Strategies for selection against recessive lethals

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Genetic defects

- Recessive defects
  - The result is a poor or non functional enzyme or no enzyme product at all
- The defect must be inherited from both parents to effect the individ. One copy of the functional gene is sufficient for a normal development and life.

Genetic defects

Earlier

- It took about 30 years for a genetic defect to be spread in the population so the defected gene could be present in both mates when a maximum inbreeding of about 3 % was accepted
- The phenotype of the defect was the way we observed the defect.
- Only defects found in very influential bulls were detected.

Genetic defects

Today

- We can identify “missing homozygotes” in a population which are indicators of a lethal defect.
- “Missing homozygotes” = heterozygotes and one of the two homozygotes are present in the population.
- Chromosome deletions and aberrations can also be identified by genetic markers.
- The chance to find defects increase with increased number of DNA-markers.

Genetic defects, classification

- Patten of inheritance, is it recessive or not?
- Is the defect lethal or not?
- Can we find phenotypes or not?
- At what age is the phenotype expressed?
- Is a marker test available for the defect or not?
- **To what extent is the defect present in the population?**

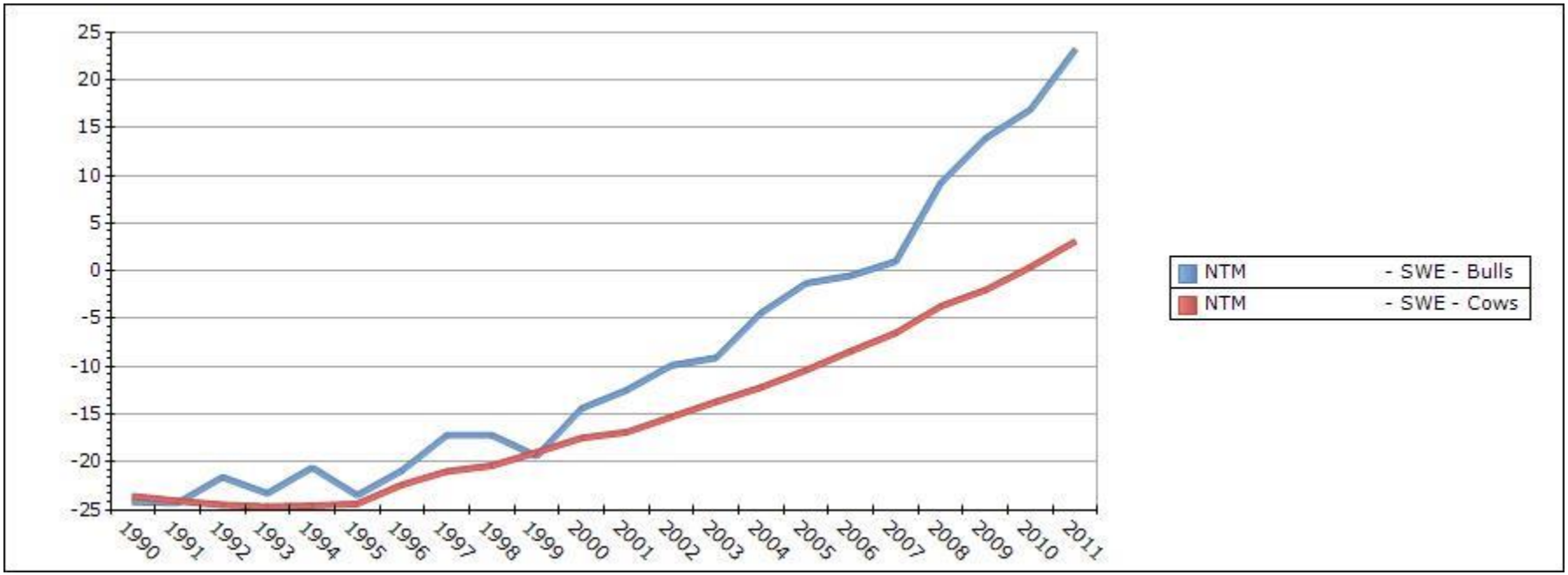
Genetic defects, strategies controlling males

- **Controlling inbreeding will decrease the risk of new defect to get a high frequency in the population.**

- Stop the use of carriers and test all bull calves before purchase
- Use carriers in this generation and test all bull calves before purchase
- Continue to use carriers on free tested females
- Continue to use carriers if the frequency in the population is low
- Continue to use carriers and only inform about who is a carrier

Genetic trend in Swedish Holstein bulls and cows

Bulls born 1999 were culled due to CVM.



What happens if we have many genetic defects?

Unexpected high proportion of carriers!

- The proportion of carriers have in some cases been considerable higher than the genetic contribution of any bull.
- We have actively selected for a traits that has a close link to the defect.
- About 30 % of the AI-candidates in the age class of 1999 were CVM carrier in Sweden and therefor not started in AI.

Genetic defects, strategy controlling females

- Is the marker test included in the LD-chip?
- Can females be tested free?
- We need to include a control of individual genes in the standard mating programs to avoid not recommended mating; controlling inbreeding on gene level!

- This control of inbreeding on specific allels might be a major reason for taking DNA-tests on herd level in the future.
- It is important that markers for genetic defects are included in the LD chip

Genetic defect index, proposal

Genetic defect index (GDI)

- We need a risk assessment for using a bull that is carrier of genetic defect compared with the use of other bulls.
- Lethal GDI = carrier (1) or not (0) of defect 1 * population gene frequency of defect 1 + .. + carrier (1) or not (0) of defect n * population gene frequency of defect n
- GDI = copies of the defected allel X1 (0, 1 or 2) * relative weight of X1 * population gene frequency of allel X1 +…+ copies of the defected allel Xn (0, 1 or 2) * relative weight of Xn * population gene frequency of allel Xn + lethal GDI

Conclusions

- More genetic defects will be found in the future, also defects that have a low frequency.
- It is important to have knowledge of the inheritance of the defect.
- It is important to have knowledge of the frequency of the unfavorable allel.
- We need to develop a tool for VG to evaluate the risk of including a bull in the breeding program in comparison with the inclusion of another bull, Genomic Defect Index.