

# ROSBREED

Pedigree Based Analysis

using **FlexQTL™** software

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# Outline

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- ✓ Current Status:
  - ✓ QTL mapping in pedigreed populations
  - ✓ IBD probability of founder alleles
  - ✓ Marker consistency checking
- ✓ Targets for RosBREED project
- ✓ Questions & discussion



# FlexQTL software - 2 modules + ....

## Quantitative trait analysis (QTL mapping)

- Pedigree, trait phenotypes, markers genotypes, linkage map
- Dissection of quantitative traits into non-genetic and genetic components

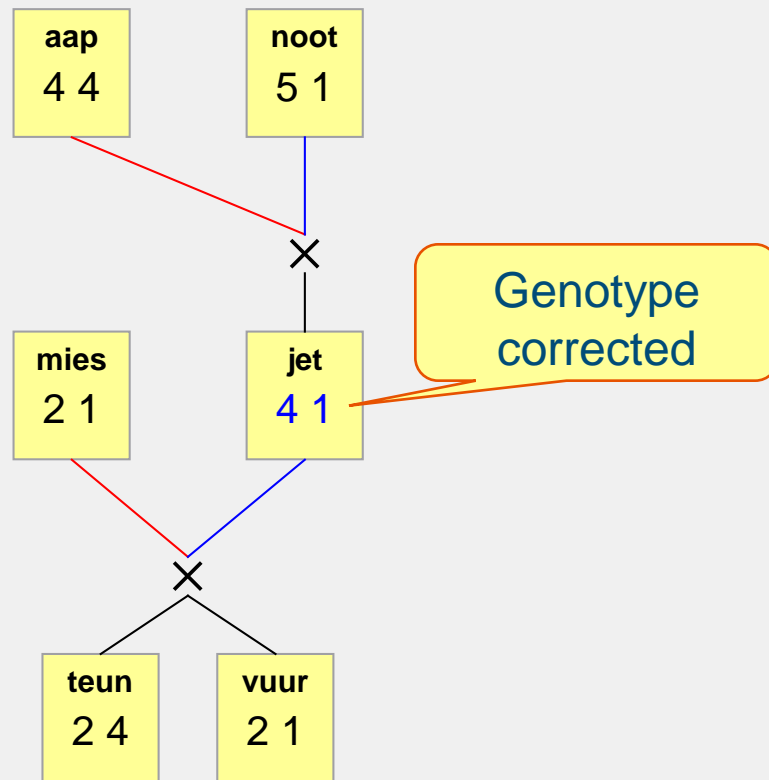
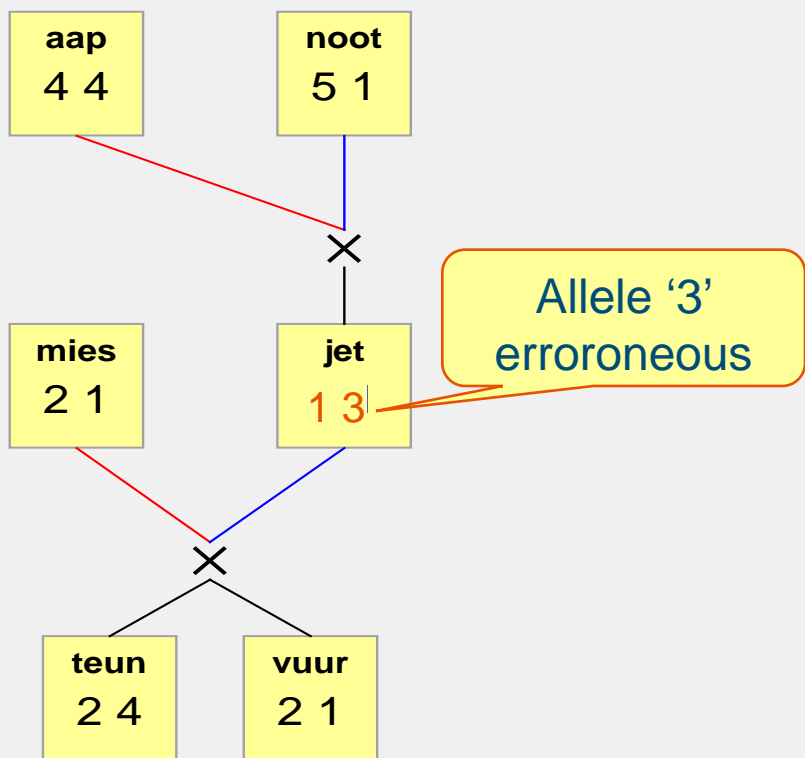
## ■ Identity By Descent (IBD) probability

- Pedigree, marker genotypes, linkage map
- Founder alleles
  - Visualization via Pedimap software
  - Regression analyses , e.g., fitting multiple alleles or interactions
  - Founder allele representation

## ■ Marker data checking

- Pedigree, marker genotypes, linkage map
- Consistency & informativeness

# Example on segregation errors



# Example - Reporting marker consistency

```
LocusName marker001 (1)
  1 2 3 4 5 ;alleles coded in FlexQTL
  1 2 3 4 5 ;alleles original in data
```

```
jet marker001 1,3 1,3
  1 3 0.0000 (<-)
  4 1 0.9999
```

```
Putative deletion: -log(p)
                   jet marker001 99.31
```

```
Realised deletion:
                   jet marker001 99.31 1 3
```

## Augmentation step:

```
jet marker001 1,3 4,1
  4 1 0.9999 (<-)
```

```
Detected segregation errors: 0
Augmented marker genotypes : 1
```

# MARKER GENOTYPE ERRORS

```
*****
Indicator : FlexQTL-code : Explanation
.          : 0          : (OC)  observed & consistent
^          : 2          : (OpE) observed & putative error
!          : 1          : (OdE) observed & deleted error
#          : 5          : (OdEA)observed & deleted error & augmented
/          : 8          : (ME)  missing & error
+          : 4          : (MA)  missing & augmented
_          : 9          : (M)   missing

|          :           : next linkage group
*****
```

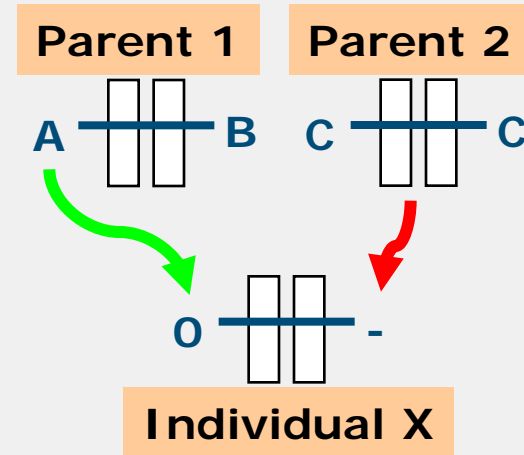
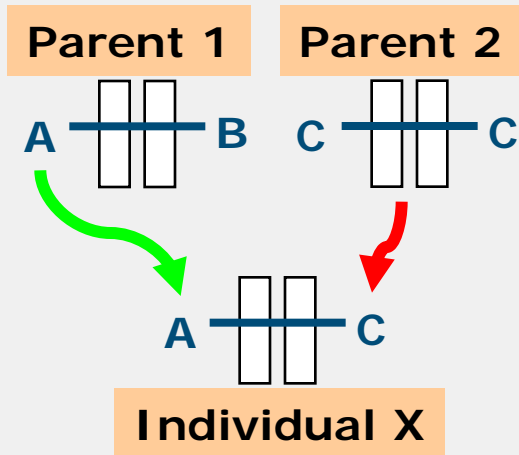
## MARKER GENOTYPE PATTERN

```
|..... ; aap
|..... ; noot
|..... ; mies
|..... ; wim
|..... ; zus
|..... ; jet
```

## MARKER GENOTYPE PATTERN

```
|..... ; aap
|....._ ; noot
|.....+ ; mies
|.....+ ; wim
|.....+ ; zus
|#.##+ ; jet
```

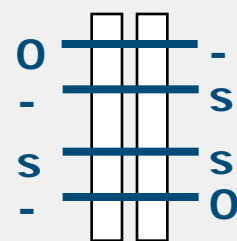
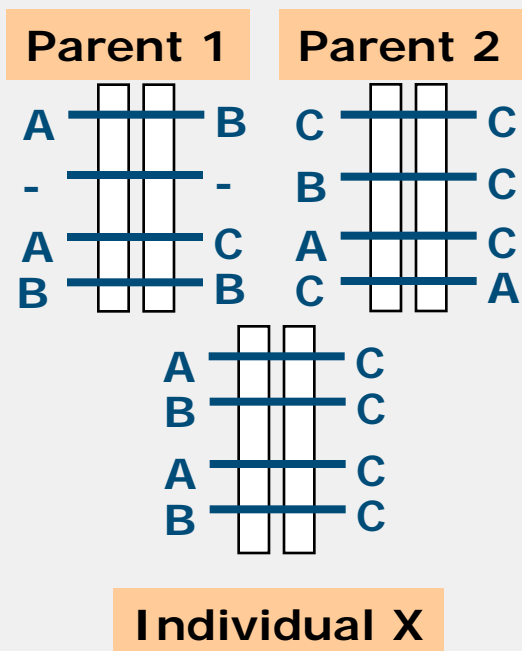
# Marker information: segregation indicators



## Used symbols & their explanation:

- \_ not available or not informative (parent homozygous genotype)
- 0 paternal allele of its parent
- 1 maternal allele of its parent
- s undetermined, i.e., similar heterozygous genotypes indiv & parent(s)

# Marker information: segregation indicators



**Individual X**

4 polymorphic markers: limited information on segregation

## Used symbols & their explanation:

- \_ not available or not informative (parent homozygous genotype)
- 0 paternal allele of its parent
- 1 maternal allele of its parent
- s undetermined, i.e., similar heterozygous genotypes indiv & parent(s)

# Segregation indicators (SI):

Used symbols & their explanation:

- \_ not available or not informative (parent homozygous genotype)
- 0 paternal allele of its parent
- 1 maternal allele of its parent
- s undetermined, i.e., similar heterozygous genotypes indiv & parent(s)

		Marker loci					
		1	2	3	4	5	6
haplotype	parent	4	4	5	5	1	5
SI values		4	5	1	4	1	1
progeny	400	_	1	1	1	_	1
	401	_	1	0	1	_	1
	402	_	0	0	0	_	
	403	_	0	s	0	_	
	404	_	1	1	1	_	
	405	_	1	s	0	_	0

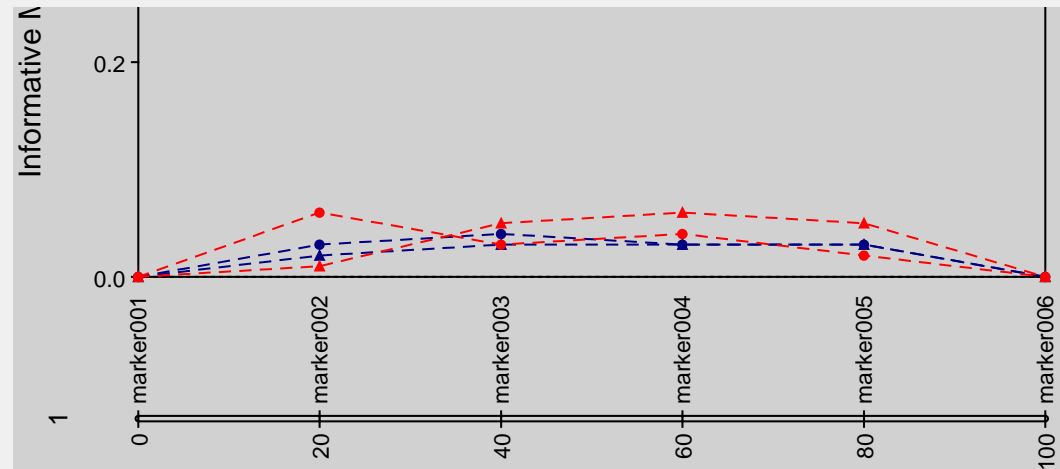
Double recombinant

# Marker consistency checking

- Consecutive triplets of informative meioses
  - Two meiosis events per non-founder individual
  
- Observed number of Double Recombinants
  - Based on observed dataset
- Expected number of Double Recombinants
  - Based on informative meiosis events
  - Based on given marker map distances

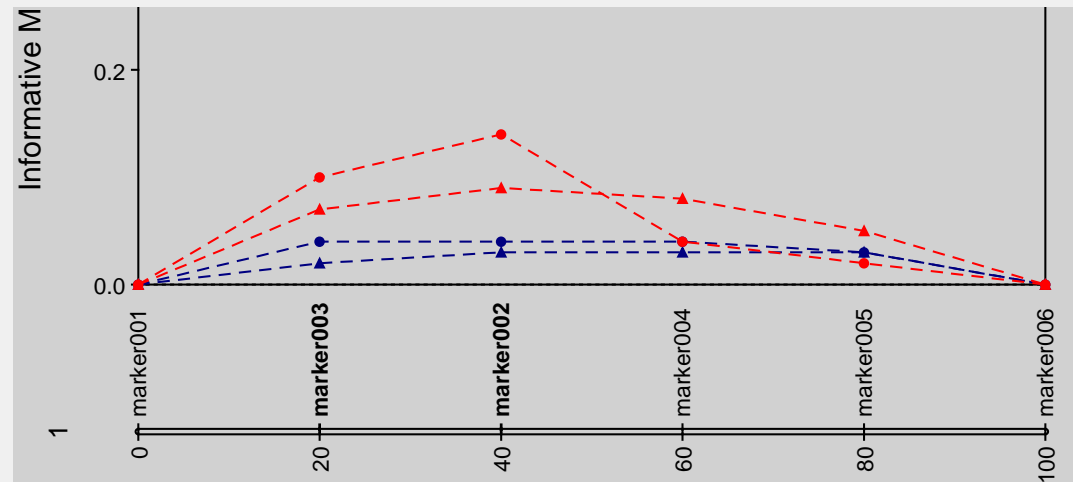
# Map order & Double Recombinants: Example

■ Correct order



■ Erroneous order

● Switched markers 2 & 3



# FlexQTL™ targets: where to go?

- *Training course materials (ASHS meeting – July'10)*
- *User friendliness of FlexQTL:*
  - *Improve current functionalities*
    - *Graphical User-Interface, e.g., window-based input*
    - *User-friendly output (easier to interpret)*
  - *Add new functionalities*
    - *Infer linkage phase of marker and QTL alleles.*
    - *Ability to deal with semi-founders*
    - *Founder (allele) representation*
- *Data exchange*
  - *Add-on tools (decision support) & Database*
- *Scientific extensions:*
  - *Polyploids (conceptually challenging; extension programming)*
  - *Marker density: High density maps & founder alleles relatedness*
  - *QTL modeling: multi-allelic & interactions (env'ment & epistasis)*