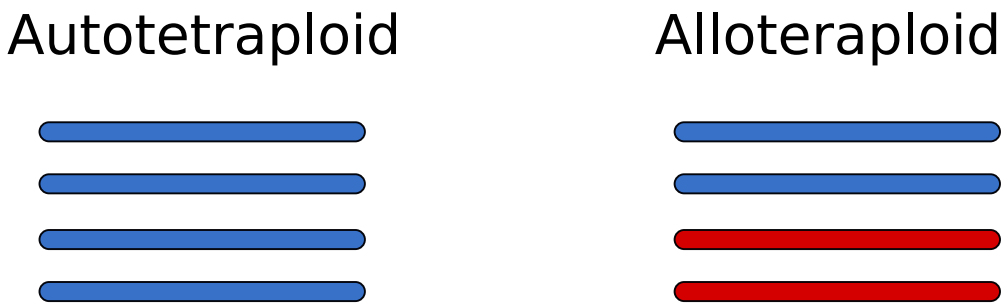


(Comai, 2005 - Nat. Rev. Genet.)

Polyploid species - Allo vs. Autopolyploids

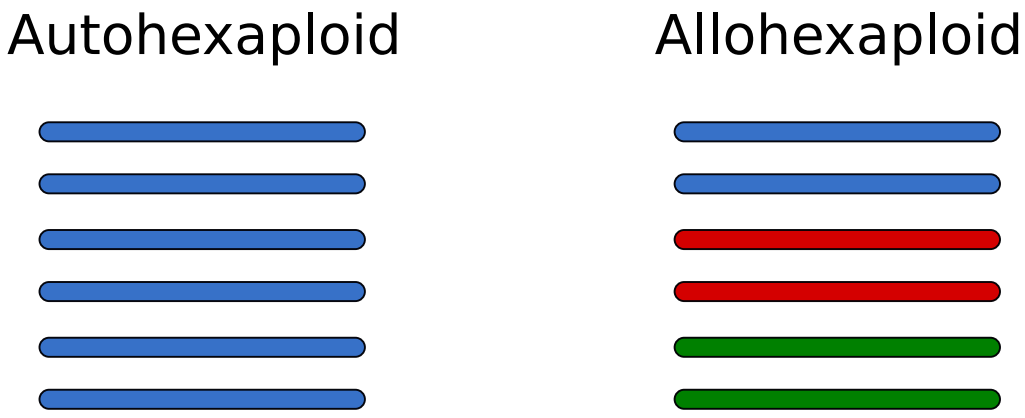
- Homologous pairing and inheritance pattern play a important role in this definition.
- Segmental allopolyploids: concept proposed by Stebbins in 1950. Occurs when a polyploid is originated from related species leading to intermediate modes of inheritance.

Polyploid species - Allo vs. Autopolyploids



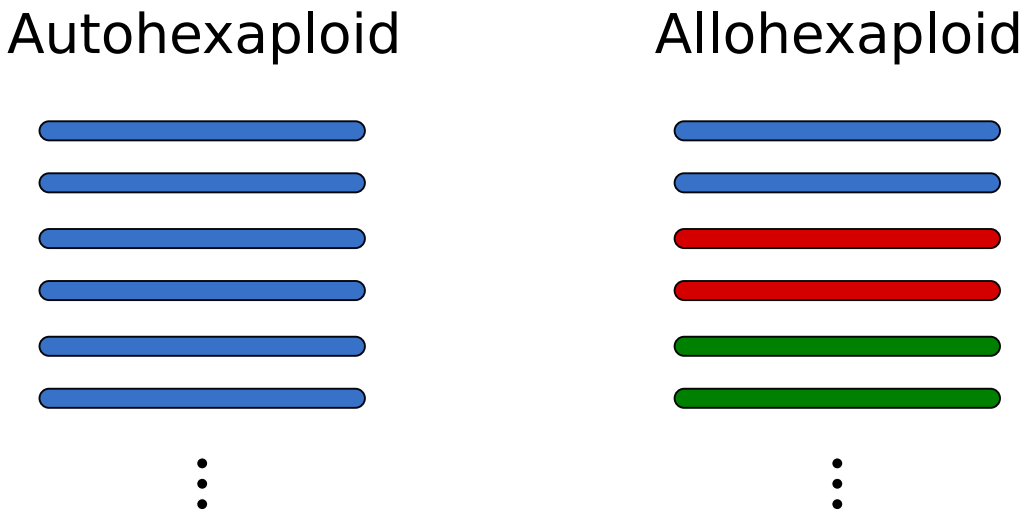
- Homologous pairing and inheritance pattern play a important role in this definition.
- Segmental allopolyploids: concept proposed by Stebbins in 1950. Occurs when a polyploid is originated from related species leading to intermediate modes of inheritance.

Polyploid species - Allo vs. Autopolyploids



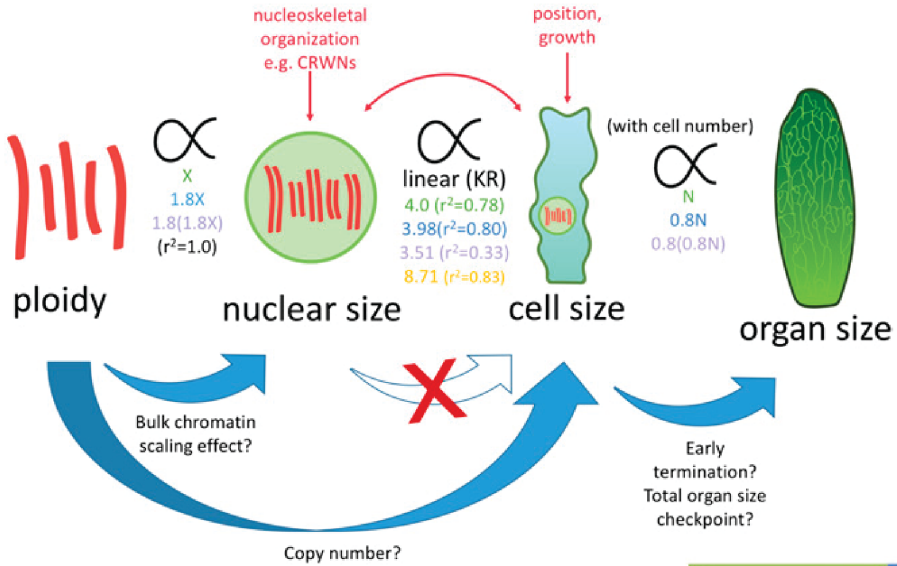
- Homologous pairing and inheritance pattern play a important role in this definition.
- Segmental allopolyploids: concept proposed by Stebbins in 1950. Occurs when a polyploid is originated from related species leading to intermediate modes of inheritance.

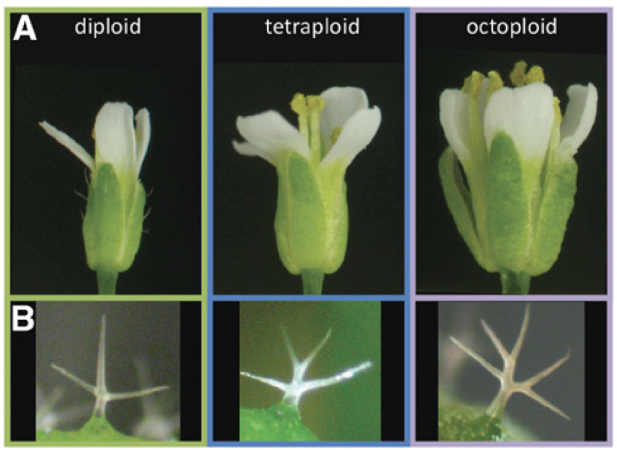
Polyploid species - Allo vs. Autopolyploids

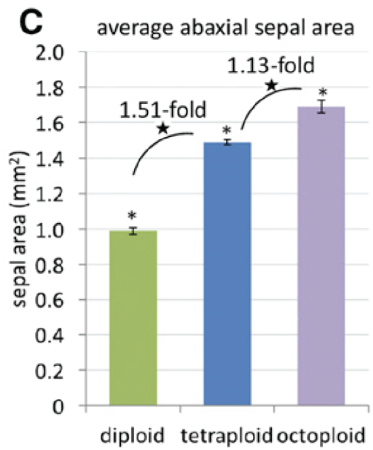


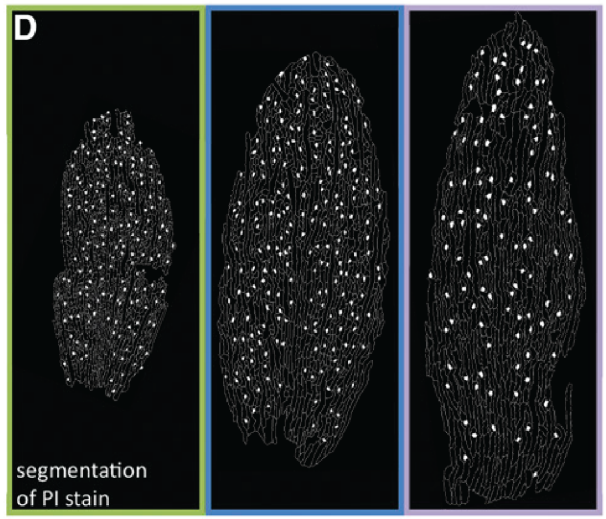
- Homologous pairing and inheritance pattern play a important role in this definition.
- Segmental allopolyploids: concept proposed by Stebbins in 1950. Occurs when a polyploid is originated from related species leading to intermediate modes of inheritance.

Ploidy and Size









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

Polyploidy consequences

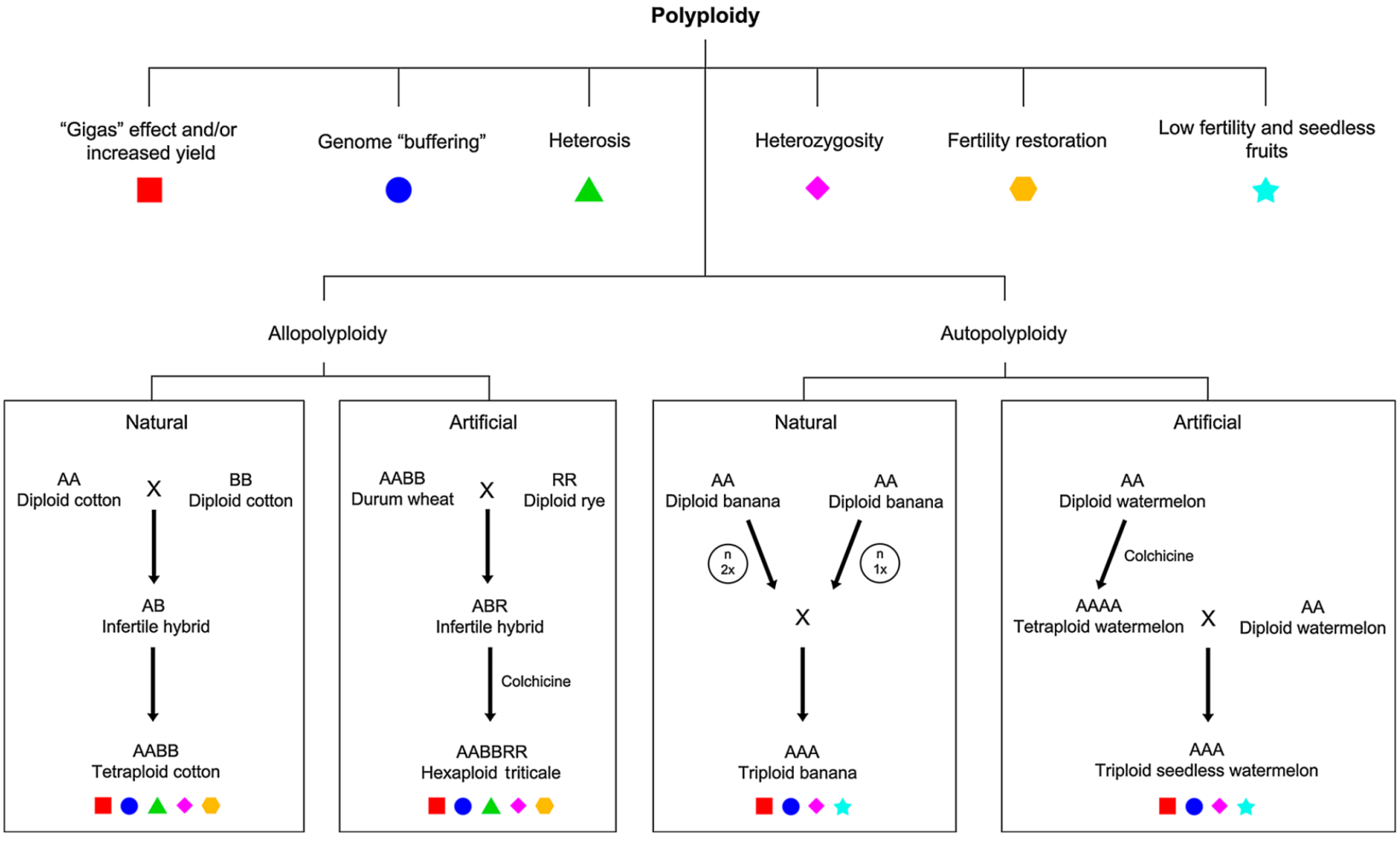
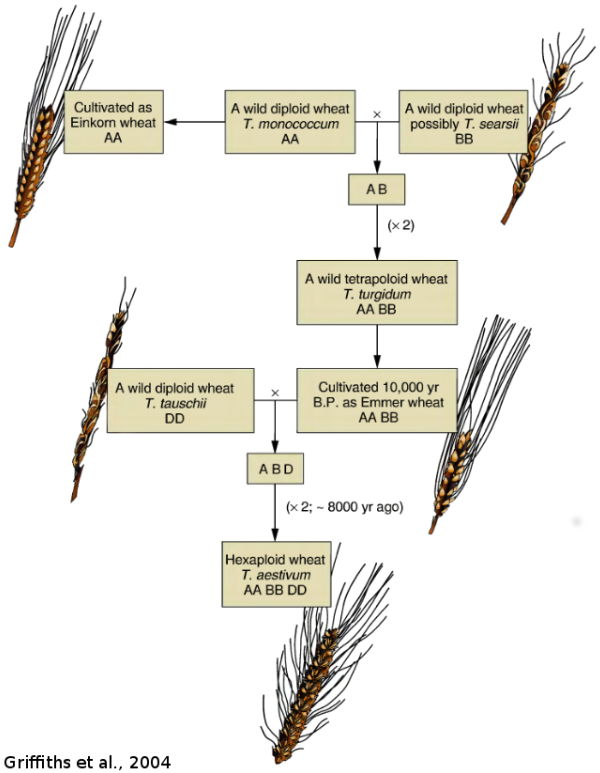


Fig. 1 Schematic representation of four cultivated species and some of the main polyploidy consequences for application in crop improvement. The *symbols* "n 1*x*" and "n 2*x*" refer to reduced and unreduced reproductive cells, respectively

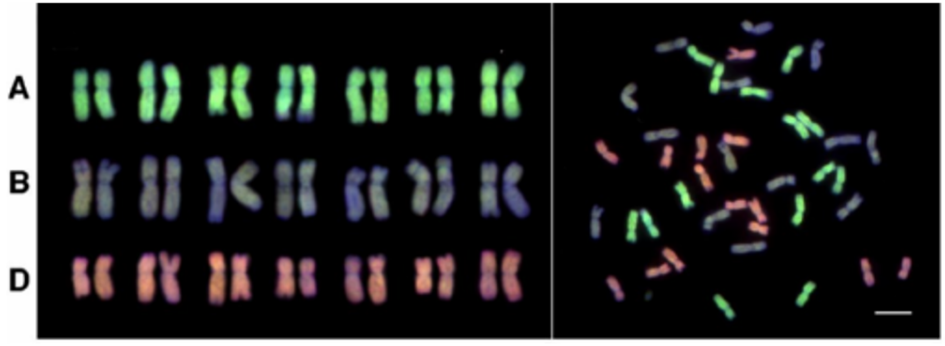
(Sattler *et al.* 2015)

Allopolyploid example - Wheat

Wheat - *Tritucum aestivum*

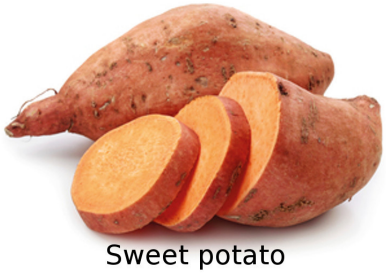


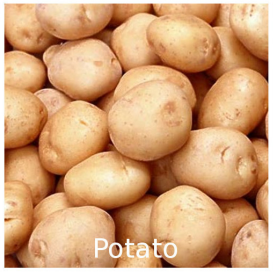
Griffiths et al., 2004



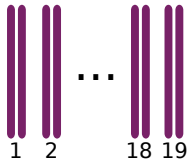
Zhang H et al. PNAS 2013;110:3447-3452

Autopolyploid example

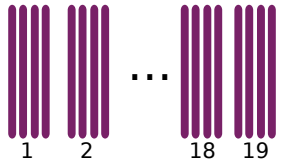




Diploid

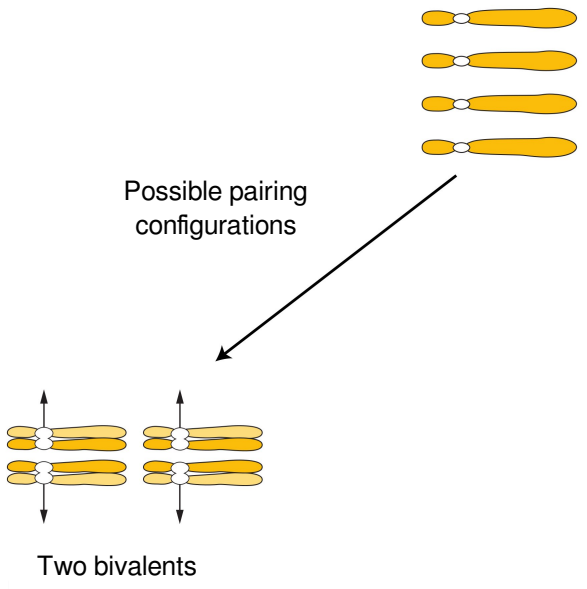


Autotetraploid



Meiotic pairing and gamete formation in autopolyploids

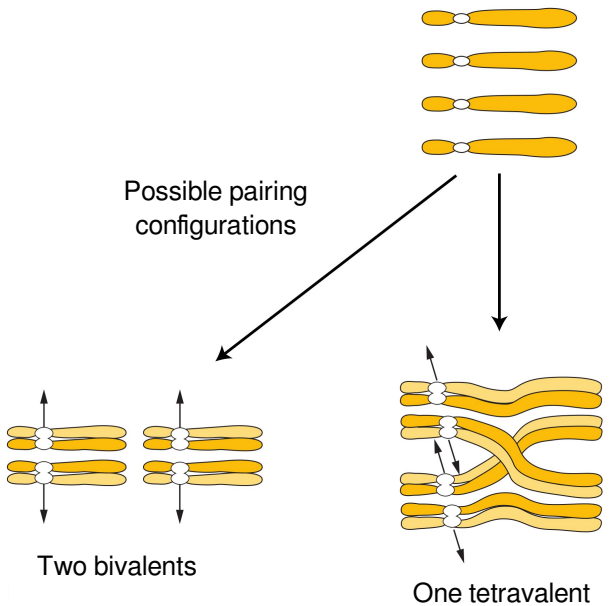
Tetraploid example



Griffiths et al. (2004)

Meiotic pairing and gamete formation in autopolyploids

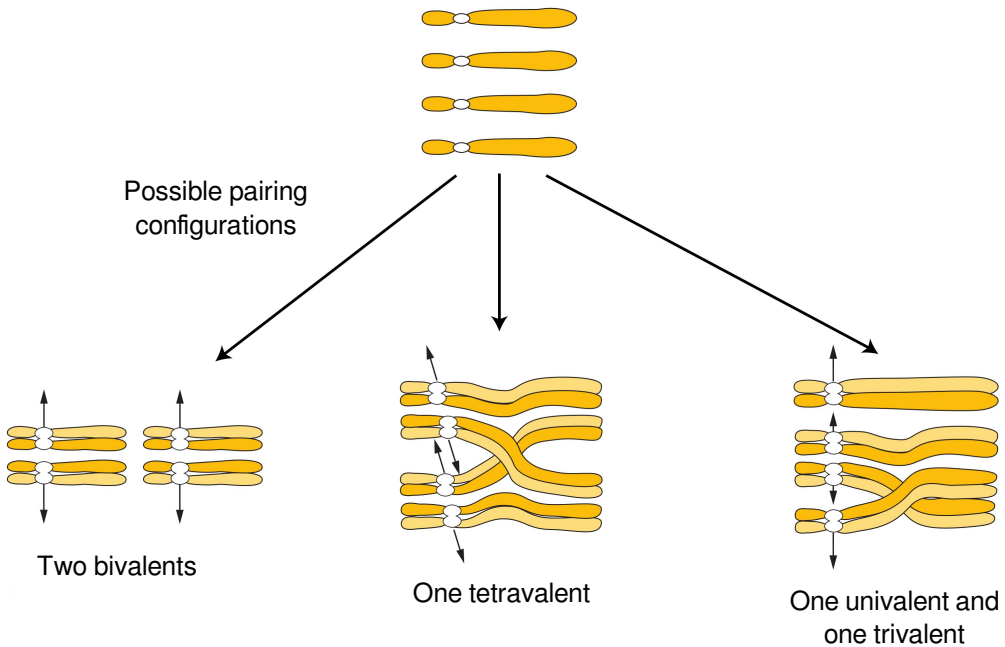
Tetraploid example



Griffiths et al. (2004)

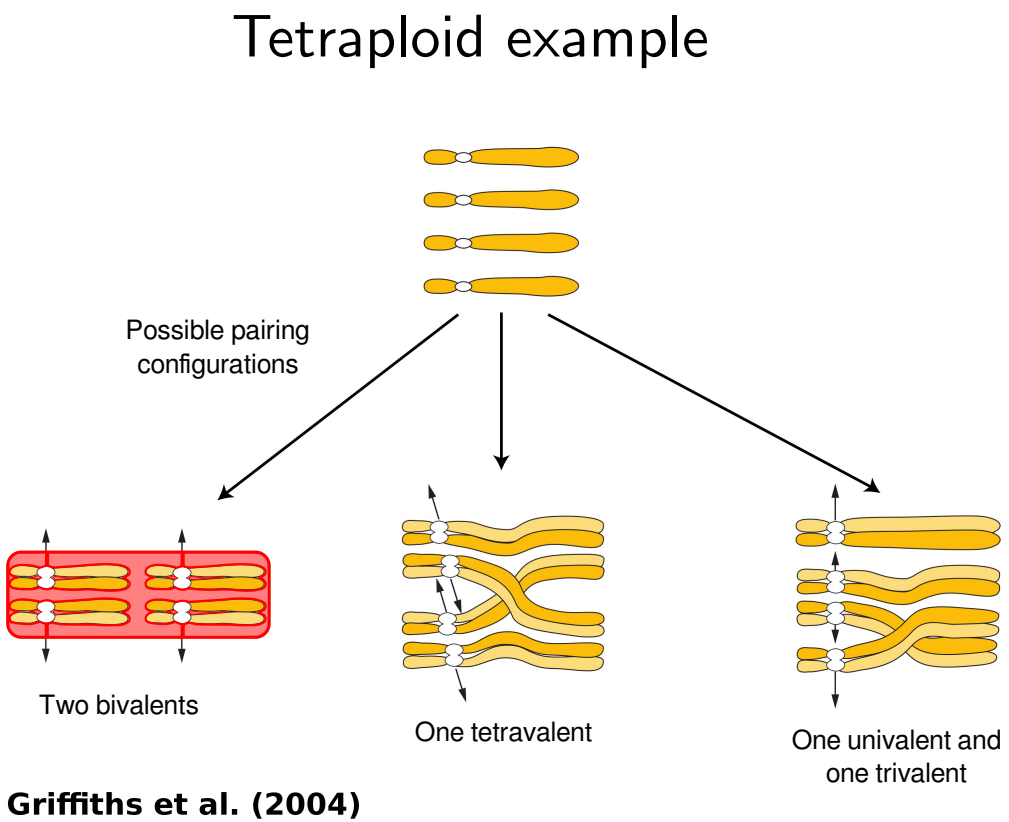
Meiotic pairing and gamete formation in autopolyploids

Tetraploid example



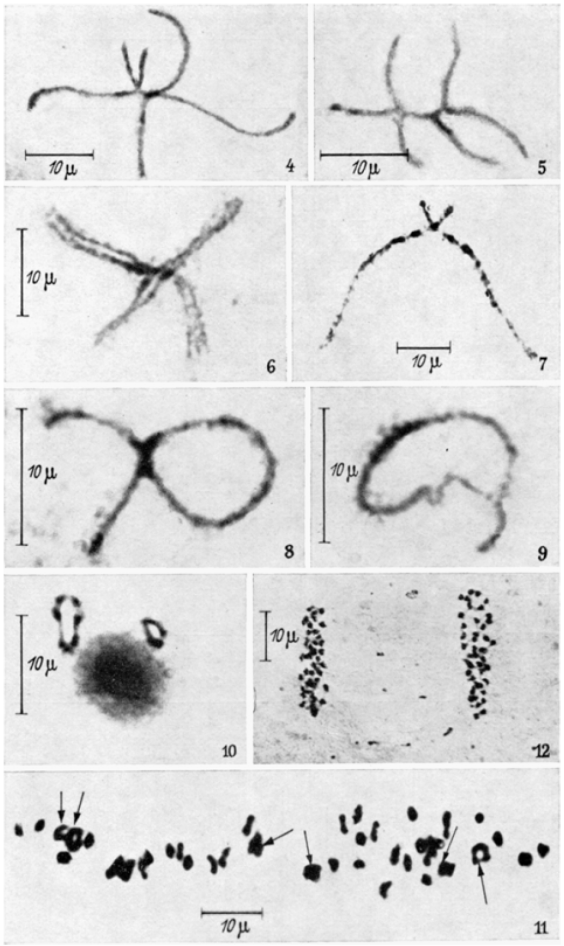
Griffiths et al. (2004)

Meiotic pairing and gamete formation in autopolyploids



Meiotic pairing in autopolyploids

Sweetpotato



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

Sugarcane

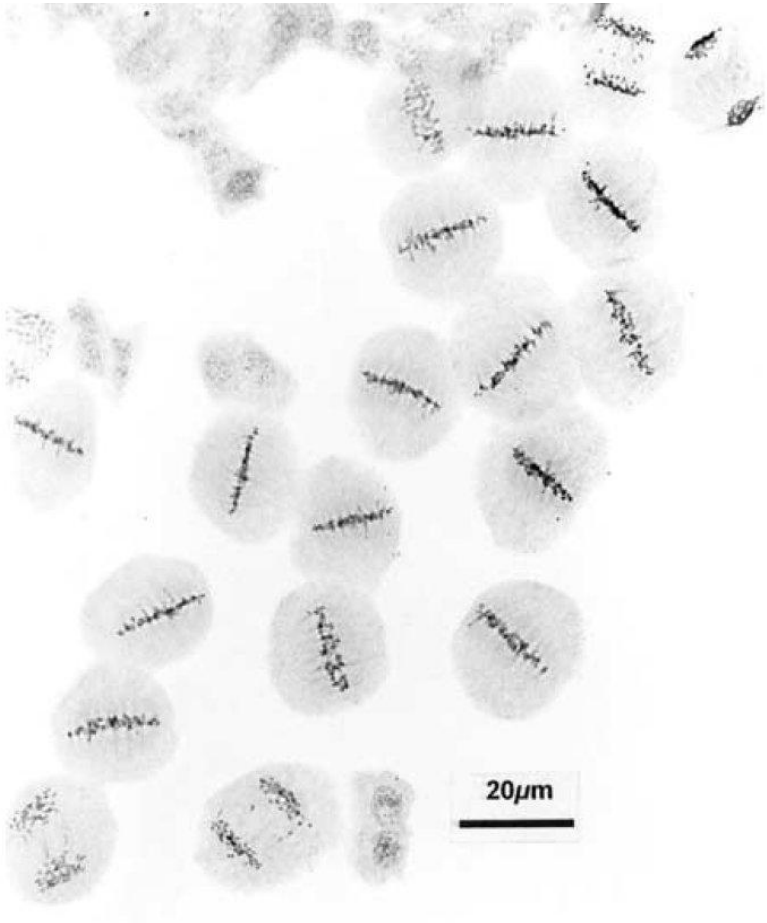


Figure 1. PMCs of *Saccharum* spp. hybrid clone 79N9059 at meiosis. As was the case in other clones, pairing was regular, bivalents generally formed. (Bielig *et al.* 2003)

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 1936

(*Galton Laboratory, University College, London*)

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

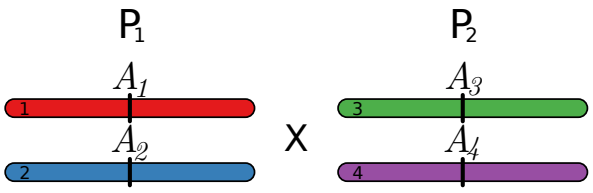
Statistical Laboratory, Iowa State College, Ames, Iowa

Received June 3, 1954

"Old" literature: still important!

Accessing the allelic variation - Diploids

Multiallelic



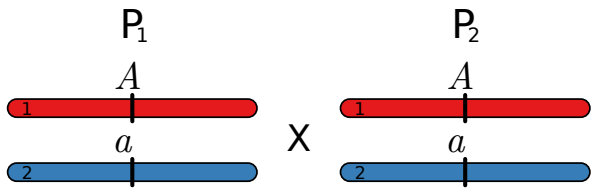
Gametes

P2 \ P1	A_1	A_2
A_3	A_1A_3	A_2A_3
A_4	A_1A_4	A_2A_4

4 possible genotypes

1:1:1:1

Biallelic



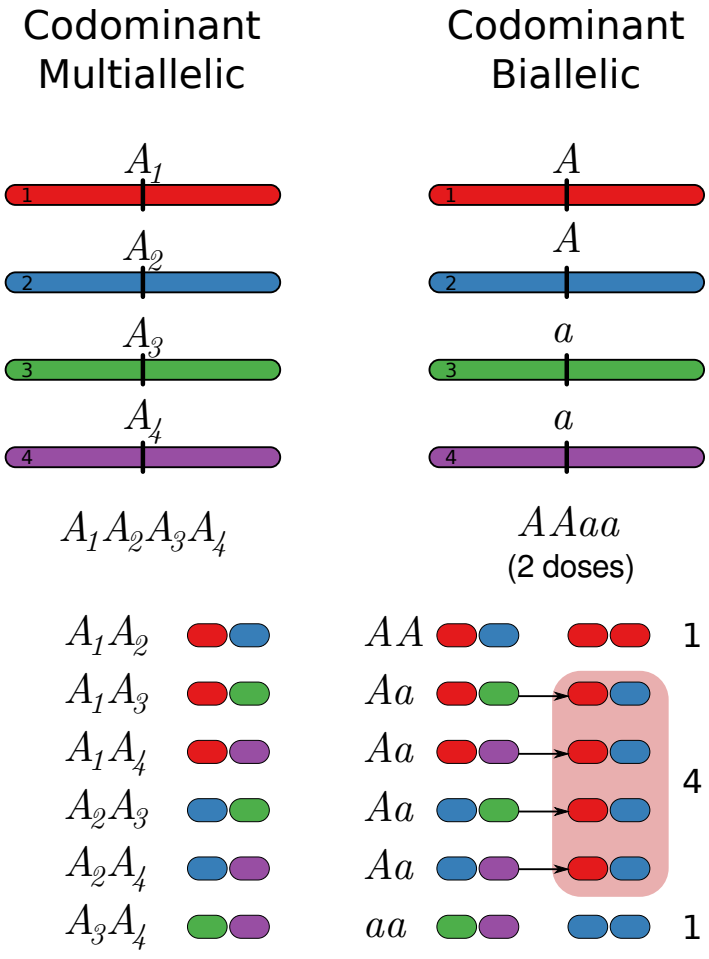
Gametes

P2 \ P1	A	a
A	AA	Aa
a	Aa	aa

3 possible genotypes

1:2:1

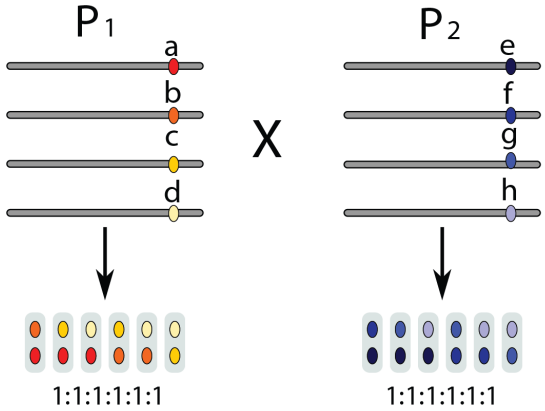
Biallelic vs. Multiallelic - Polyploids



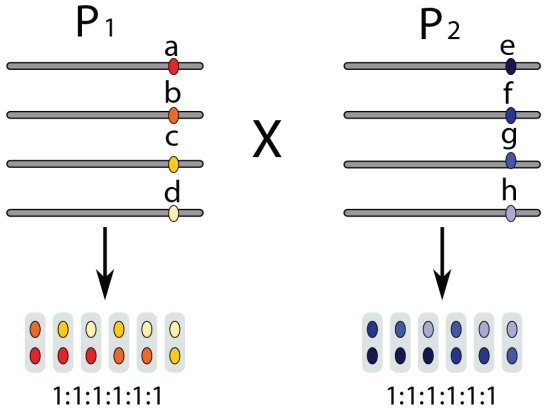
Number of possible gametes considering one locus in one and two parents

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

Inheritance pattern - Autotetraploid example



Inheritance pattern - Autotetraploid example

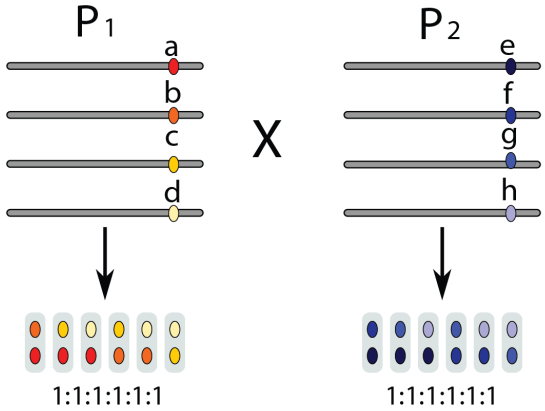


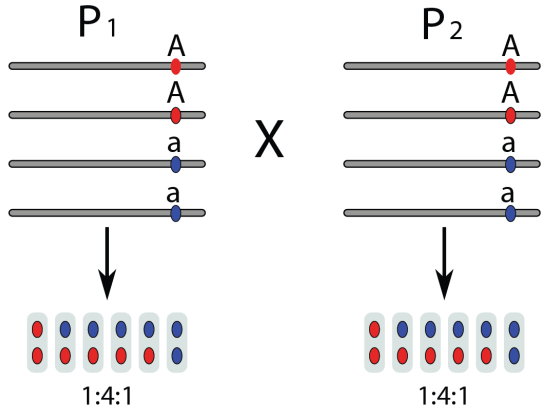
	ef	eg	eh	fg	fh	gh
ab	ab ef	ab eg	ab eh	ab fg	ab fh	ab gh
ac	ac ef	ac eg	ac eh	ac fg	ac fh	ac gh
ad	ad ef	ad eg	ad eh	ad fg	ad fh	ad gh
bc	bc ef	bc eg	bc eh	bc fg	bc fh	bc gh
bd	bd ef	bd eg	bd eh	bd fg	bd fh	bd gh
cd	cd ef	cd eg	cd eh	cd fg	cd fh	cd gh

Segregation 1 abef : 1 acef : ... : 1 bdhg : 1 cdgh

36 genotypes

Inheritance pattern - Autotetraploid example



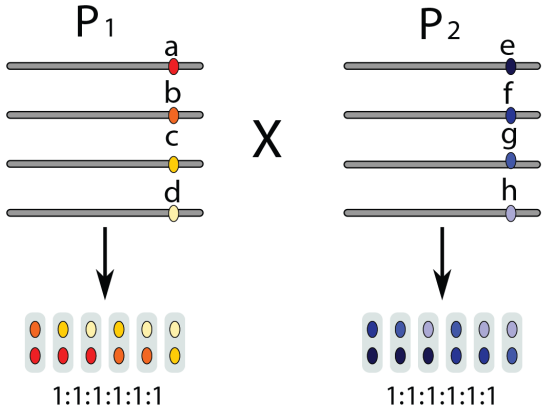


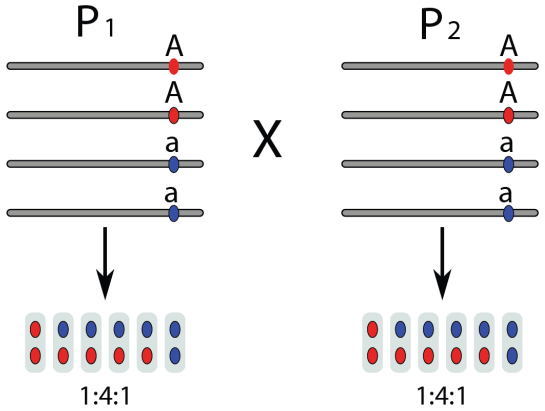
	ef	eg	eh	fg	fh	gh
ab	ab ef	ab eg	ab eh	ab fg	ab fh	ab gh
ac	ac ef	ac eg	ac eh	ac fg	ac fh	ac gh
ad	ad ef	ad eg	ad eh	ad fg	ad fh	ad gh
bc	bc ef	bc eg	bc eh	bc fg	bc fh	bc gh
bd	bd ef	bd eg	bd eh	bd fg	bd fh	bd gh
cd	cd ef	cd eg	cd eh	cd fg	cd fh	cd gh

Segregation 1 abef : 1 acef : ... : 1 bdhg : 1 cdgh

36 genotypes

Inheritance pattern - Autotetraploid example





	ef	eg	eh	fg	fh	gh
ab	ab ef	ab eg	ab eh	ab fg	ab fh	ab gh
ac	ac ef	ac eg	ac eh	ac fg	ac fh	ac gh
ad	ad ef	ad eg	ad eh	ad fg	ad fh	ad gh
bc	bc ef	bc eg	bc eh	bc fg	bc fh	bc gh
bd	bd ef	bd eg	bd eh	bd fg	bd fh	bd gh
cd	cd ef	cd eg	cd eh	cd fg	cd fh	cd gh

Segregation 1 abef : 1 acef : ... : 1 bdhg : 1 cdgh

36 genotypes

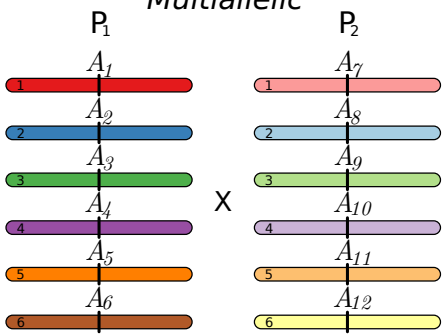
	AA	Aa	Aa	Aa	Aa	aa
AA	AAAA	AAAa	AAAa	AAAa	AAAa	AAaa
Aa	AaAA	AaAa	AaAa	AaAa	AaAa	Aaaa
Aa	AaAA	AaAa	AaAa	AaAa	AaAa	Aaaa
Aa	AaAA	AaAa	AaAa	AaAa	AaAa	Aaaa
Aa	AaAA	AaAa	AaAa	AaAa	AaAa	Aaaa
aa	aa AA	aa Aa	aa Aa	aa Aa	aa Aa	aaaa

Segregation 1 AAAA : 8 AAAa : 18 AAaa : 8 Aaaa : 1 aaaa

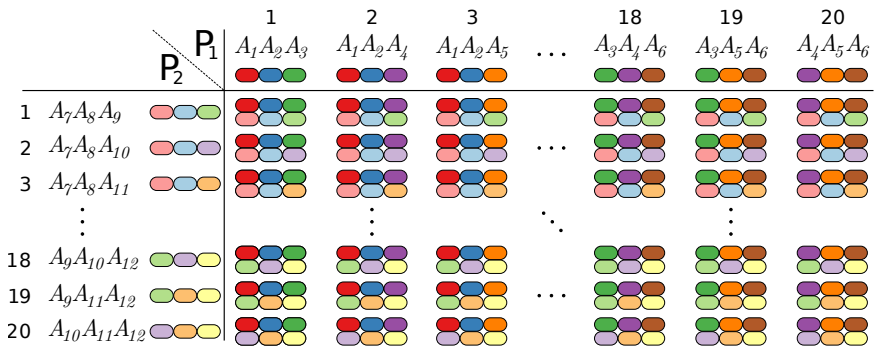
5 genotypes

Inheritance pattern - Autohexaploid Example

Multiallelic



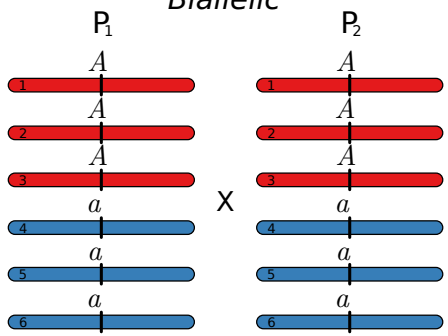
Gametes



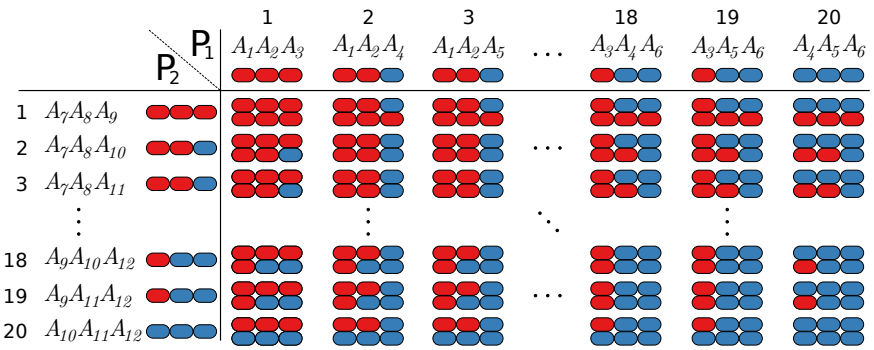
400 possible genotypes

1 : 1 : ... : 1 : 1

Biallelic

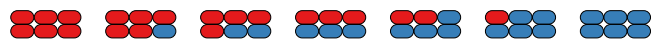


Gametes



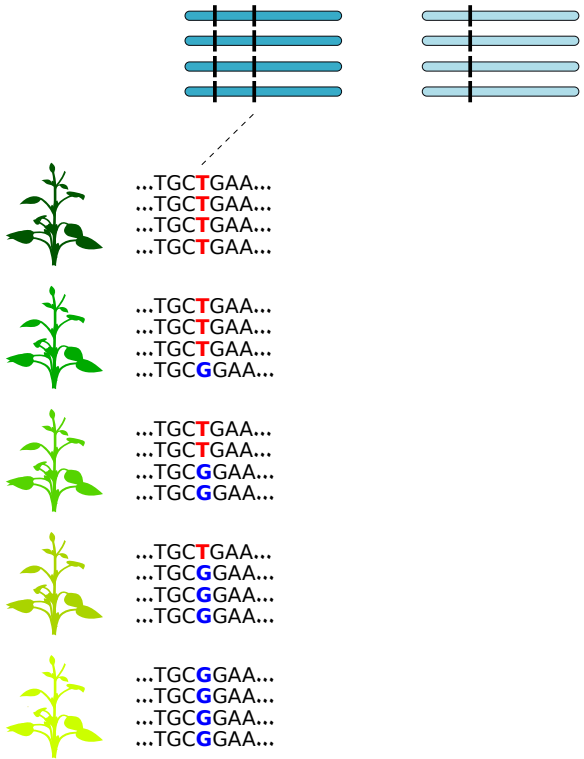
7 possible genotypes

1 : 18 : 99 : 164 : 99 : 18 : 1



Genotyping in polyploids

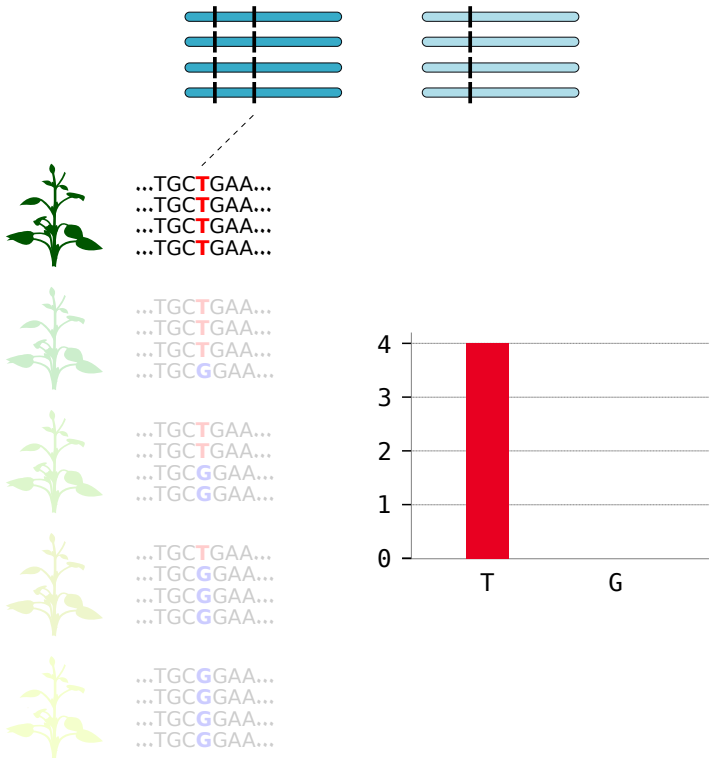
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

Genotyping in polyploids

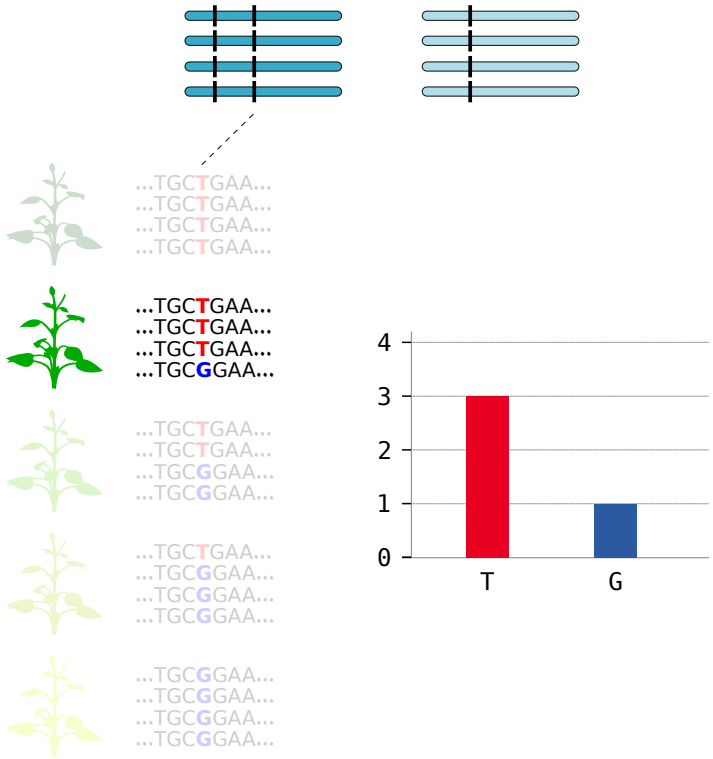
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

Genotyping in polyploids

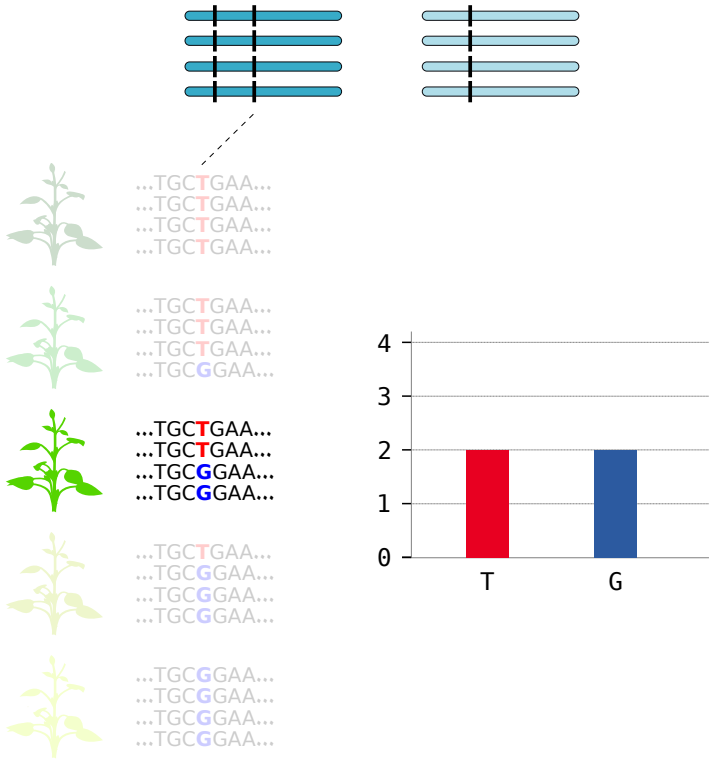
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

Genotyping in polyploids

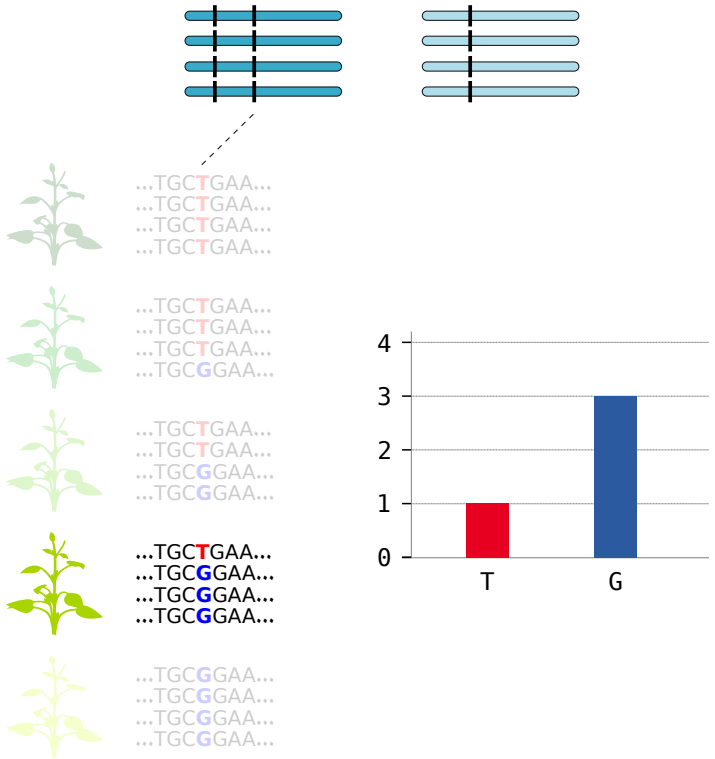
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

Genotyping in polyploids

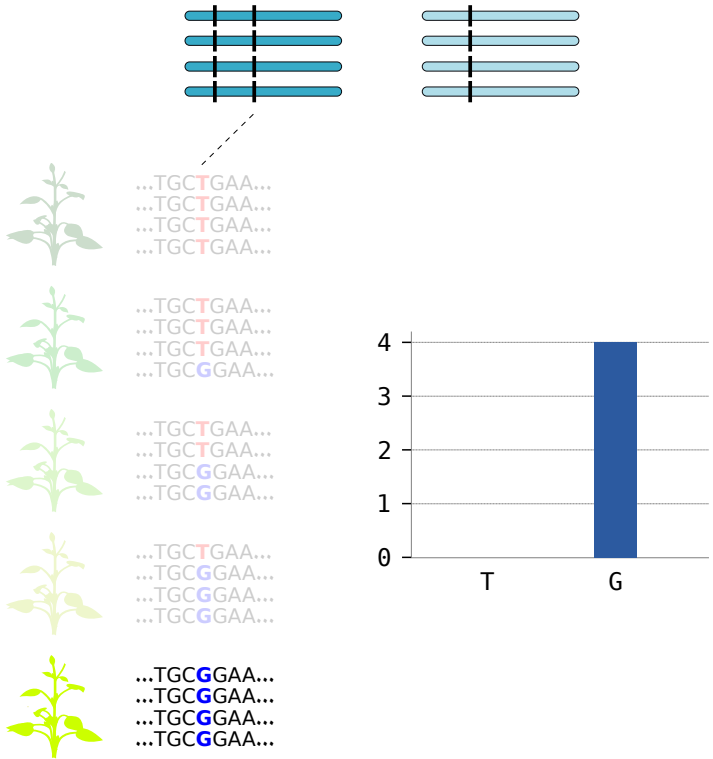
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

Genotyping in polyploids

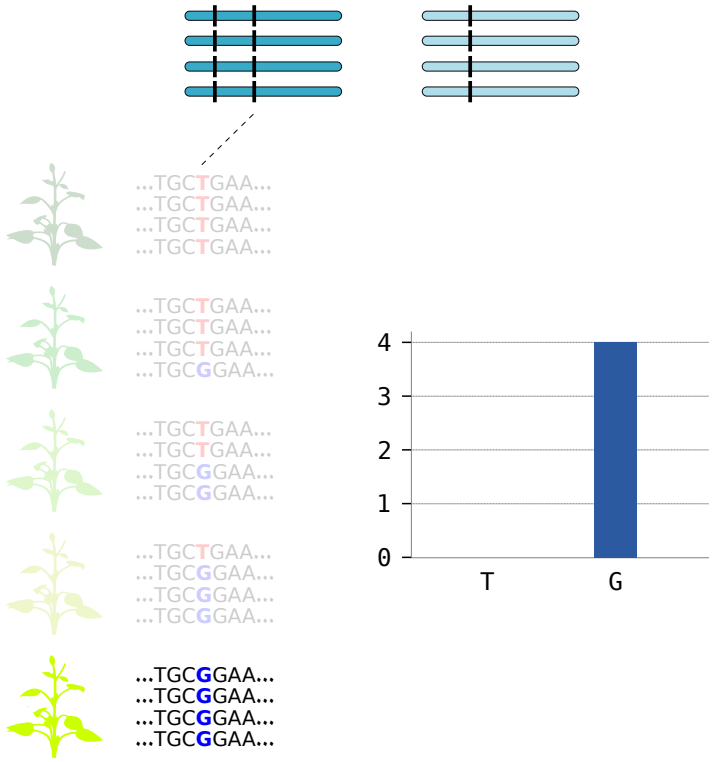
Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

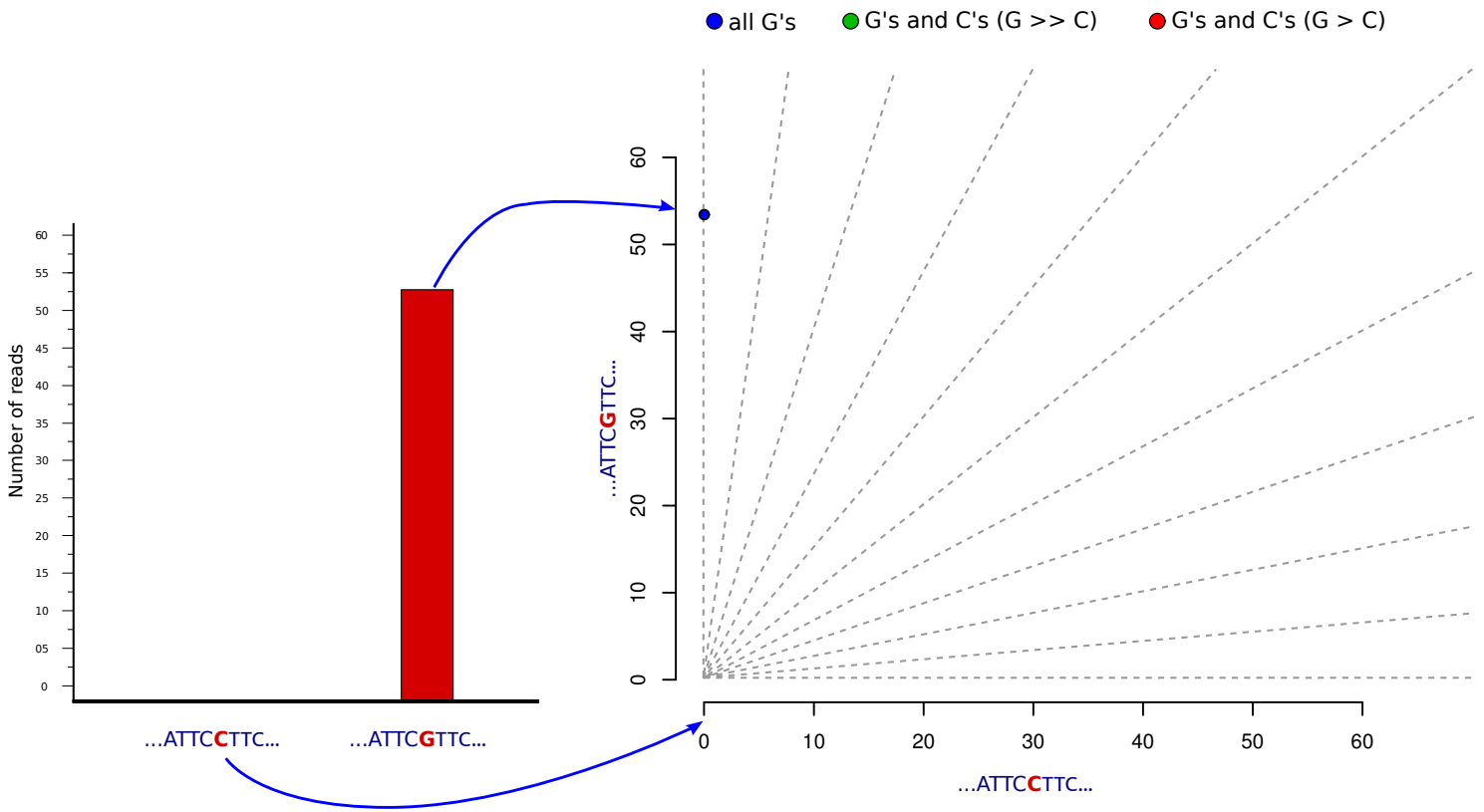
Genotyping in polyploids

Biallelic co-dominant marker



Quantitative genotyping: access the abundance of each one of the allelic forms (in a biallelic marker)

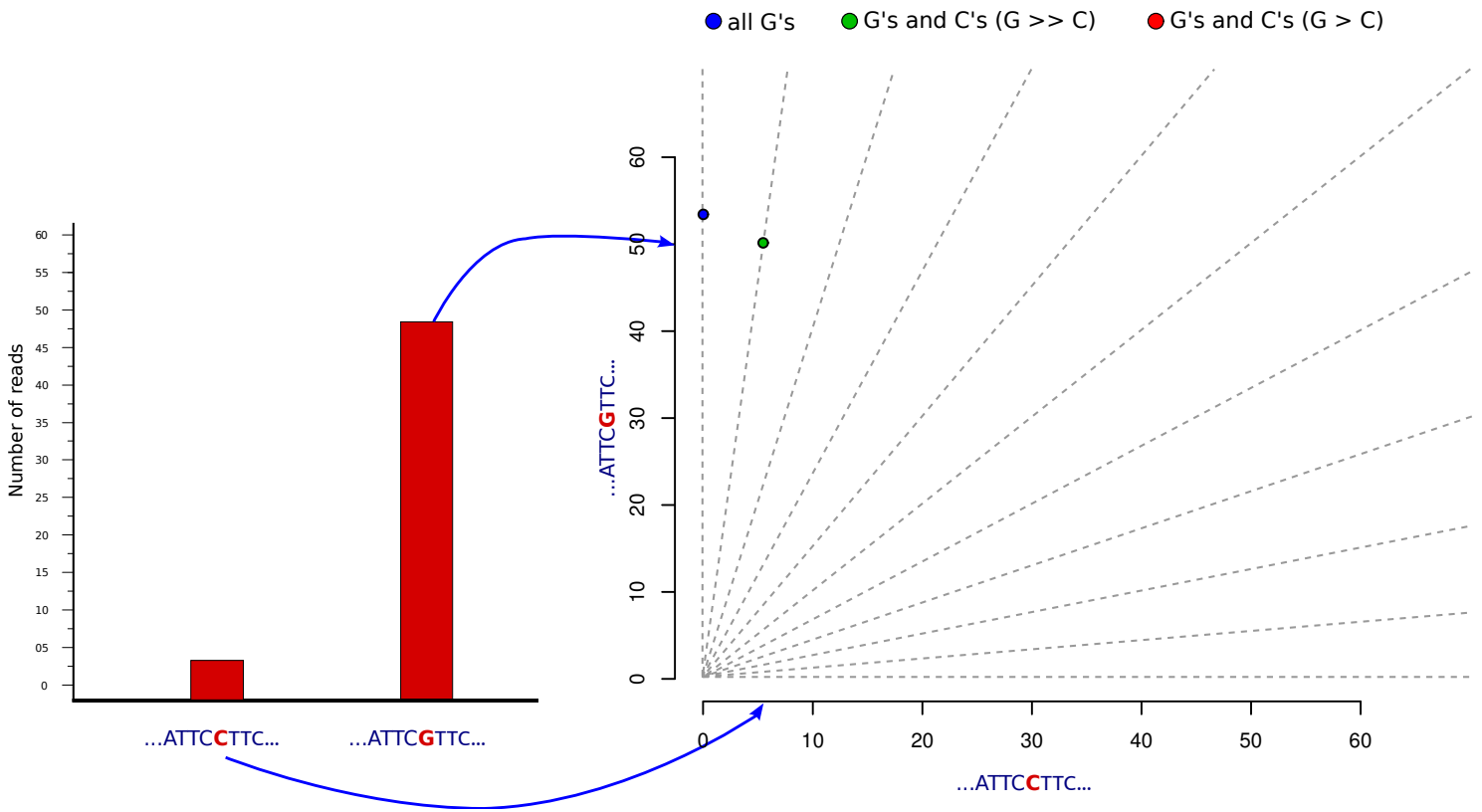
GBS scatter plot



Important

- Ratios of peaks or areas (angle): dosage

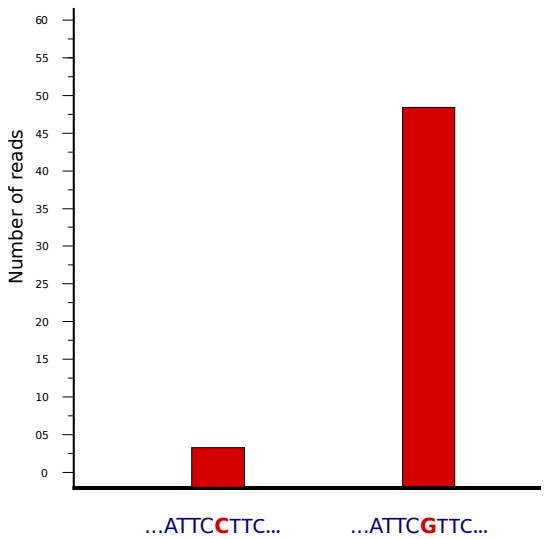
GBS scatter plot

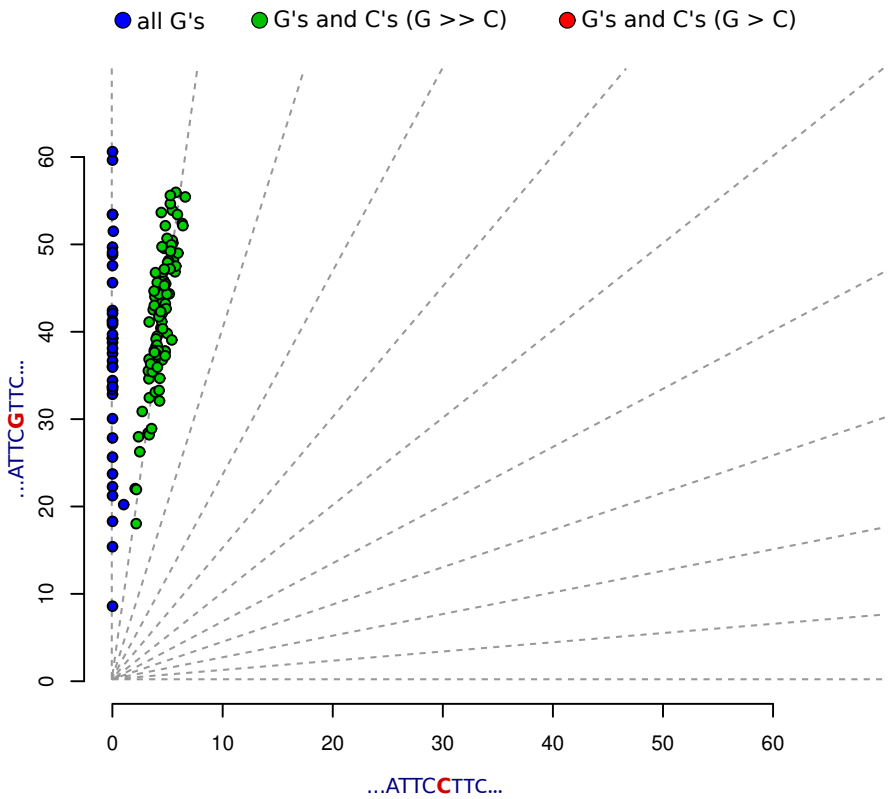


Important

- Ratios of peaks or areas (angle): dosage

GBS scatter plot

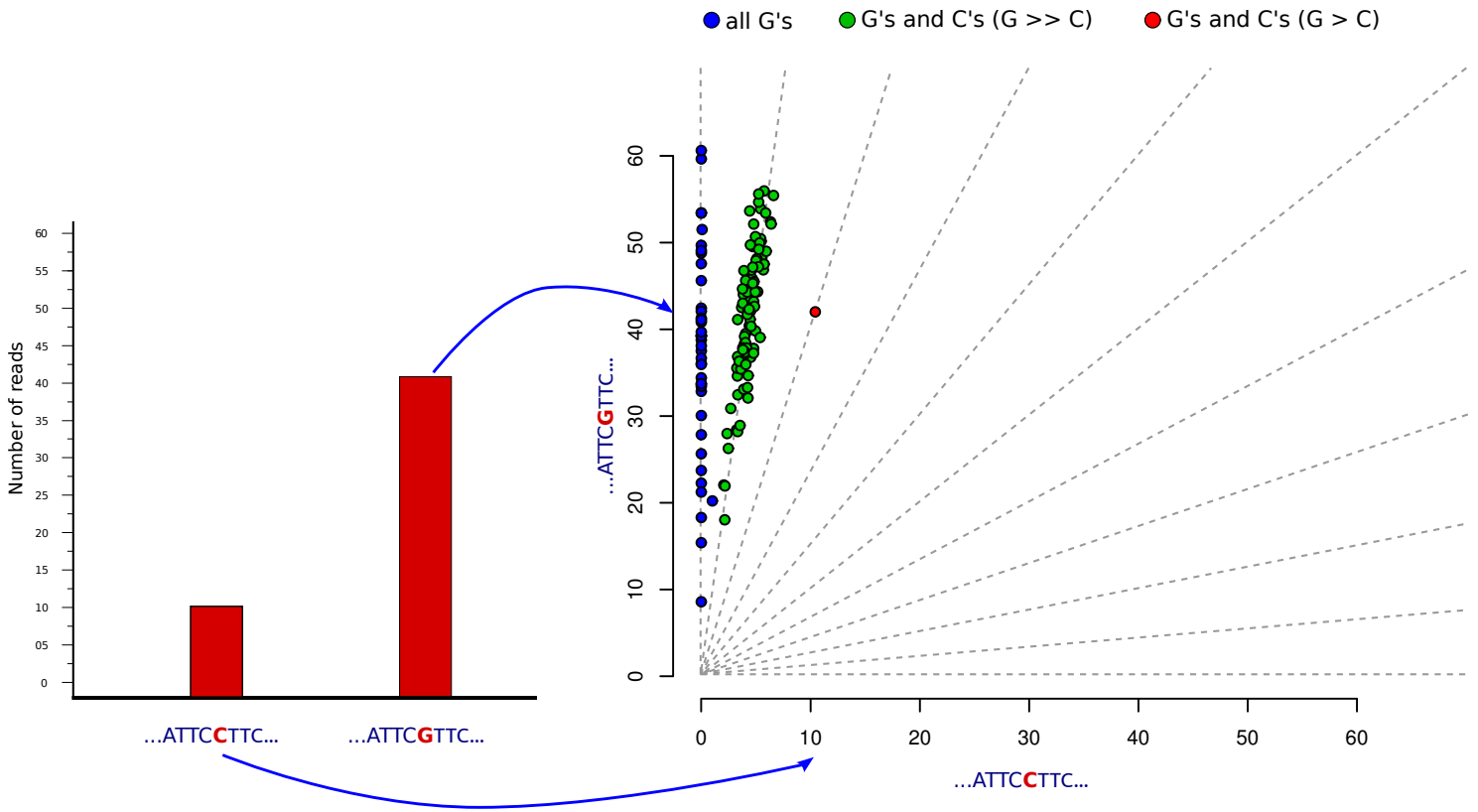




Important

- Ratios of peaks or areas (angle): dosage

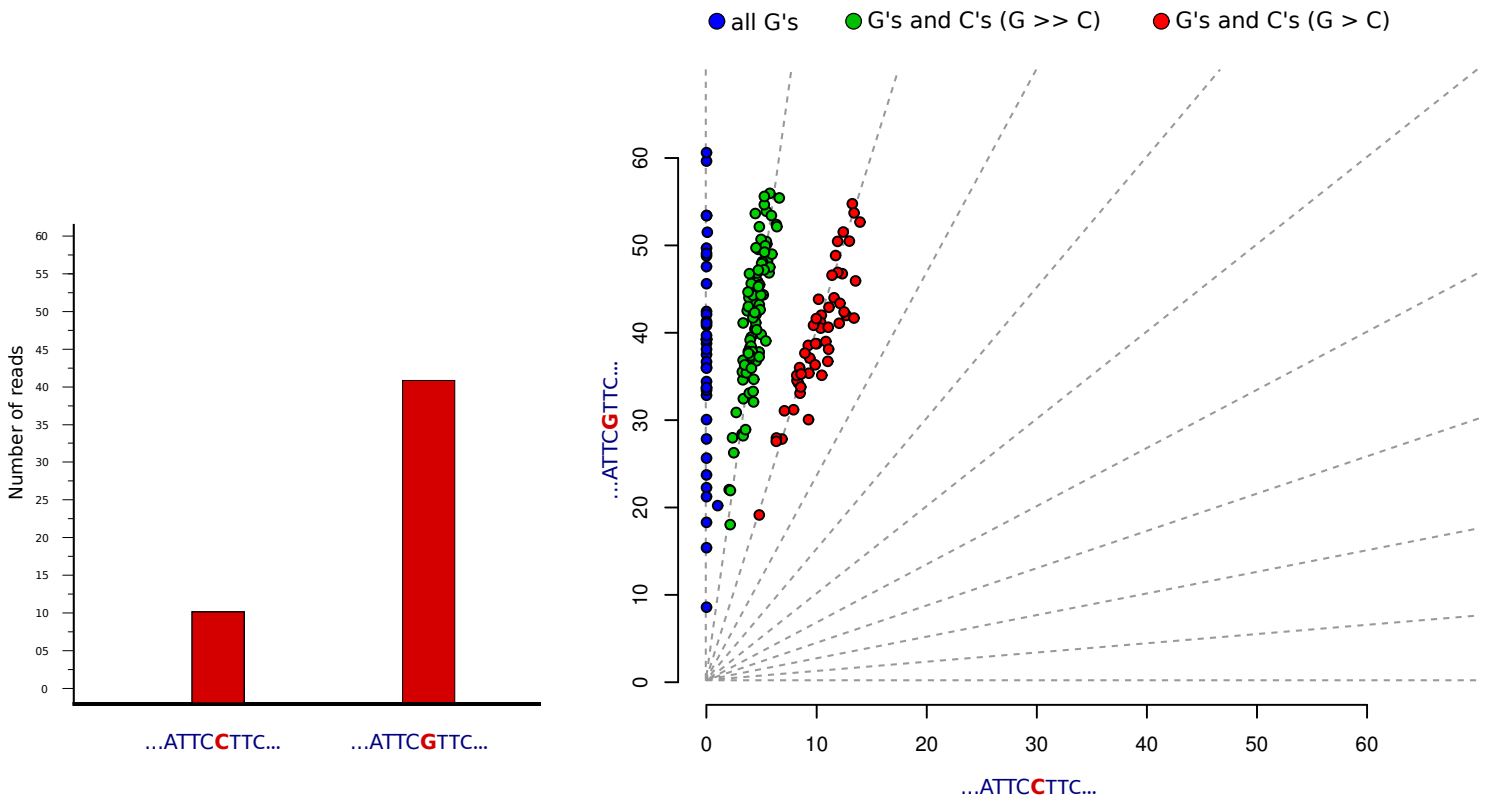
GBS scatter plot



Important

- Ratios of peaks or areas (angle): dosage

GBS scatter plot

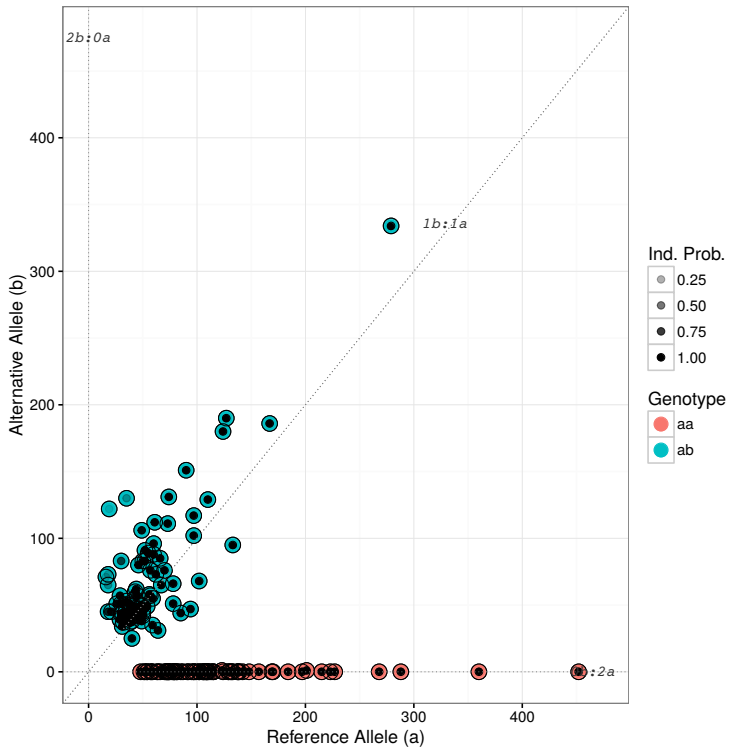


Important

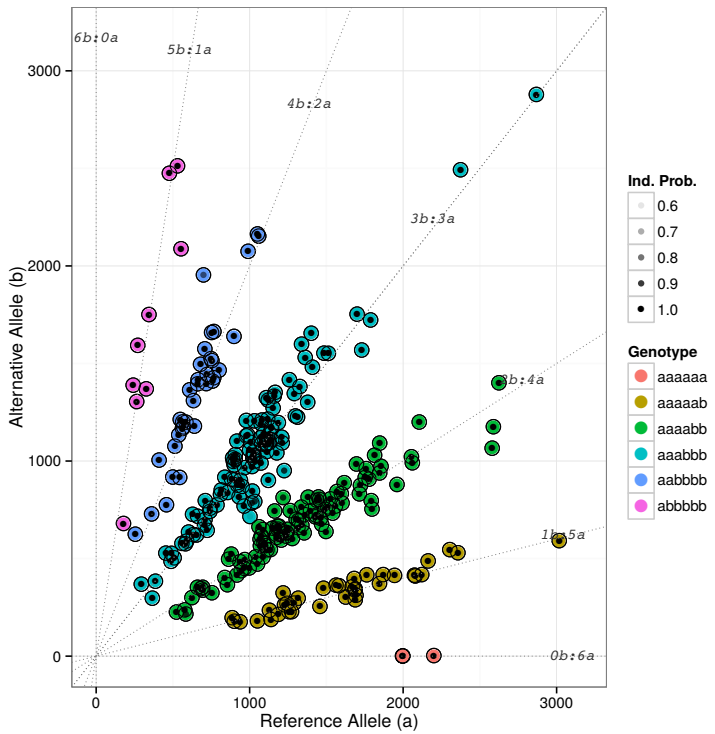
- Ratios of peaks or areas (angle): dosage

Example: SNPs in Sweet potato

Diploid



Hexaploid



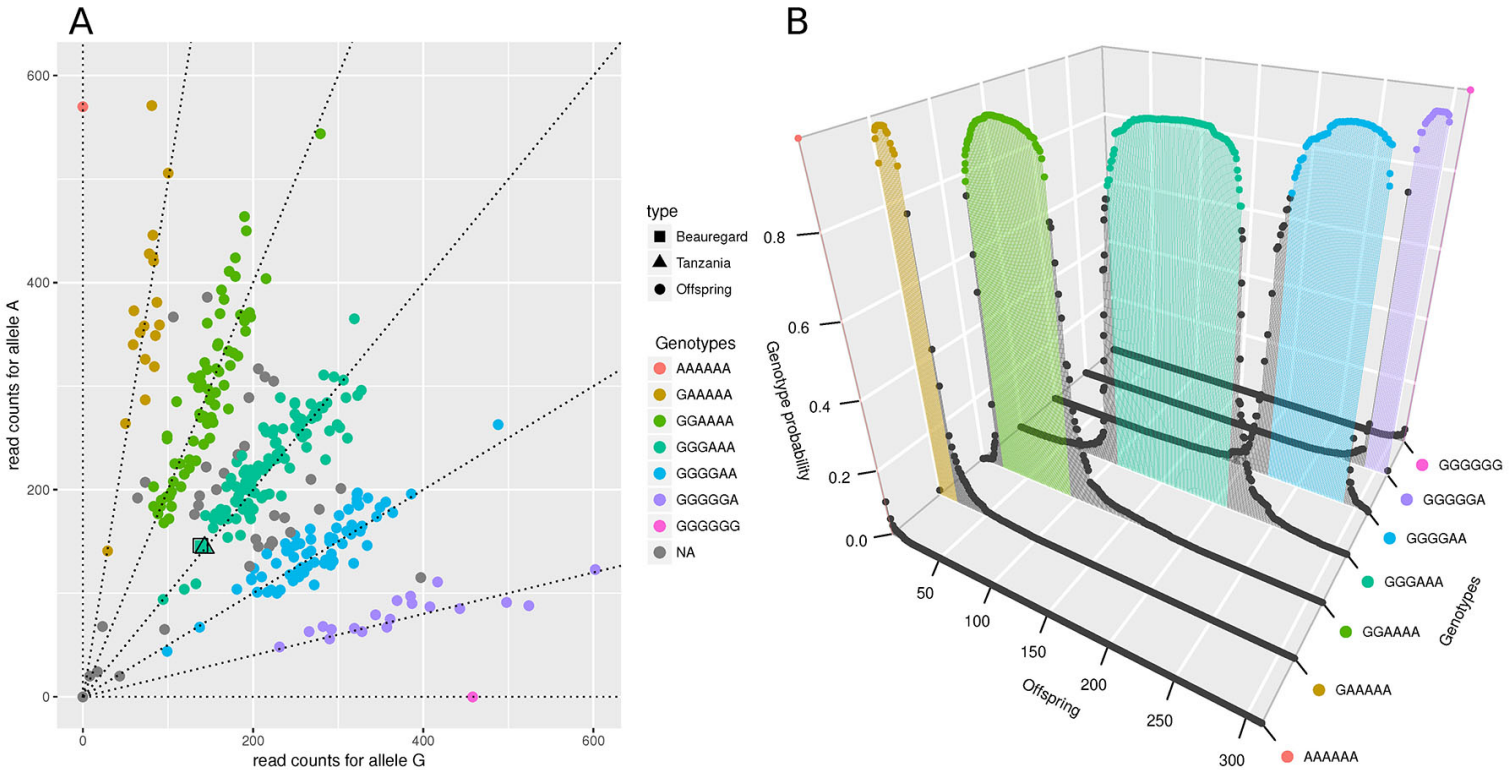
Genotype calling in polyploids

- fitTetra (tetraploids):
  `https://www.wur.nl/en/show/Software-fitTetra.htm`
- SuperMASSA (any ploidy level):
  `https://bitbucket.org/orserang/supermassa`
- updog (any ploidy level, allows preferential pairing):
  `https://github.com/dcgerard/updog`
- polyRAD (any ploidy level, reads VCF, BAM, etc):
  `https://github.com/lvclark/polyRAD`

Unraveling the hexaploid sweetpotato inheritance using ultra-dense multilocus mapping

Segregation in double triplex SNPs

Beauregard × Tanzania

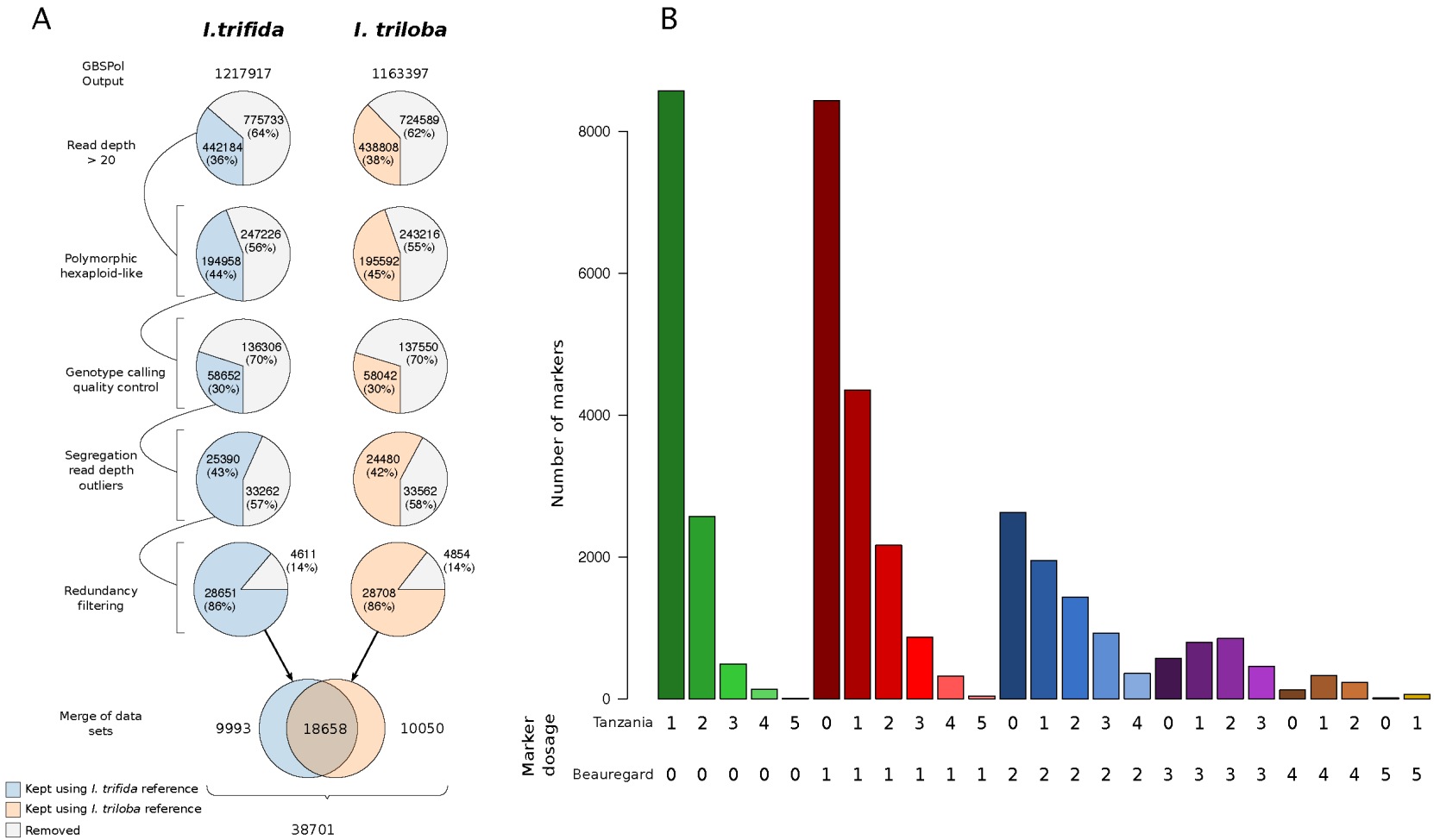


Example of genotype call using SuperMASSA.

- A Scatter plot of the read counts for the two allelic variants A and G. Segregation 1:18:99:164:99:18:1.
- B Probability distribution of genotypes for each individual.

Filtering

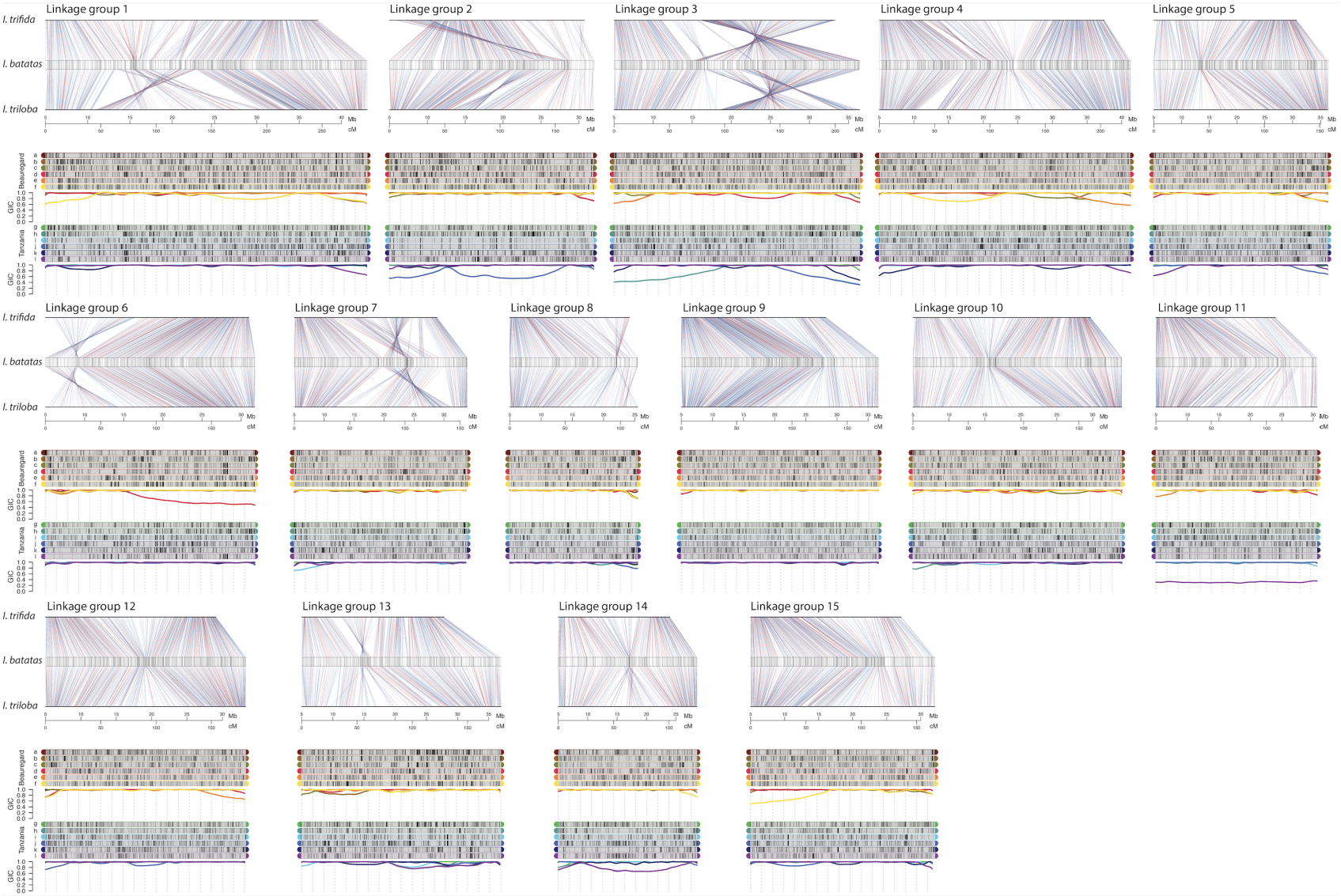
Beauregard × Tanzania



SNP filtering process and resulting dosage distribution for Beauregard × Tanazania population.

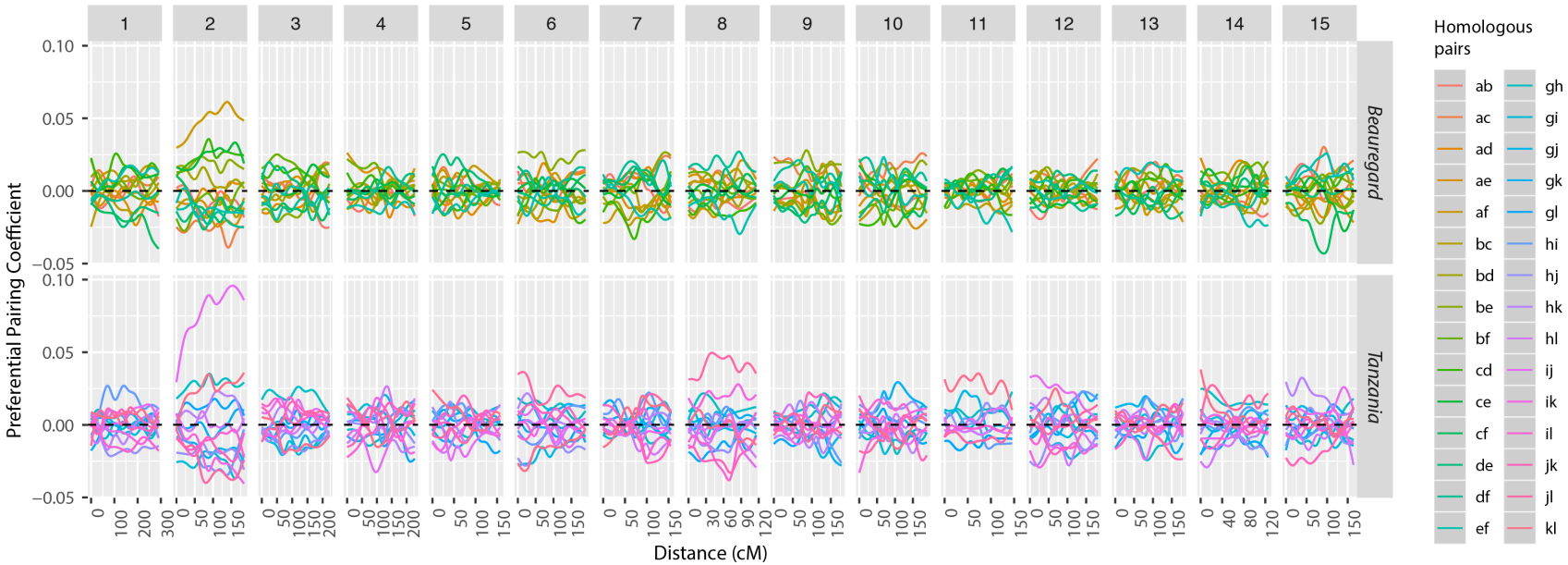
Sweetpotato genetic map

Beauregard × Tanzania



Preferential pairing profiles

Beauregard × Tanzania



Examples of multivalent formation

Beauregard