The French observatory on genetic defect: an assessment after 15 years of operation

Coralie DANCHIN-BURGE, Institut de l’Elevage, France
Cécile GROHS, INRA GABI, France

coralie.danchin@idele.fr
Genetics defects in cattle

- Ne, the Effective Population Size (=diversity) in cattle is very low due to high selection pressure

Examples with French selected populations:
- Ne Holstein = 96
- Ne Charolais = 704

- Inbreeding rate ↠ ↠ = homozygous animals ↠ ↠

- Several crisis due to genetic defects since the 90s (BLAD, CVM, brachyspina, weaver syndrom etc.)

- Need to implement a specific monitoring on the emergence of genetic defects
Welfare and genetic defect

Axonopathy - Blonde d'Aquitaine
Welfare and genetic defect
Monitoring the emergence of genetic defects

French national observatory on genetic defects created in 2002 - ONAB

- Monitoring of genetic defects emergence: simple statistics based on a declaration form (paper, web site)
- Triggers scientific research when an emergence is detected in field
- Communication on genetic defects: newsletter, seminars, website...
Our goal: To be **effective**, despite a low rate of reporting

=> Using a participative science approach
“Devoted to the cause”

Core of proactive people - inseminator, veterinarian etc.
- Systematic report of any defects
- Early warning of emergences
First example: overbent fetlocks in the Montbeliardiad breed
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Information letter + website

ONAB

• AI center
• Breed’s organization

« Watchmen »

• Specific work with a vet school

Global network

Numerous reports (declaration forms) overbent fetlocks

• Specific forms
• Blood/ear samples

On going research
Second example: “no hair or teeth” in the Charolais breed
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*Anhidrotic Ectodermal Dysplasia*

- **First alerts:**
  - Participative website restricted to vets only, “vetofocus” (2013) – however the ONAB is not contacted
  - A veterinarian specialized in cattle dermatological diseases contacts the ONAB at the same time

- **First specific work by the ONAB:**
  - Anhidrotic Ectodermal Dysplasia= a well known genetic defect. But what triggers our attention: both males and females have the disease (in the literature: only males)
  - Analysis of the pedigree structure: a mutual ancestor is found => recessive allele

=> Specific calls for new cases with biological samples
Second example: “no hair or teeth” in the Charolais breed

• Samples genotyping with a 50K SNP chip (< 10 animals)

=> Large common haplotype found in a gene responsible for hair growth

=> New mutation, that starts to spread in the Charolais population (0.5% of the population is carrier, mutation in AI and natural service lines)

=> Development of a specific genotype test (now combined with other genetic defects such as ataxia and progressive blindness in Charolais)
**Perspectives for monitoring the emergence of genetic defects**

- **ONAB - French national observatory on genetic defects**
  - Monitoring of genetic defects emergence
  - Triggers scientific research (including fine phenotyping)
  - Communication: newsletters, website, seminars...

- **National genetic database**
  - Bulls genetic evaluations on vitality
  - Mortality observatory (bases on calves exit dates)

- **Genomic data**
  - Detection of haplotypes associated with prenatal death
  - Reverse genetic approach

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Year Timeline:
- 2002
- 2016
Acknowledgment

And all the ONAB partners: