Session 18, anu.sironen@mtt.fi



#### Causes for the rapid proliferation of the immotile short tail sperm defect within the Finnish Yorkshire population

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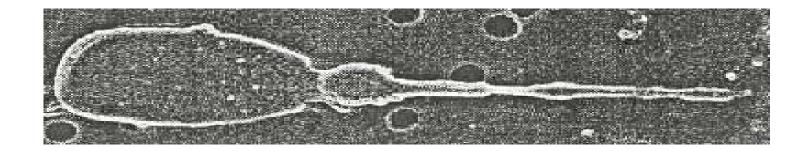
MTT Agrifood Research Finland/Animal Genomics



# The immotile short tail sperm (ISTS) phenotype

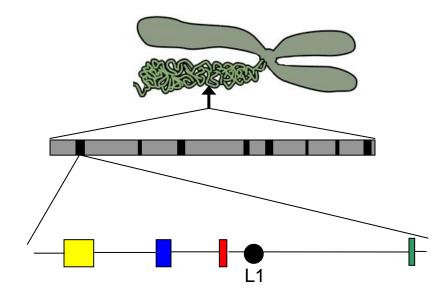
- Immotile sperm
- Shortened sperm tail
- Oligospermia
- All sperm tail structures are severely altered
- No effect on other ciliary tissues

→ only affects sperm flagella





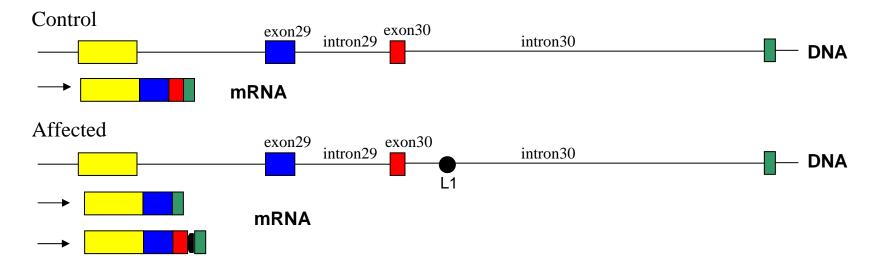
## Mapping of the ISTS mutation



- 1. Porcine chr 16
- 2. Human chr 5,
  - →8 genes
- 3. KPL2 (SPEF2) gene
- 4. L1 insertion within intron 30



## L1 affects KPL2 splicing in the testis



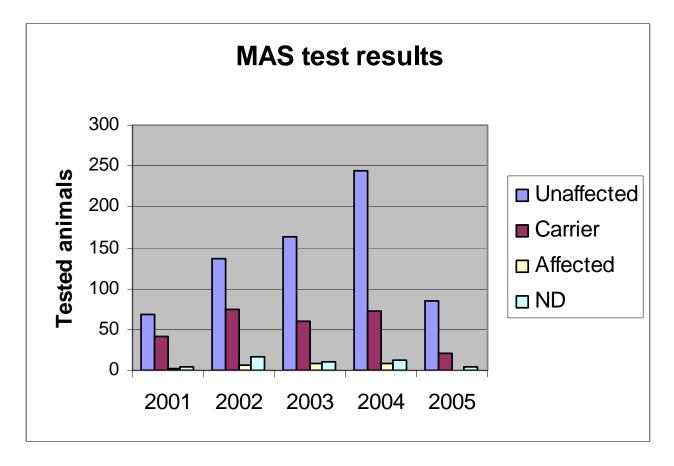
 In most affected testis specific transcripts exon 30 is skipped

•In a few cases, exon 30 is present together with part of the insertion sequence

•Translation stop codons are created in both cases



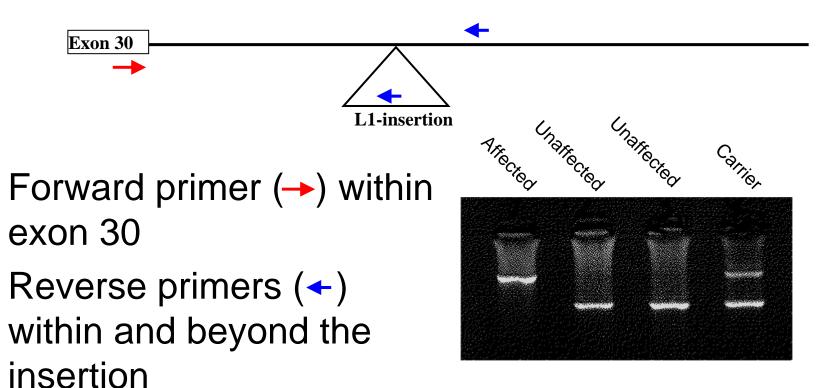
## Marker assisted selection 2001-2005



Markers: SW2411 and SW419

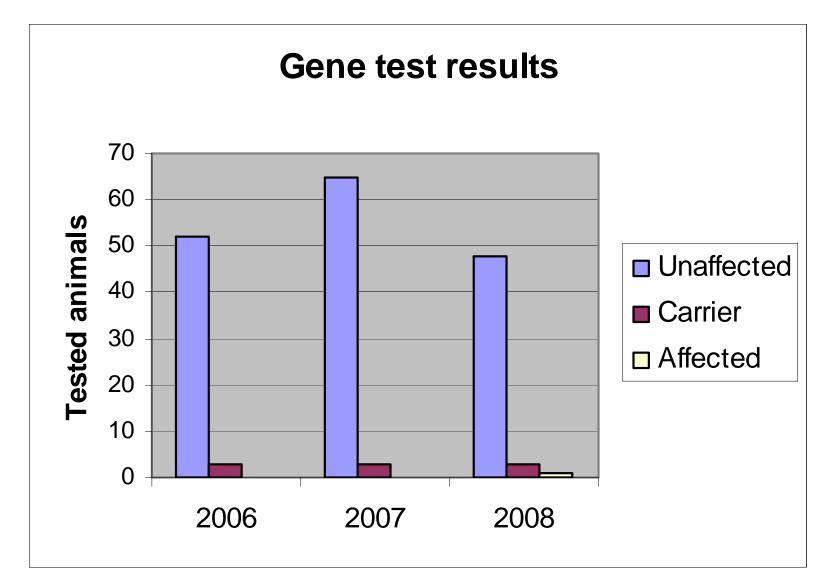


## 100% DNA-test



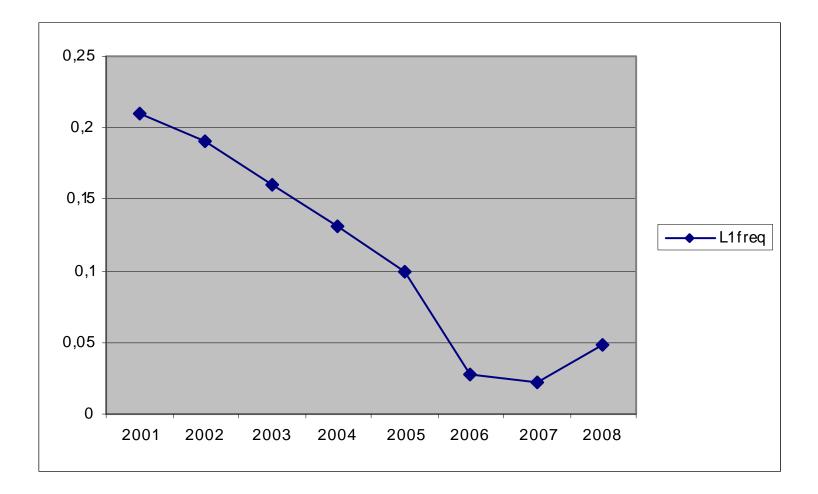


## Gene assisted selection 2006→





## Frequency of the L1-insertion





# Association of ISTS with production and reproduction traits

#### 361 sows

- GLM, fixed effect: sire (161), covariate: ISTS status
- 93 boars, 6 families
  - T-test between ISTS control vs. carriers
  - Nested effect GLM, ISTS status tested within sire families
- 421 piglets from carrier-carrier crosses
  - GLM, fixed effect: gender, tester (7), sire (41), dam (40)
    Covariate: ISTS status
- Marker: Line-1 insertion within KPL2



## **Reproduction traits**

- Age at first farrowing
- First and second farrowing interval
- Total number of piglets born in first and later parities
- Number of stillborn piglets in first and later parities
- Piglet mortality between farrowing and weaning for first and subsequent parities
- Fertility-index (calculated based on all reproduction traits)

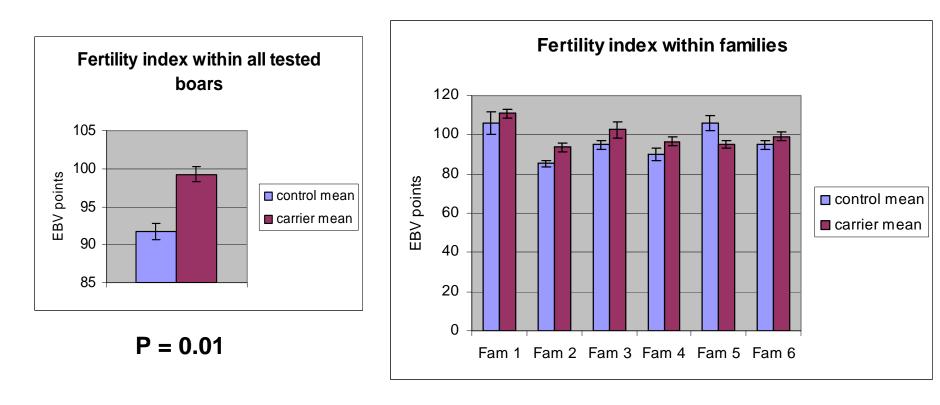


## Production and product quality traits

- Feed conversion ratio feed unit/kg
- Growth 0-30 kg and 30-100 kg
- Fat %,
- Meat %,
- Meat quality (pH and Minolta colour values)



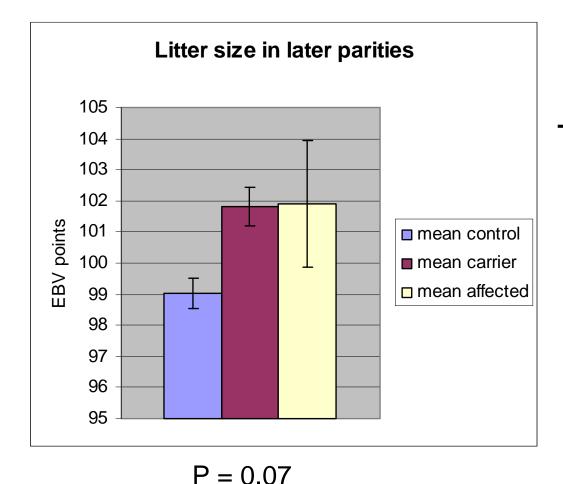
## Boar data results: reproduction



**P** = 0.33



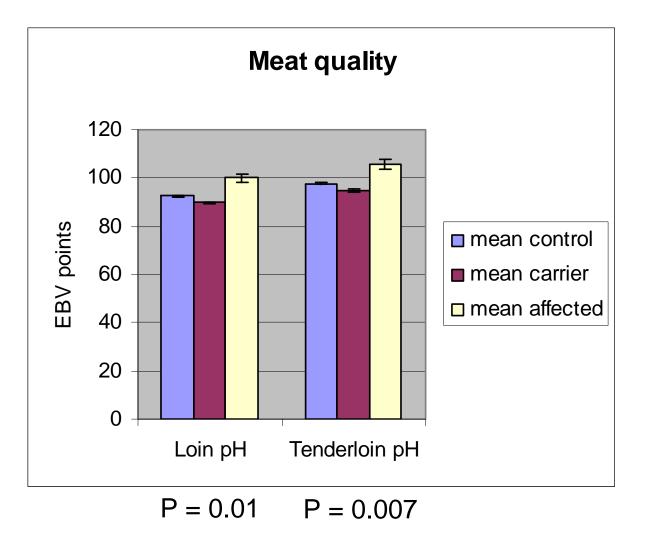
## Sow data results: reproduction



→ carriers produce
 0.12 extra piglets
 in parities >1



### Sow data results: production





## Conclusions

- Marker and gene assisted selections have reduced the frequency of ISTS mutation from 0.21 (2001) to 0.05 (2008)
- L1 insertion within KPL2 gene appears to be positively association with litter size
- L1 insertion is associated with meat quality

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## Thank you for your attention!

Collaborations: Faba Breeding, Finland Finnzymes Diagnostics, Finland

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