Observatoire National des Anomalies Bovines

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

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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

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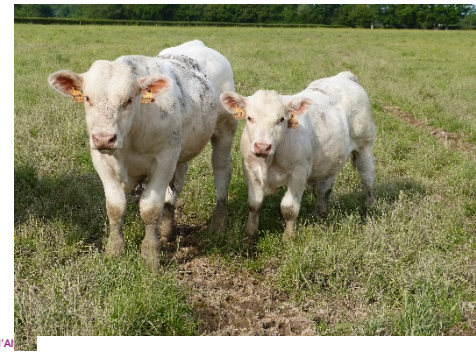
Monitoring the emergence of genetic defects

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…



Dwarfism in Charolais

Limiting factors:

- Restricted number of cases reported
- Inaccurate phenotypic descriptions which can lead to a wrong diagnosis of the situation




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 Montbeliard breed

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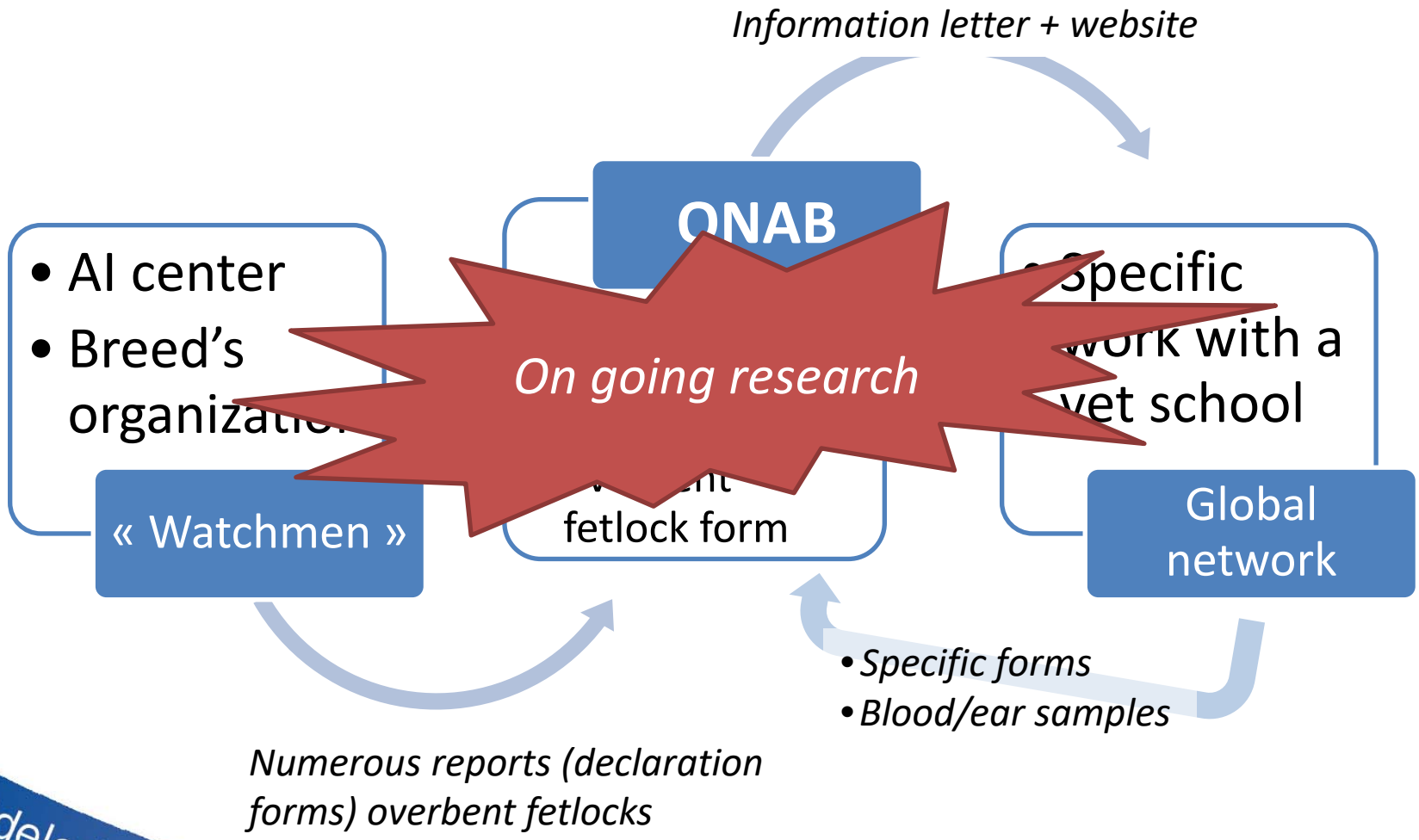

Second example: “no hair or teeth” in the Charolais breed

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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

2002

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

2016

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