Meiotic recombination analyses in pigs carrying different balanced structural chromosomal rearrangements

Nicolas Mary, Harmonie Barasc, Stéphane Ferchaud, Aurélia Priet, Anne Calgaro, Anne-Marie Loustau-Dudez, Nathalie Bonnet, Martine M. Yerle, Alain Ducos, Alain Pinton

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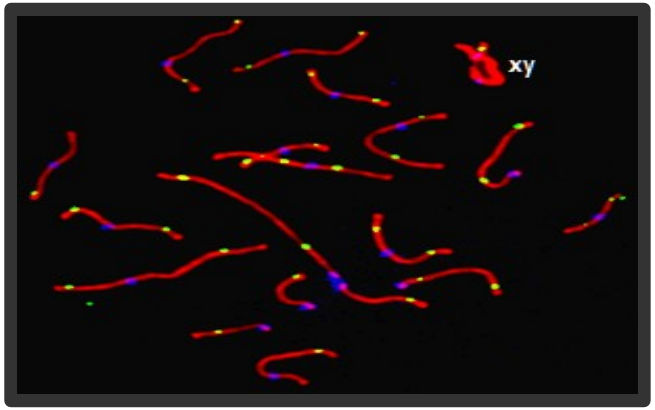
Nicolas Mary1*, Harmonie Barasc1, Stéphane Ferchaud2, Anne Calgaro1, Anne-Marie Loustau1, Nathalie Bonnet1, Martine Yerle1, Alain Ducos1, Alain Pinton1



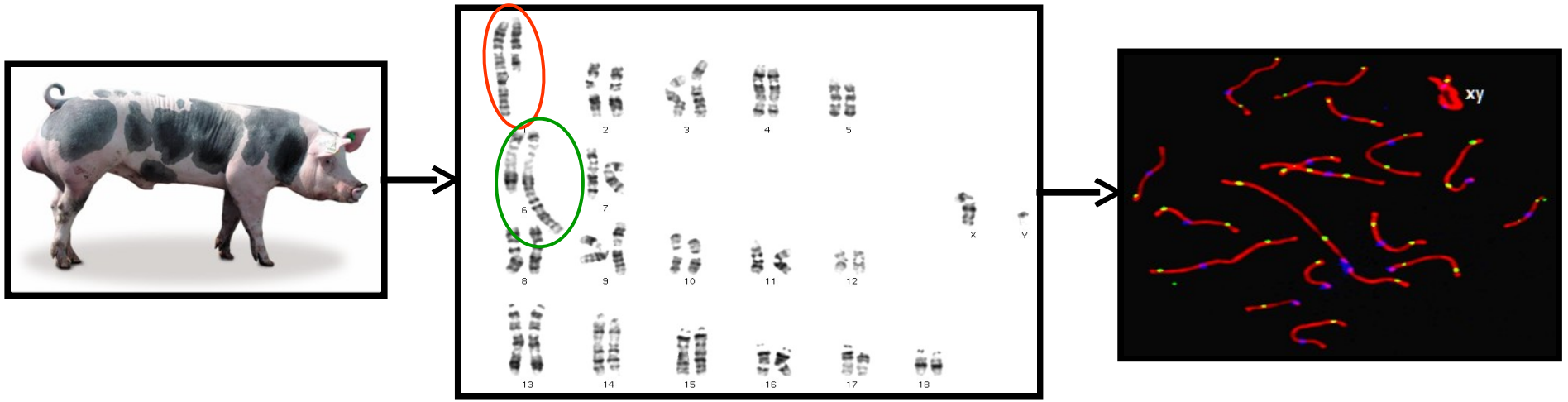
1. **INRA-ENVT GenPhySE, Toulouse, France**
2. **INRA GenESI, Surgères, France**

Previous study :

- Meiotic recombination study on normal boars :
  - Published and presented in 2014 (Mary *et al.* 2014-PlosOne)

Present study :

- Effects of chromosomal abnormalities on meiotic recombination :

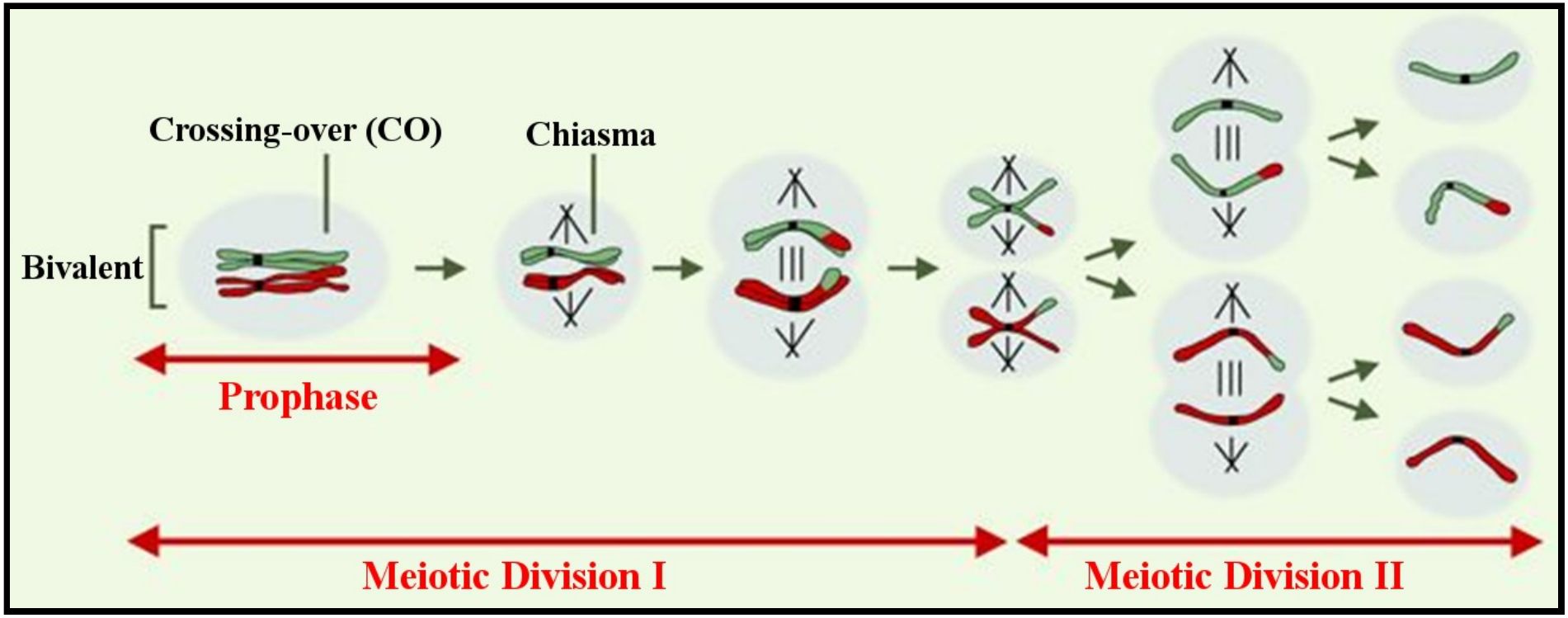


- Number of crossing-over (CO).

- Distribution of CO along the chromosomes.

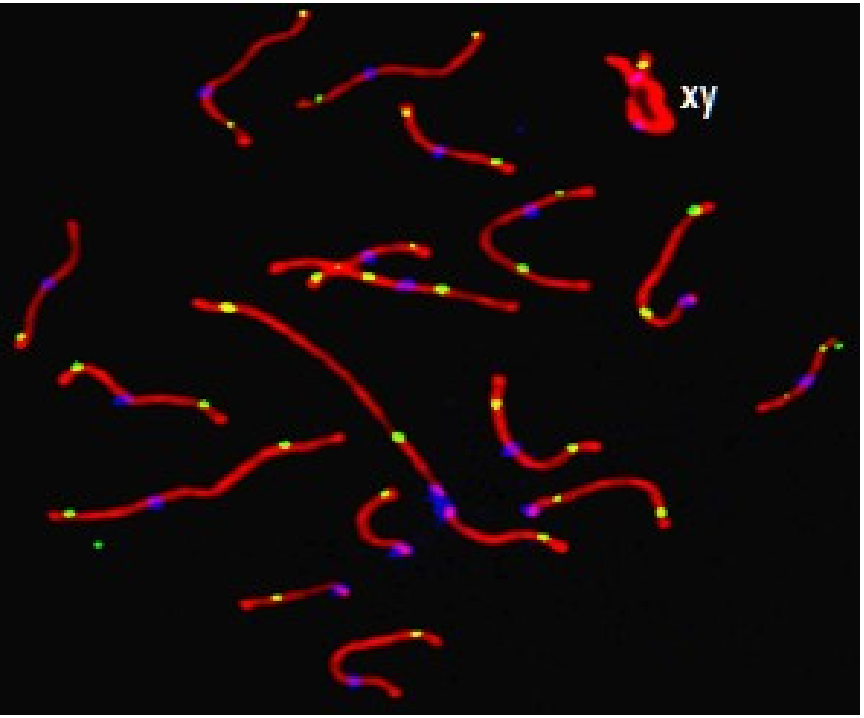
Introduction

- **Prophase of the first meiotic division :**



Methods

- **Immunostaining :**



Red Synaptonemal complexes (SCP1-3)

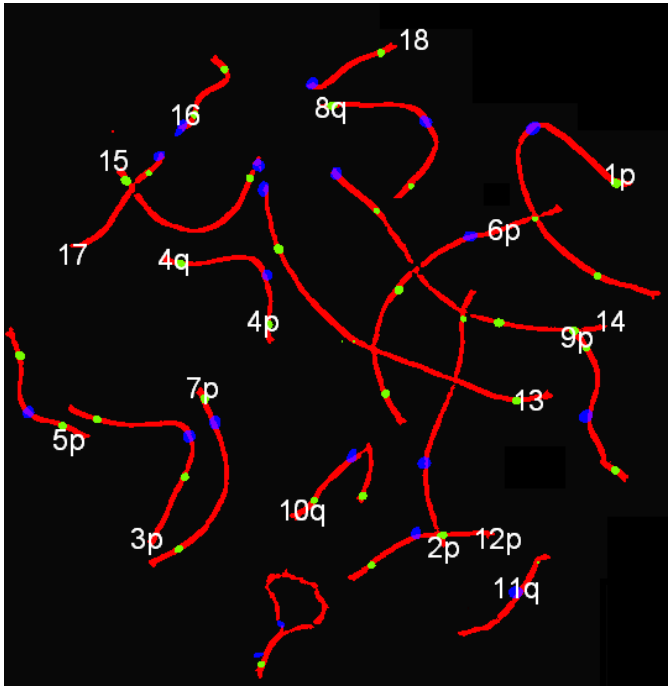
Blue Centromeres

Green Crossing-over (MLH1)

1 foci = 1 recombination event (CO)

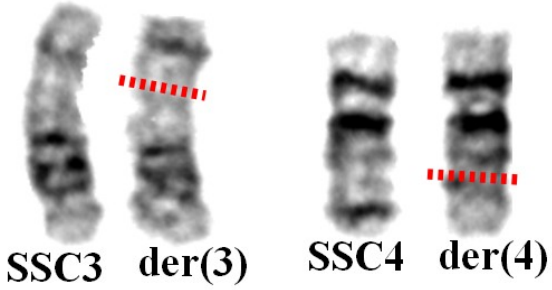
Fluorescent in situ hybridization (FISH) :

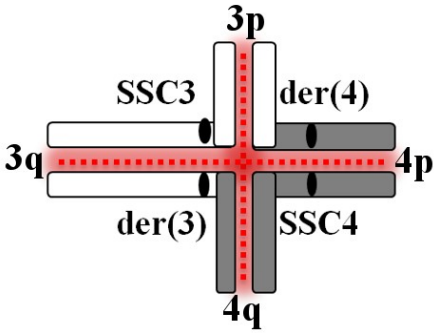
-BAC probes

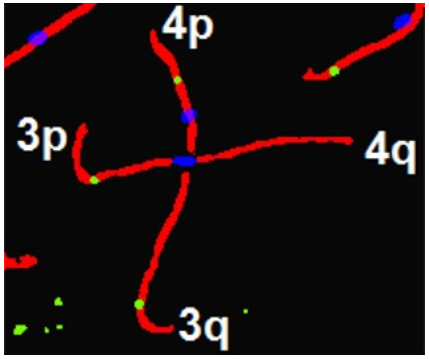


Results

- Boar T34he, <u>heterozygous</u> for the t(3;4) reciprocal translocation.

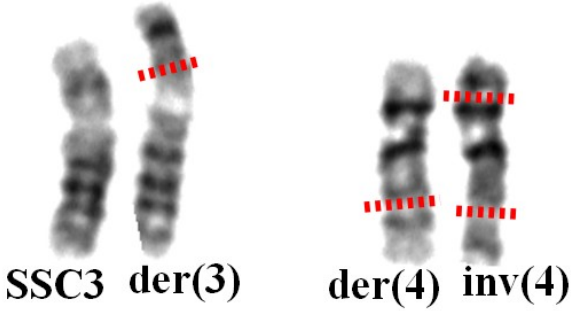


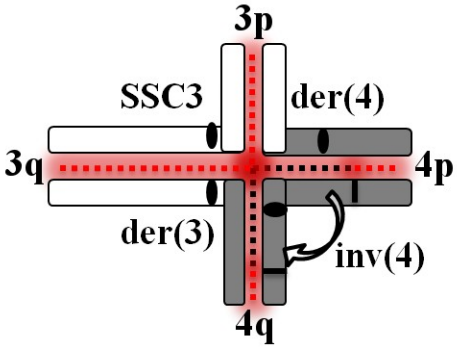


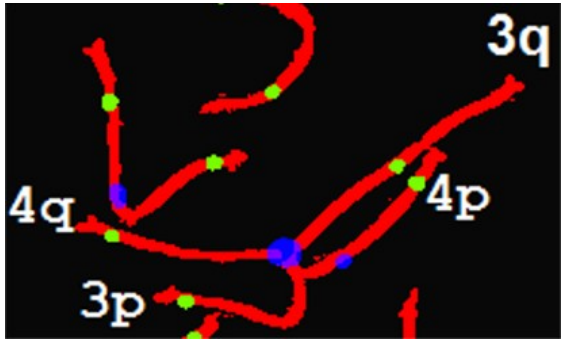


- Quadrivalent formation allowing synapsis between the homologous regions of the different chromosomes.

- Boar T34Inv, <u>heterozygous</u> for the t(3;4) reciprocal translocation and for the inversion inv(4).



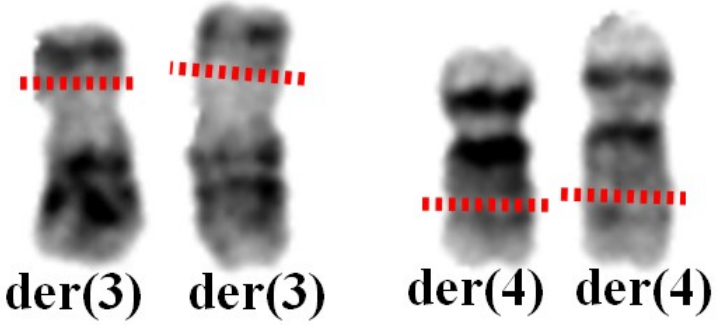


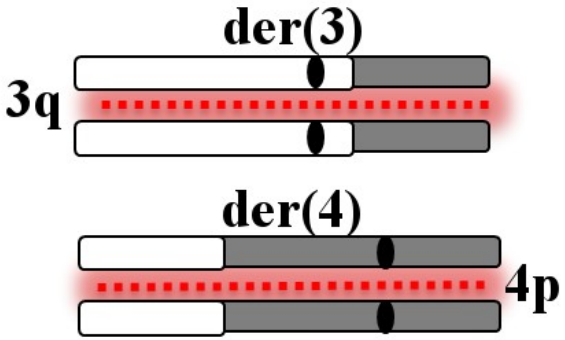


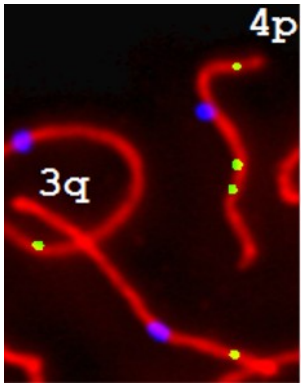
- Synapsis for the chromatin from chromosome 4 is :
  - -homologous on the telomeric parts,
  - -heterologous on the inverted parts.

Results

- Boar T34Inv, homozygous for the t(3;4) reciprocal translocation.





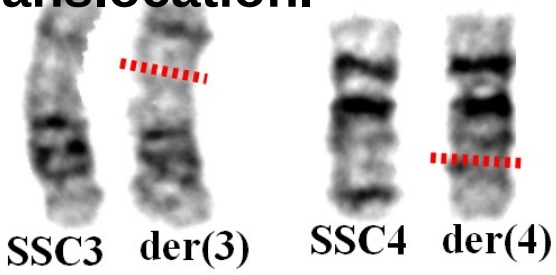


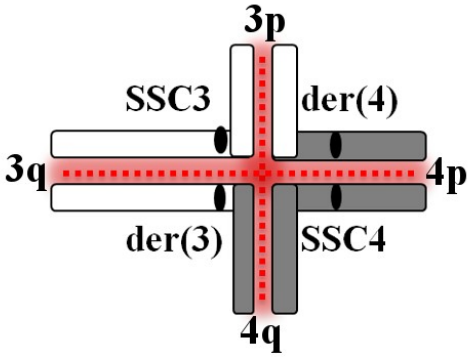
- ✓ Homologous synapsis between the same derivatives chromosomes (neo-chromosome).

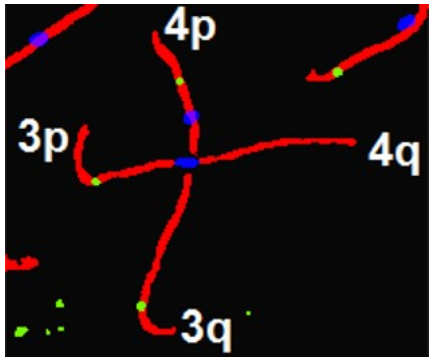
- **Recombination rate :**
  - ✓ No significant change in the mean number of CO on chromosome 3 and 4.

Results

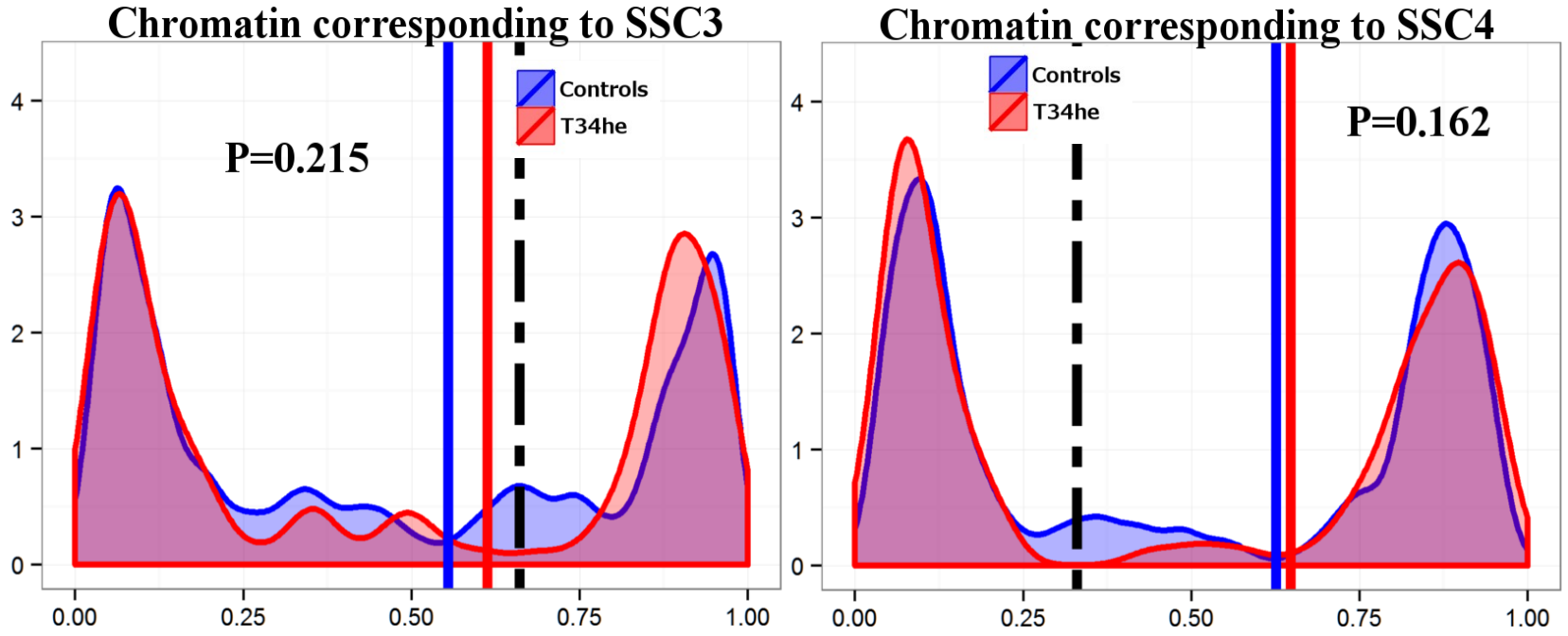
- **Boar T34he, heterozygous for the t(3;4) reciprocal translocation.**





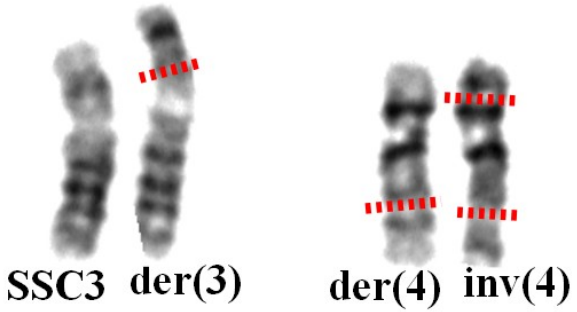


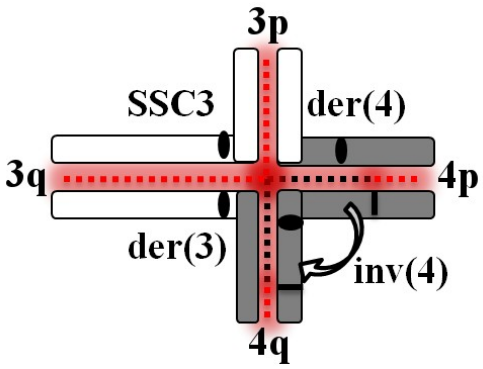
- **No significant change on CO distribution :**

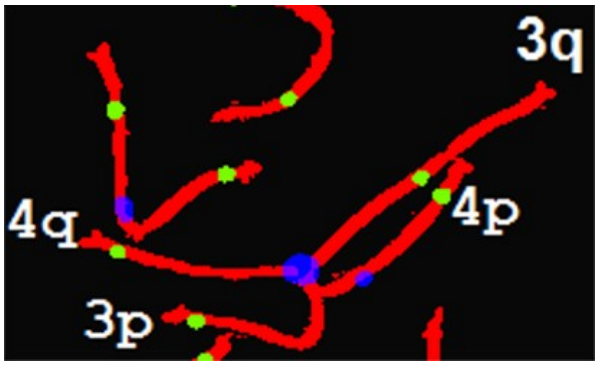


Results

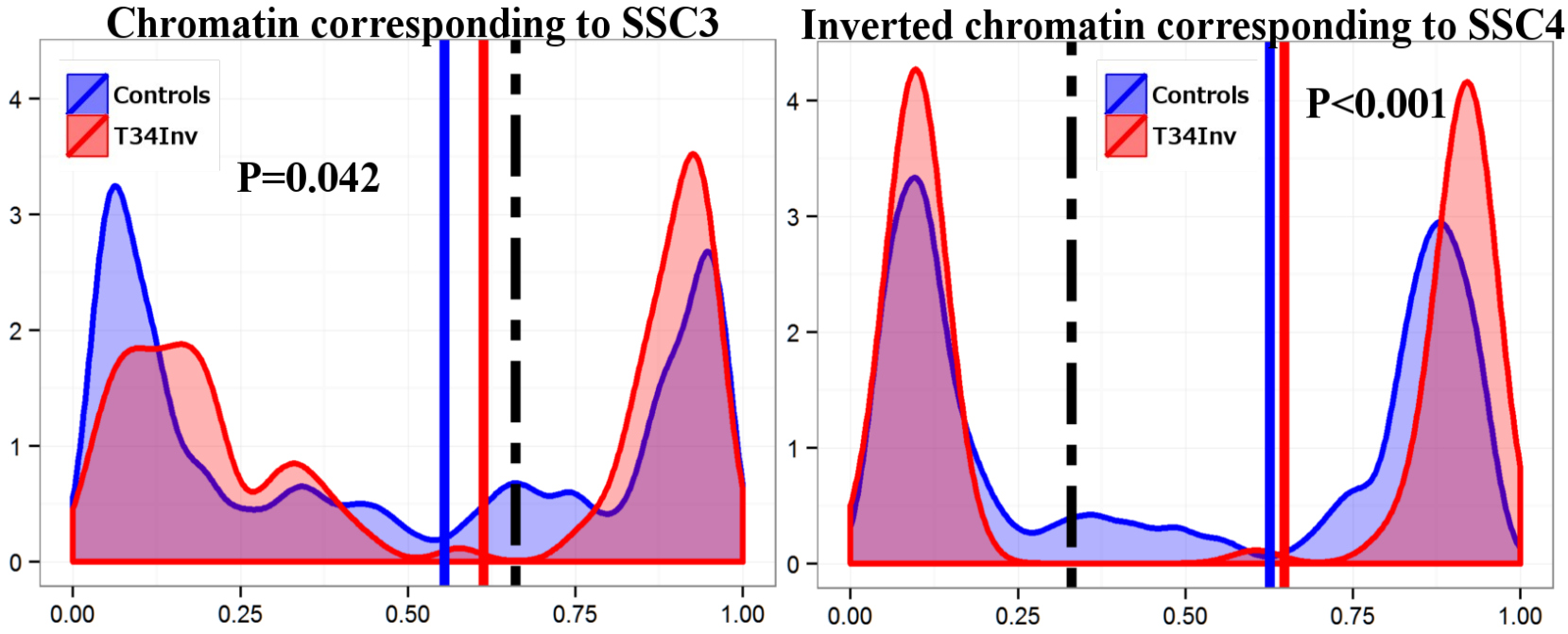
- **Boar T34Inv, heterozygous for the t(3;4) reciprocal translocation and for the inversion inv(4).**





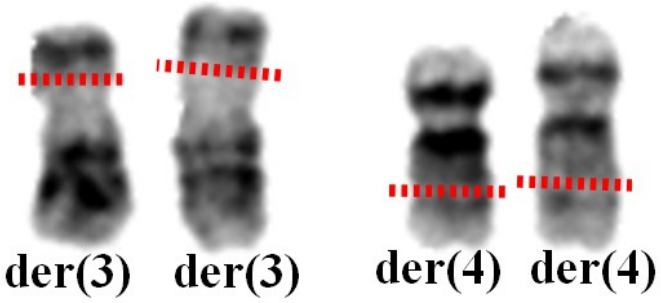


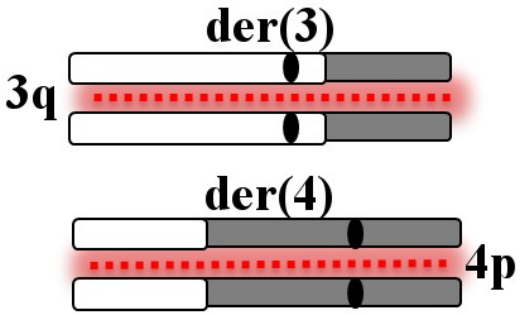
- **Significant change on CO distribution :**

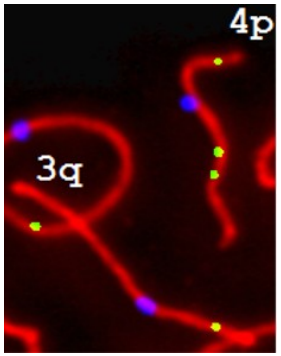


Results

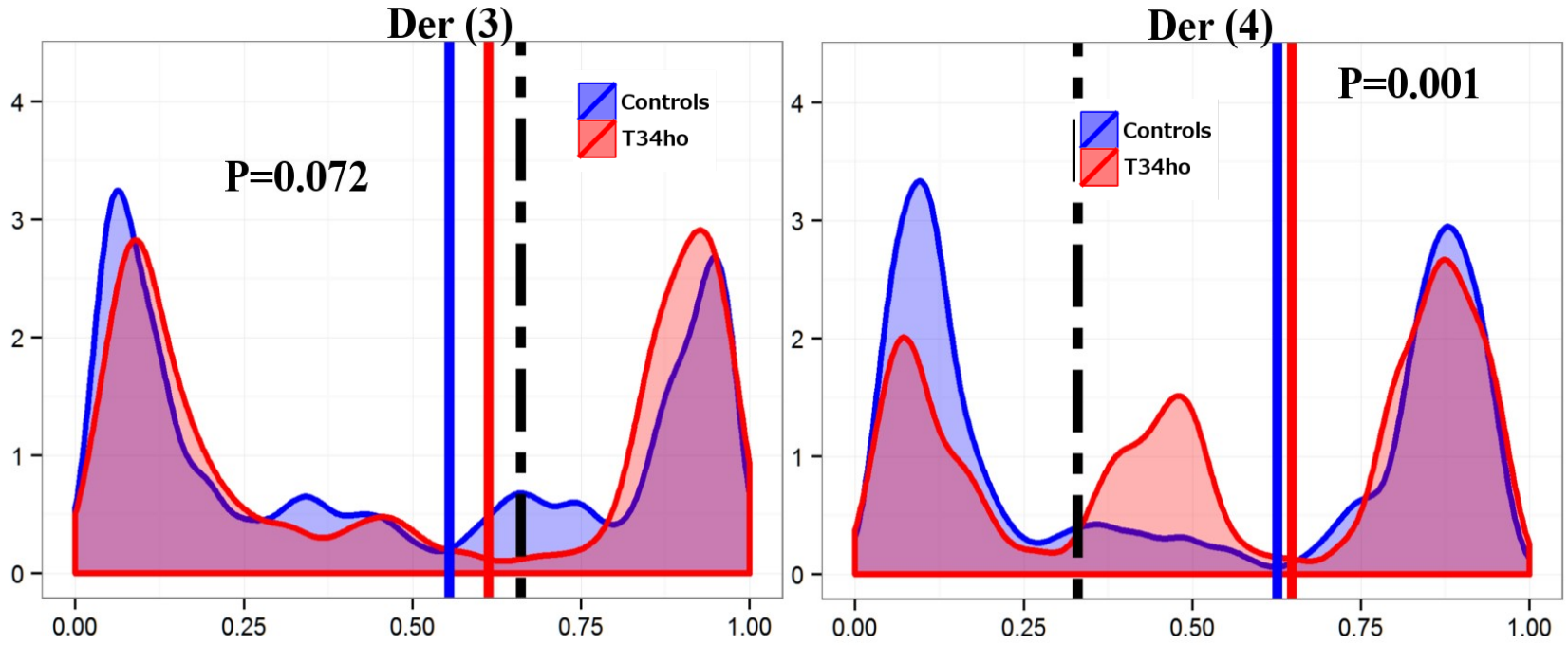
- **Boar T34ho, homozygous for the t(3;4) reciprocal translocation.**







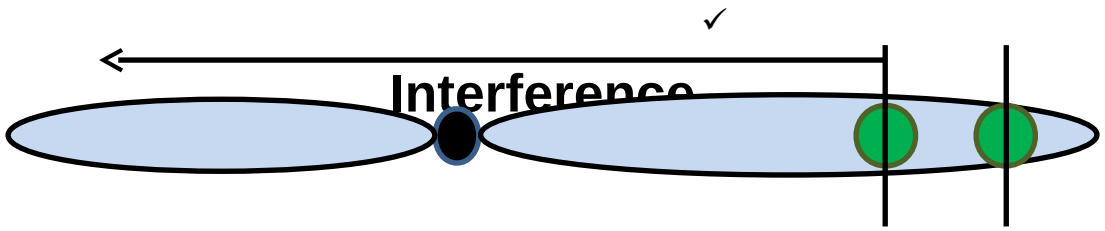
- **Significant change on CO distribution :**



Digression

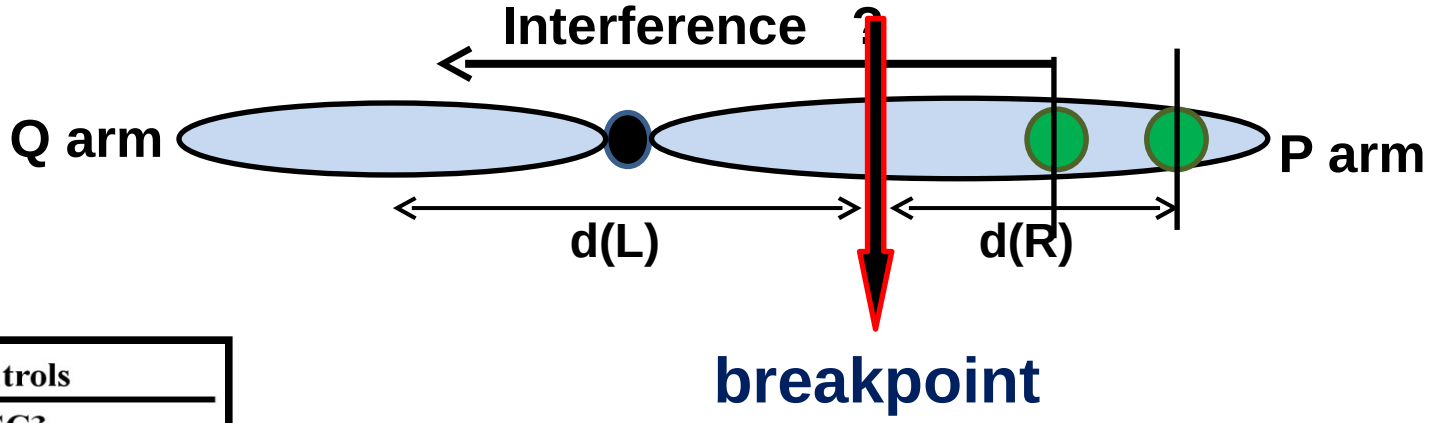
- **The interference** :

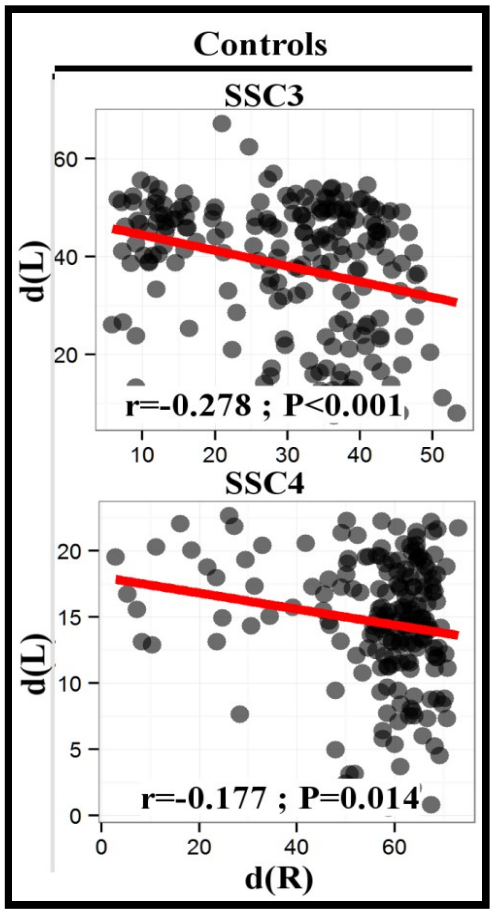
« Positive CO interference, by definition, means that the occurrence of one C
liscourages the formation of other COs in its vicinity » (Lian *et al.*, 2008)



- **Interference signals can act on long distances,**
- **Interference signals can cross the centromere,**
- **But we do not know if the signal is propagated along :**
  - **-the chromatin,**
  - **-the synaptonemal complex,**
  - **-or both.**

reakpoint, barriers to interference ?

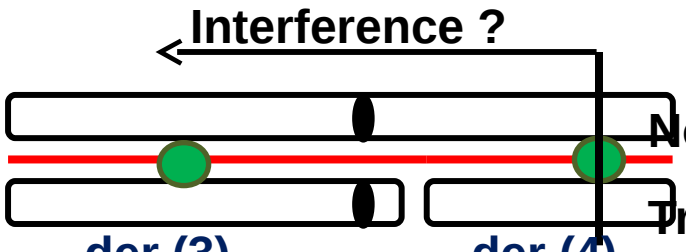




✓**There is an interference signal which cross this region (where the breakpoint will be on translocated chromosomes)**

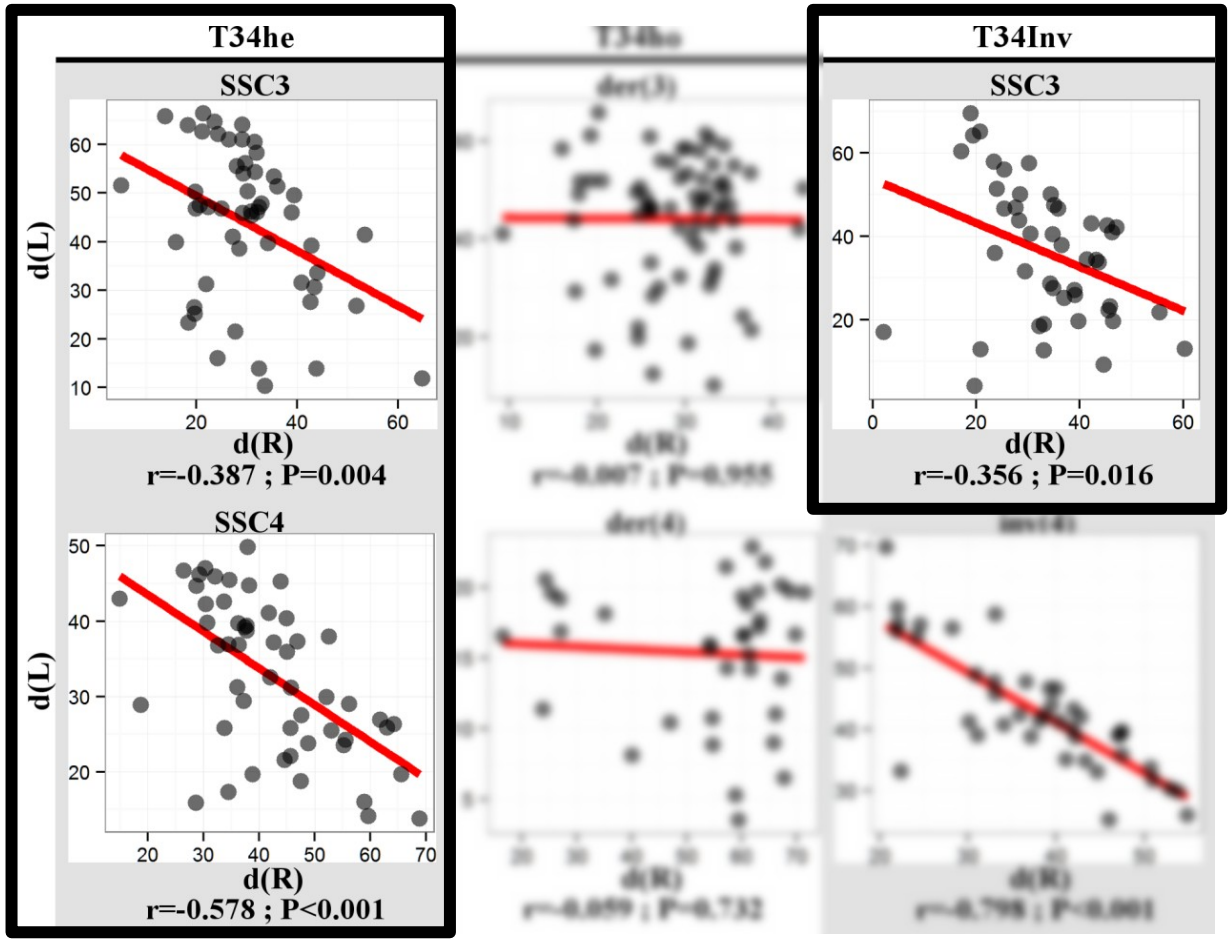
Results

Heterozygous



Normal chromosome 3

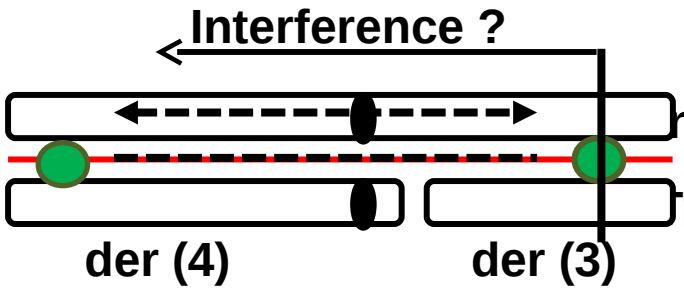
Translocated chromosomes



- Interference signal could cross the breakpoint.

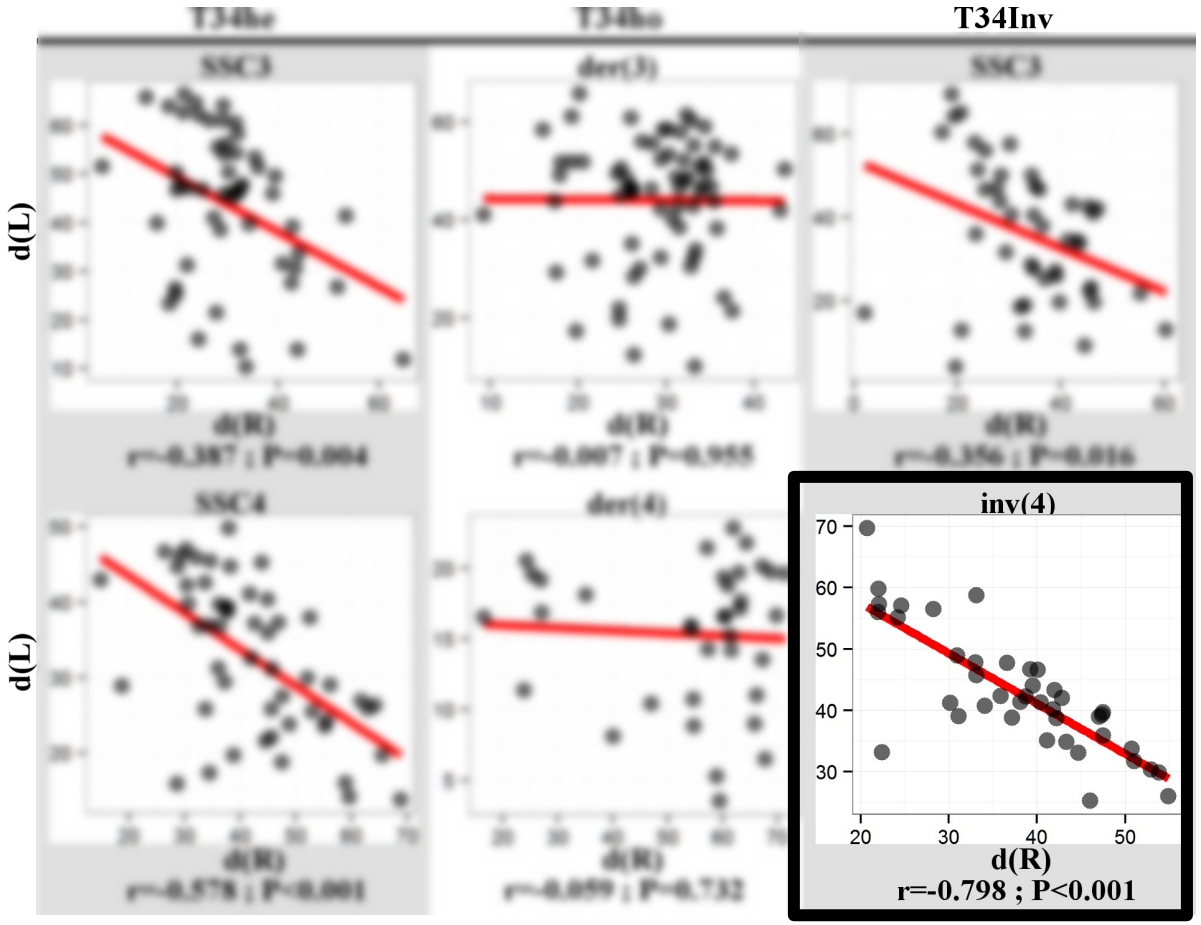
Results

Heterozygous



nverted chromosome: Inv (4)

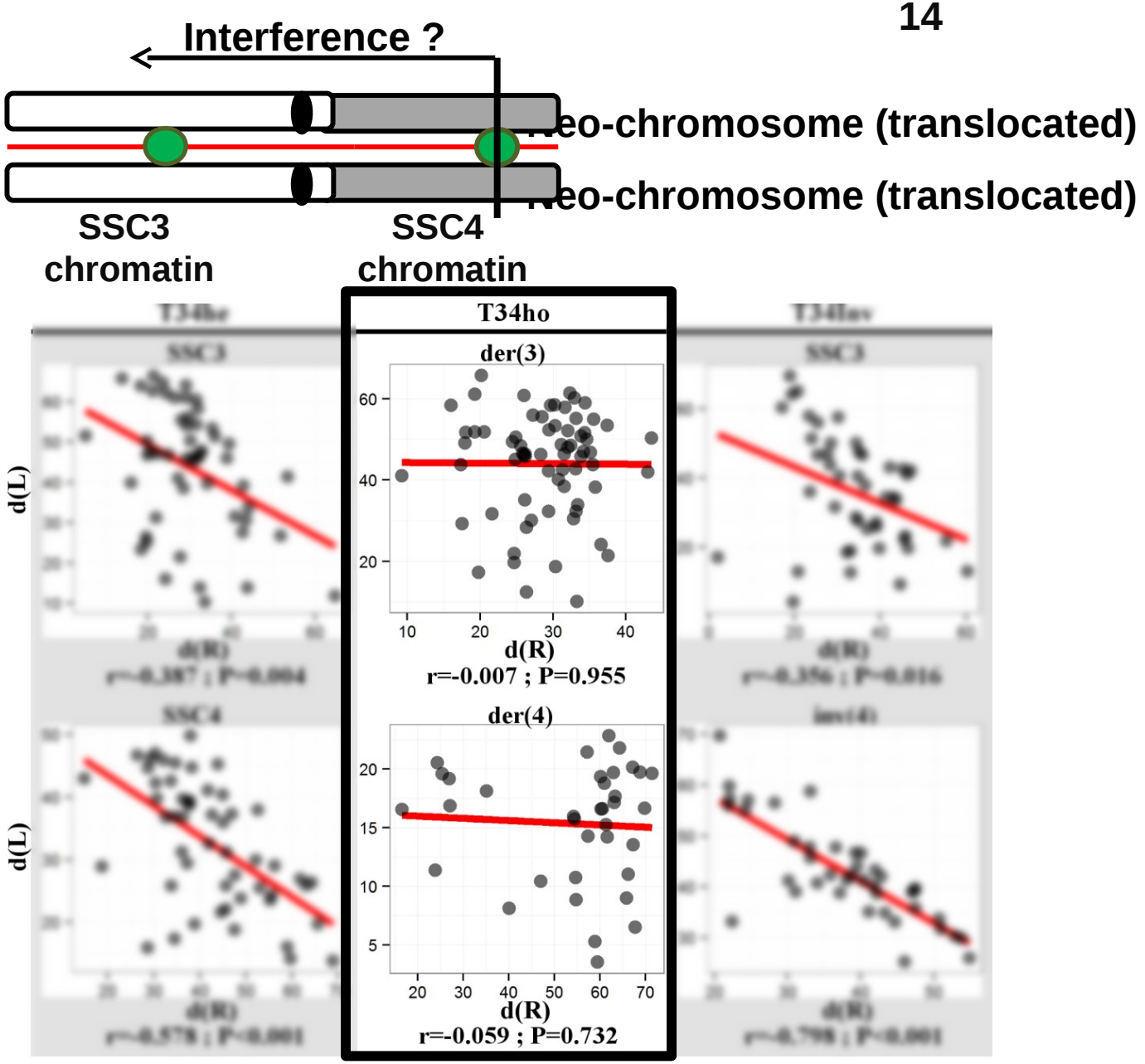
ranslocated chromosomes



- **Interference signal could cross the breakpoint.**

Results

Homozygous



- Interference signal seems to be stopped by the breakpoint.

Conclusion :

- First report on meiotic recombination in boars :
  - with two chromosomal abnormalities (translocation and inversion),
  - homozygous for a reciprocal translocation.
- No synaptic defects.
- No effect on CO rate.
- Significant change on CO distribution (interference).

✓ Normal SC is sufficient to regulate CO rate but not sufficient to propagate normal interference.

✓ Chromatin information (DNA sequence, methylation…) is important to propagate interference.

Conclusion :

- **More details and analyses on Mary *et al.* april 2016 - PlosOne**

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1 INRA, UMR1388 Génétique, Physiologie et Systèmes d'Elevage, Castanet-Tolosan, France, **2** Université de Toulouse INPT ENSAT, UMR1388 Génétique, Physiologie et Systèmes d'Elevage, Castanet-Tolosan, France, **3** Université de Toulouse INPT ENVT, UMR1388 Génétique, Physiologie et Systèmes d'Elevage, Toulouse, France, **4** UE1372 GenESI Génétique, Expérimentation et Système Innovants, Surgères, France

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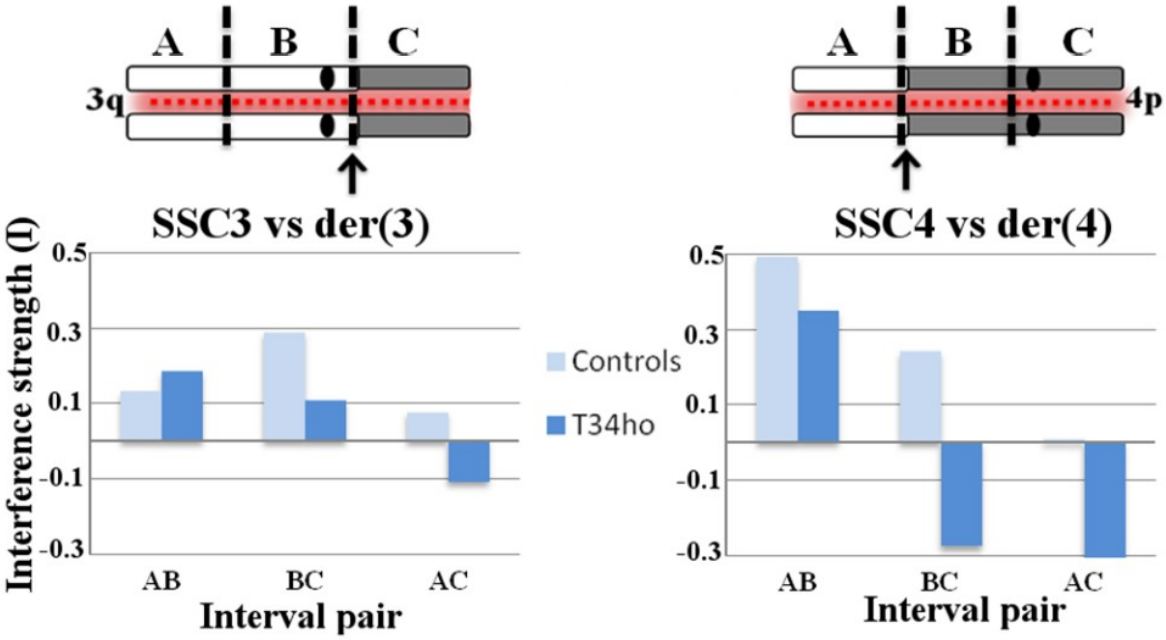
thank you

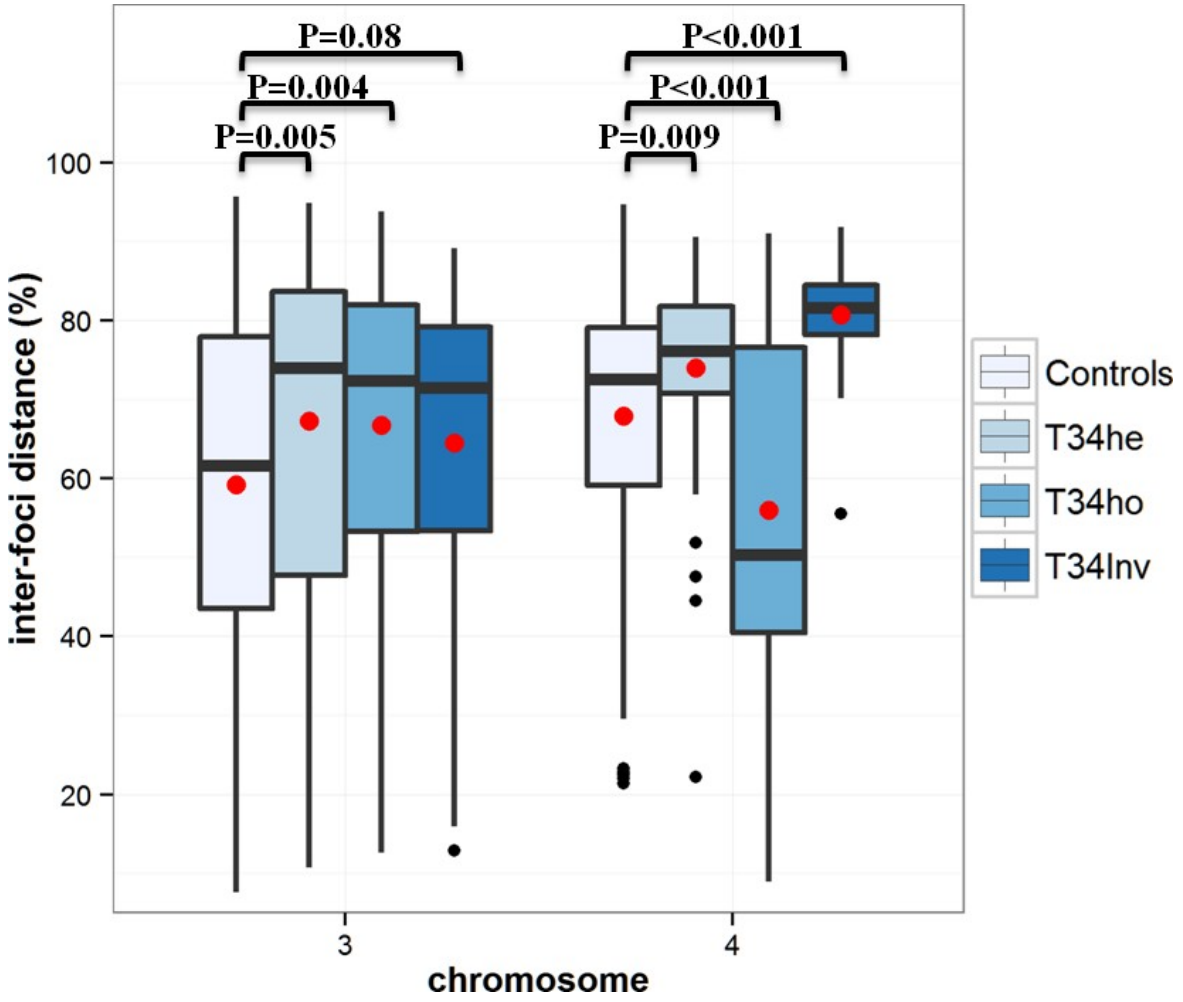
Results

Coefficient of coincidence analysis



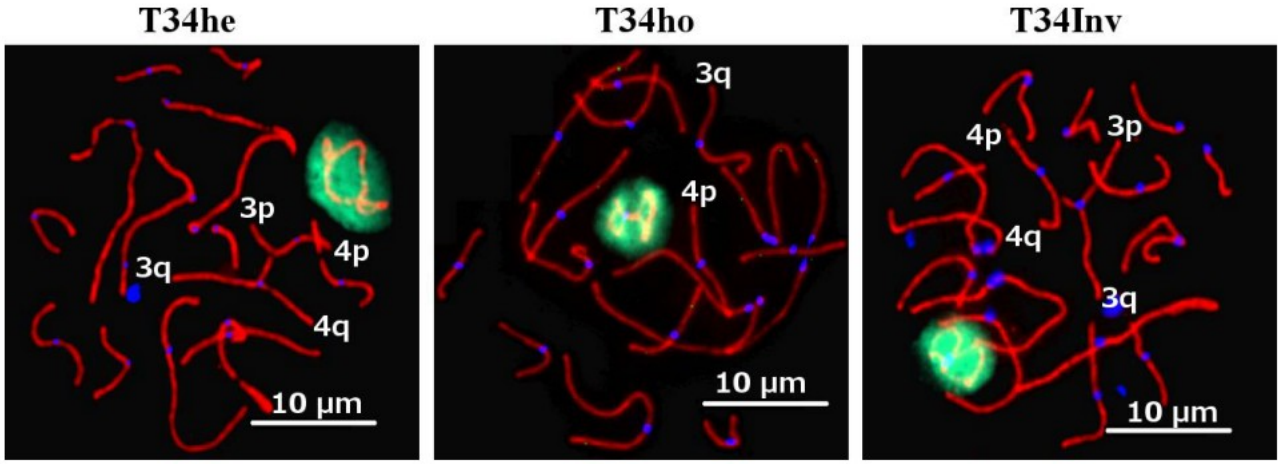
Results

- **Distances between CO (in % of synaptonemal complex length)**

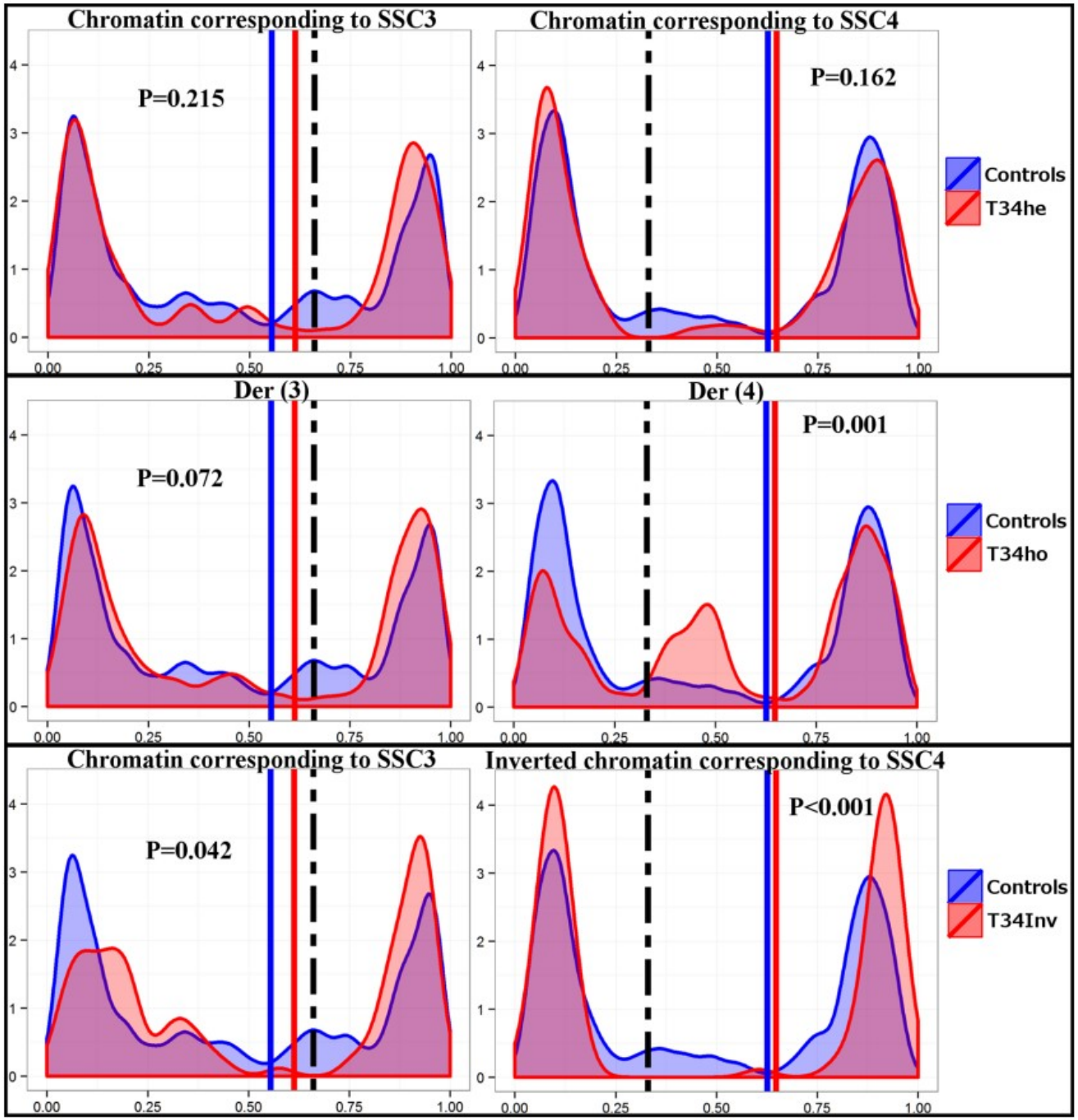


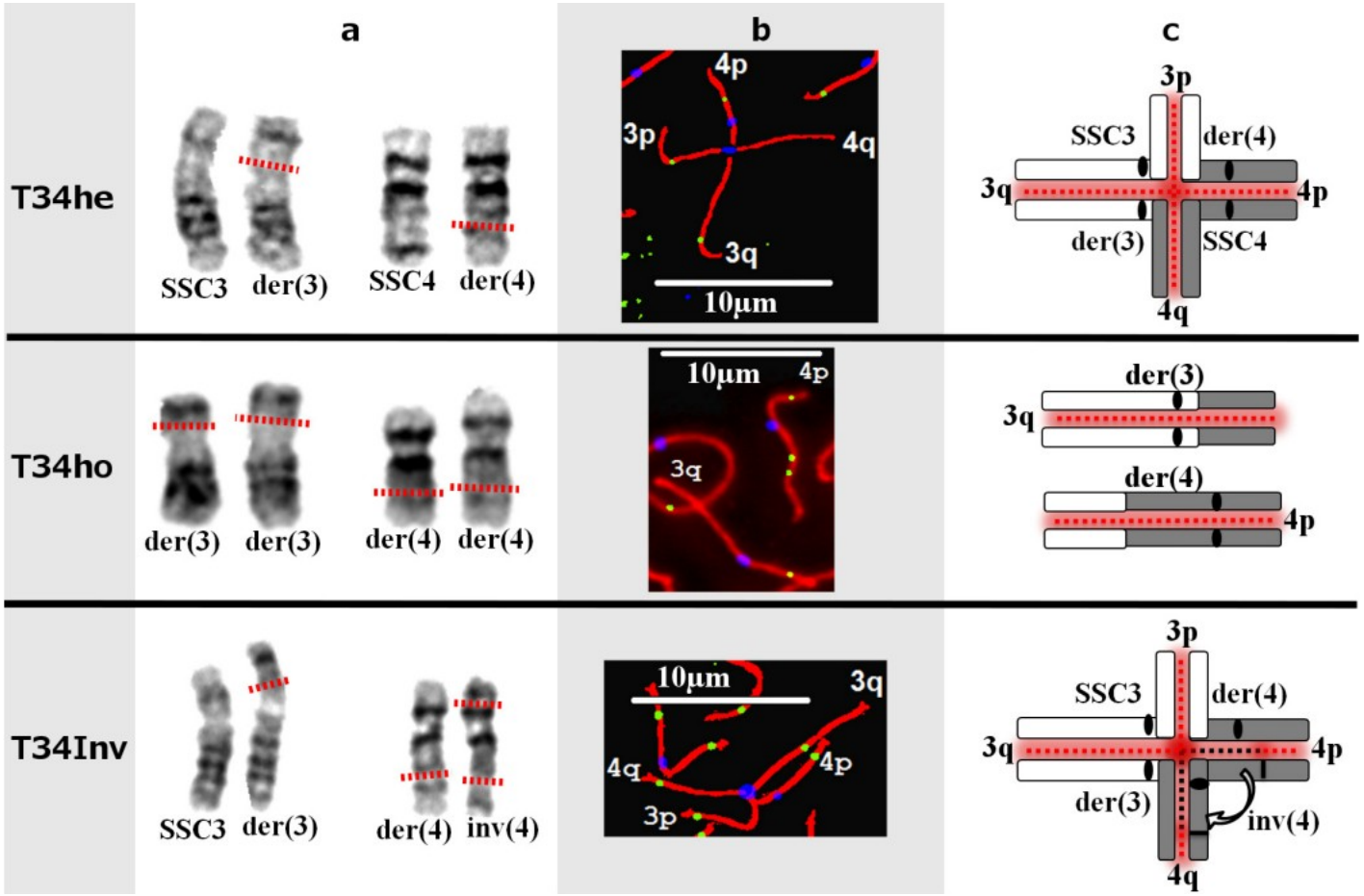
- **Chromosomal abnormalities disturb crossing-over position.**

- **Meiotic pairing analysis of pachytene cells.**



- **No γH2AX-positive region was observed, except on the XY-body**


Chromatin corresponding to SSC3
P=0.215
Chromatin corresponding to SSC4
P=0.162
Controls
T34he
Der (3)
P=0.072
Der (4)
P=0.001
Controls
T34ho
Chromatin corresponding to SSC3
P=0.042
Inverted chromatin corresponding to SSC4
P<0.001
Controls
T34Inv


a
b
c
T34he
SSC3
der(3)
SSC4
der(4)
4p
3p
4q
3q
10µm
3p
SSC3
der(4)
3q
4p
der(3)
SSC4
4q
T34ho
der(3)
der(3)
der(4)
der(4)
10µm
4p
3q
der(3)
3q
der(4)
4p
T34Inv
SSC3
der(3)
der(4)
inv(4)
10µm
3q
4q
4p
3p
3p
SSC3
der(4)
3q
4p
der(3)
inv(4)
4q