



# IDEA development

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# Additional Animal Information module

- \* Allow member organizations to upload additional information about animals in IDEA.
- \* Questionnaire sent out to 46 member organizations.
- \* Contribution of the information is voluntary.
- \* Information will be available to all organizations with login access to IDEA.



# Type of information

- \* Start with the type codes (additional information) in the questionnaire from 2015
- genotyping
- twin and et animals
- genetic defects
- coat color
- polled/not polled, scurs
- Kappa Casein
- + % RH genes in Simmental



# Additional Animal Information

- \* There will be a CheckAnimInfo.py program available for downloading from IDEA software tab.
- \* A zip file is created if the coding in the file is according to the Business Rules.
- \* Form for new type codes on ITBC homepage.
- \* Pre-study by ITBC.
- \* ITBC - Business Rules.
- \* ITBC adds the type code and Business Rules in IDEA.
- \* Information is ready to be uploaded to IDEA.



# Business rules

\* Important with the same references for the information in IDEA.

\* Business rules for type codes

Genotyped, Twin, ET (etc) – ITBC

Genetic defects – OMIA + ITBC

Exampel : Hypotricosis <http://omia.angis.org.au/OMIA000540/9913/>

Coat colors – OMIA +ITBC

Exampel: Albinism <http://omia.angis.org.au/OMIA000202/9913/>

Polled/Scurs – OMIA +ITBC

Exampel :Polled/Horns <http://omia.angis.org.au/OMIA000483/9913/>





# OMIA

- \* Include a Note in OMIA:  
*“Information reported to Interbull Centre for brd1,brd2..”*
- \* Breed codes from the 3 first characters in the animal id .
- \* OMIA will get information about breeds registering the OMIA id.
- \* If there is no Note, the OMIA id is not reported to Interbull Centre.
- \* Member organizations can send a request to ITBC.
- \* The OMIA id will be added as accepted OMIA id to report to ITBC.



You are here: [OMIA](#) / [Search](#) / [OMIA 000595](#) / [cattle](#)

## OMIA 000595-9913 : Leukocyte adhesion deficiency, type I in *Bos taurus*

[See the equivalent entry at ICBI](#)

In other species: [dog](#) , [water buffalo](#)

Possible human homologue (MIM number): [116920](#)

Mendelian trait/disorder: yes

Mode of inheritance: Autosomal Recessive

Considered a defect: yes

Key mutation known: yes

Year key mutation first reported: 1992

**Cross-species summary:** Affected animals die because of extreme susceptibility to infections, caused by an inability of white blood cells (leukocytes) to pass from the blood stream into infected tissue. This inability is due to the lack of a membrane glycoprotein called the leukocyte integrin beta-2 subunit or CD18.

**Species-specific name:** Bovine leukocyte adhesion deficiency; Haplotype HHB

**Species-specific symbol:** BLAD; HHB

**Mapping:** In the course of their large-scale study of BovineSNP50 BeadChip haplotypes that are common but never homozygous, VanRaden et al. (2011) confirmed the mapping of this disorder to BTA1 at 141-146Mb (UMD 3.0 genome assembly).

**Molecular basis:** By cloning and sequencing a very likely comparative candidate gene (based on the same disorder in humans), Shuster et al. (1992; PNAS) showed that this disorder in Holstein cattle is due to a missense mutation (c.383A>G) in the CD18 gene, now known as ITGB2.

This mutation was confirmed in Daetwyler et al. (2014)'s analysis of whole-genome sequence data from 234 cattle, including key ancestors of the Holstein-Friesian, Fleckvieh and Jersey breeds: the c.383A>G mutation was identified in only four Holstein bulls in this study, each of which had previously been identified as a carrier.

**Breed:** Holstein.

**Associated gene:**

Symbol	Description	Species	Chr acc	Chr name	Start	Stop	OMIA gene details page	Other Links
ITGB2	integrin beta-2 (complement component 3 receptor 3 and 4 subunit)	<i>Bos taurus</i>	no genomic information	-	-	-	<a href="#">ITGB2</a>	<a href="#">Homologene</a> , <a href="#">Ensembl</a> , <a href="#">NCBI gene</a>

### References

Note: the references are listed in reverse chronological order (from the most recent year to the earliest year), and alphabetically by first author within a year.



# File format

- \* New file format XML (Extensible Markup Language)
- \* Why XML?
- \* Flexible file format
- \* When fixed character width data file formats become too complex, with for example the number of columns depending on values in previous columns or from data in other files, the effort to write parsers to read and write the information and make sure it is correctly understood becomes very time- and resource consuming.
- \* ITBC will create a page in IDEA with useful documentation (manuals, file examples etc)





# Exampel coding with XML file format

```
<interbull type="animinfo" version="1.0">
  <animals>
    <a id="HOLUSAM123838482929">
      < GENOTYPE coding="Y" />
      < ET coding="Y" />
      < TWIN coding="N" />
      < COAT_COLOR value=" Albinism" coding="X" />
      < GENETIC_DEFECT value="Bulldog" coding="Y"/>
    </a>
    <a id="HOLCANF129375273837">
      <GENOTYPED coding="N" />
      <KCAS coding="AB" />
    </a>
  </animals>
</interbull>
```



# IDEA

- \* Any organization with information about an animal is allowed to upload additional information.
- \* Animal id must be registered in IDEA.
- \* New menu bar in IDEA called "AnimInfo".
- \* Submenues
  - upload
  - query
  - conflict
    - I) defect
    - ii) coat color
    - lii) etc
- \* Conflicting information will be solved according to the same principle as potential duplicate ids.
- \* Log file will be produced at uploading.





## Animal Info Structure overview

### Allowed animal information types and their specifications

- **COAT:** Information pertaining to the coat of the animal
  - red (restricted set): prevalence of red coat
    - **Allowed values:**
      - yes
      - no
  - red\_carrier (restricted set): animal with B&W phenotype but producing red offsprings
    - **Allowed values:**
      - yes
      - no
- **GENETICS\_DEFECTS:** Information pertaining to recessive lethal genes
  - blad (restricted set): bovine leukocyte adhesion deficiency (BLAD)
    - **Allowed values:**
      - no
      - yes
  - cvm (restricted set): Complex vertebral malformation (CVM)
    - **Allowed values:**
      - yes
      - no
- **GENOLIST:** Information pertaining to genotyped animals
  - genotyped (restricted set): animal has been genotyped
    - **Allowed values:**
      - no
      - yes
  - snp\_chip (free text): type of chip used for genotyping the animal
- **RH\_GENES:** % RH genes in Simmental
  - percent (free text): percent rh genes in simmental

### Value type definitions

- **free text:** a string of text with any content
- **restricted set:** a restricted set of specified, allowed values



# IDEA - OMIA

- Increase visibility of Interbull Centre for OMIA users
- Increase visibility of OMIA to ITB organizations
- OMIA will get information about breeds registering the OMIA id .
- Interbull Centre will get standardized business rules from OMIA
- Interbull Centre will become a repository for dairy and beef cattle
  - a) pedigree
  - b) breeding values
  - c) additional animal information
    - coat color
    - genetic defects
    - et/twin animal
    - etc
  - d) genotypes



# Acknowledgements

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