

# Towards better understanding of genetic variation in dairy cattle breeds

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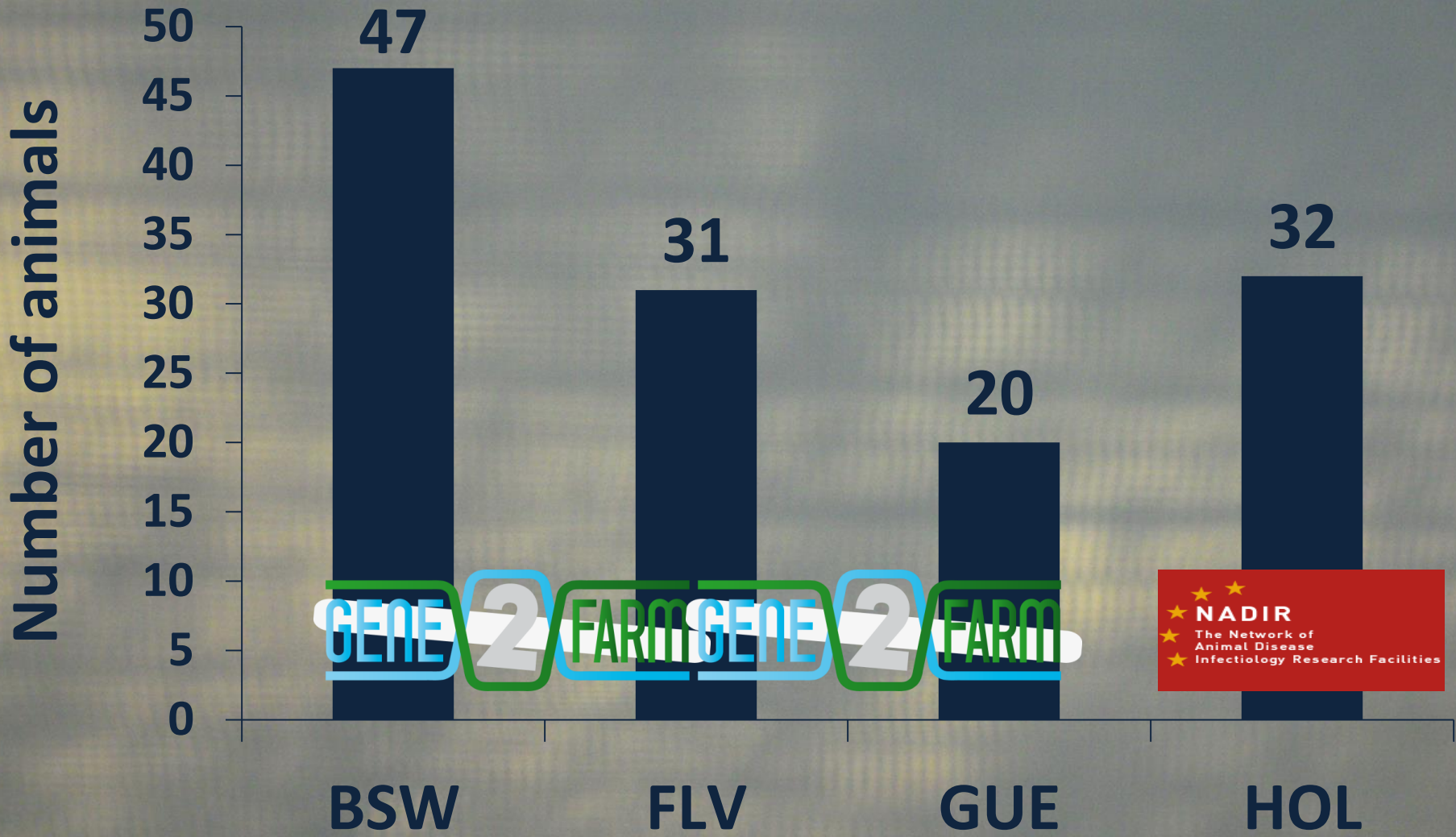


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**Describe genome-wide distribution of functional variants  
... in various dairy breeds**

- 1. Data – animals & sequence**
- 2. Methods – variant genomic annotation**
- 3. Results – Sequence Ontology**
- 4. Results – genes**
- 5. Results – pathways**
- 6. Conclusions**

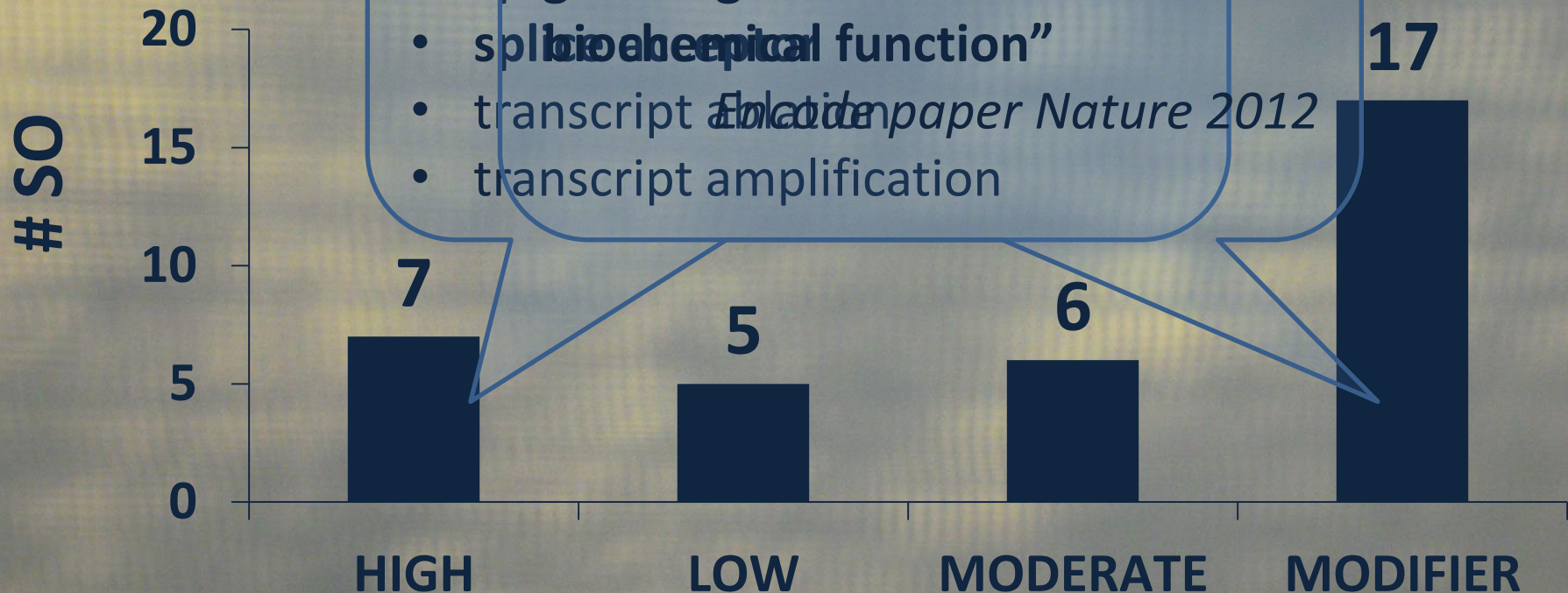
sequenced animals



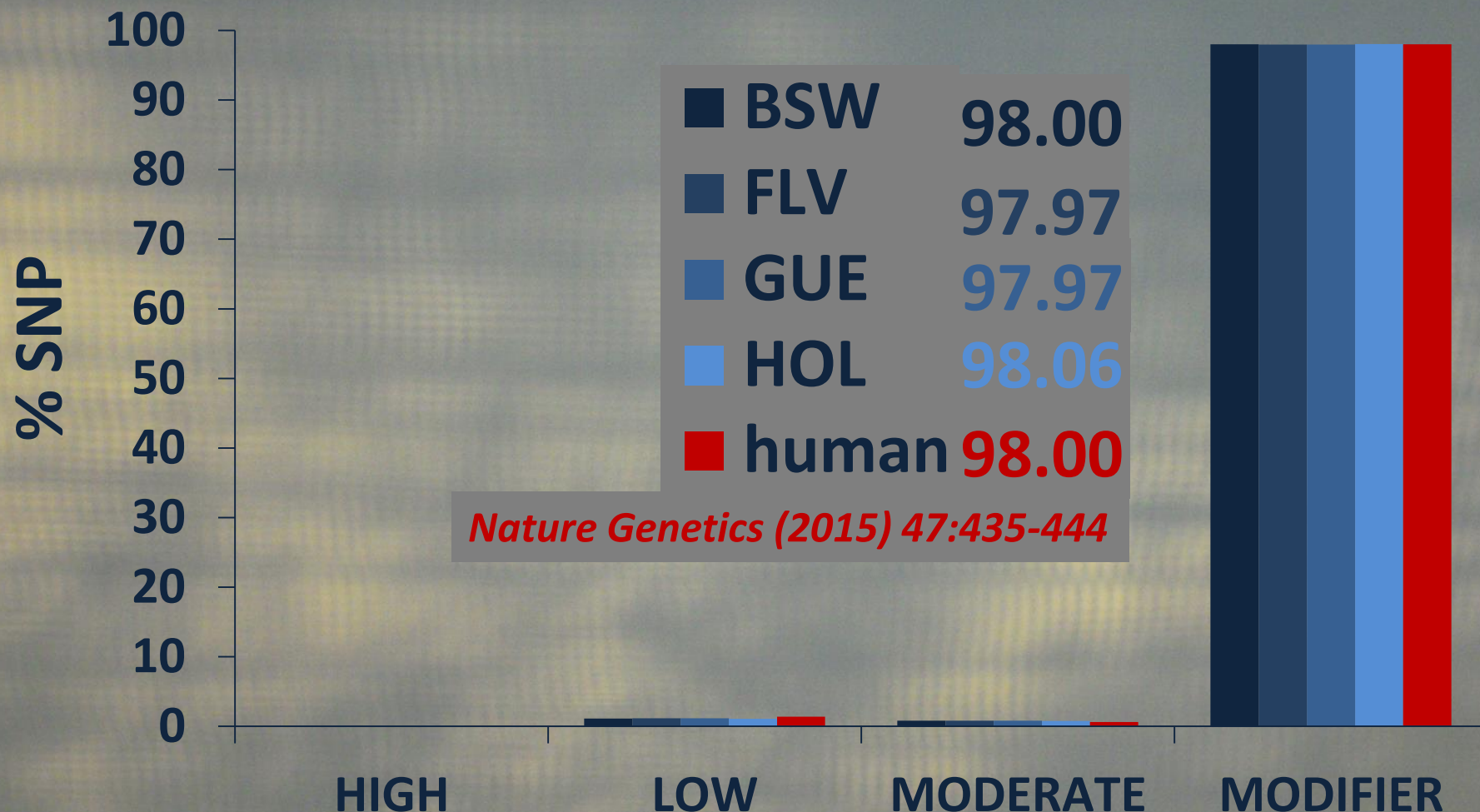


## variant annotation

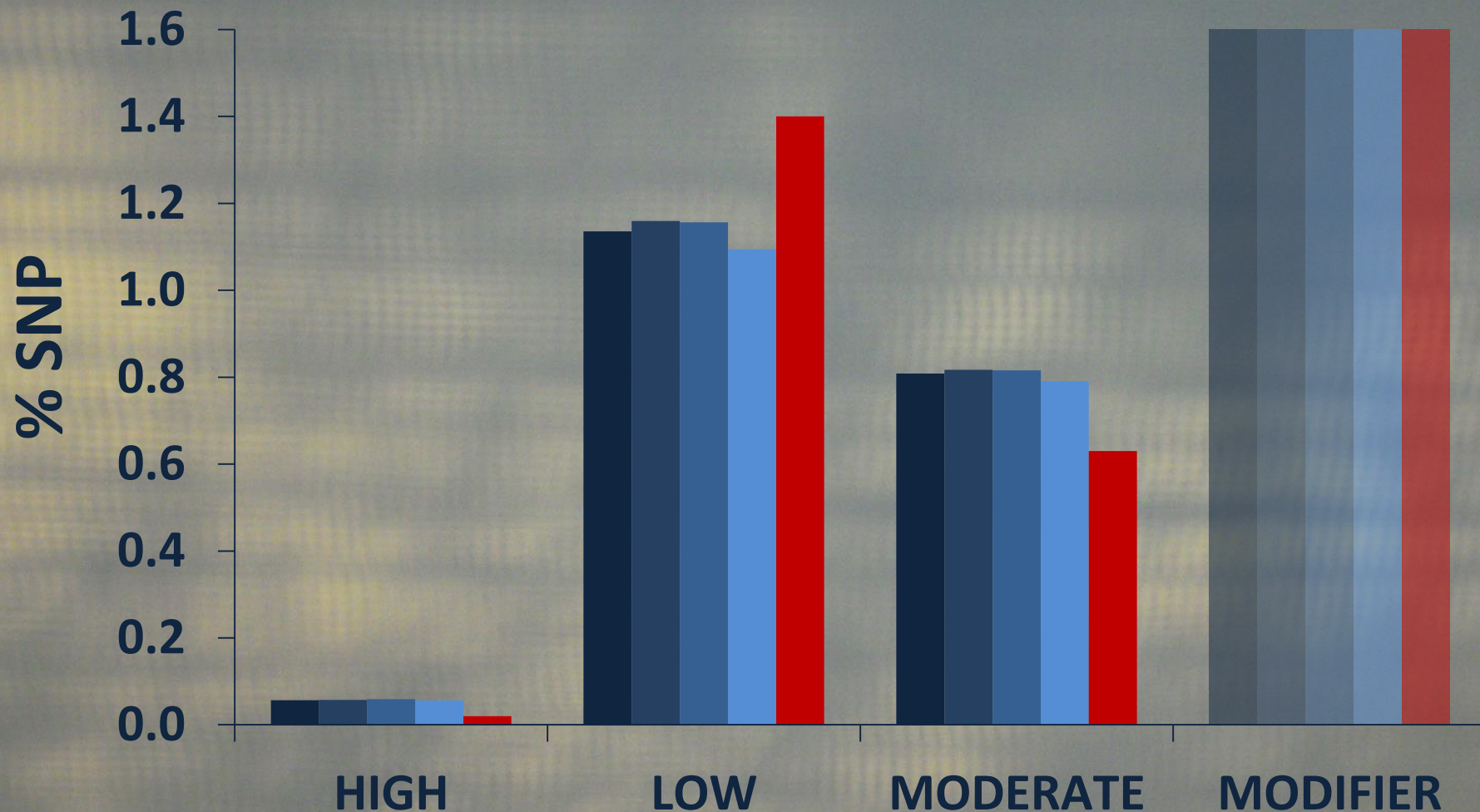
- Variant Effect Predictor → Ensembl
  - Annotation based on position in UMD3.1
  - Classification in SO terms → 24/35 terms
- frameshift variant
  - stop-gained information in the human genome may be important for gene regulation than for splice acceptor function”
  - transcript elongation paper Nature 2012
  - transcript amplification



## variants in Sequence Ontology classes



## variants in Sequence Ontology classes





# correlations between numbers of variants

	HIGH	LOW	MODERATE	MODIFIER
HIGH	1.00	0.91	0.96	0.89
LOW		1.00	0.99	0.97
MODERATE			1.00	0.96
MODIFIER				1.00

→ BSW

	HIGH	LOW	MODERATE	MODIFIER
HIGH	1.00	0.99	0.99	0.99
LOW		1.00	1.00	0.99
MODERATE			1.00	0.99
MODIFIER				1.00

→ FLV

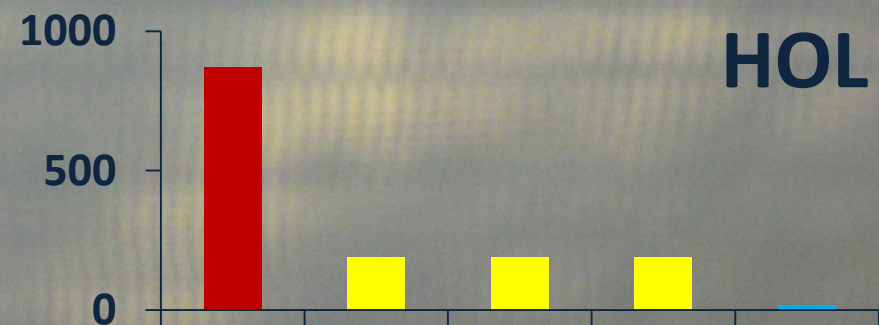
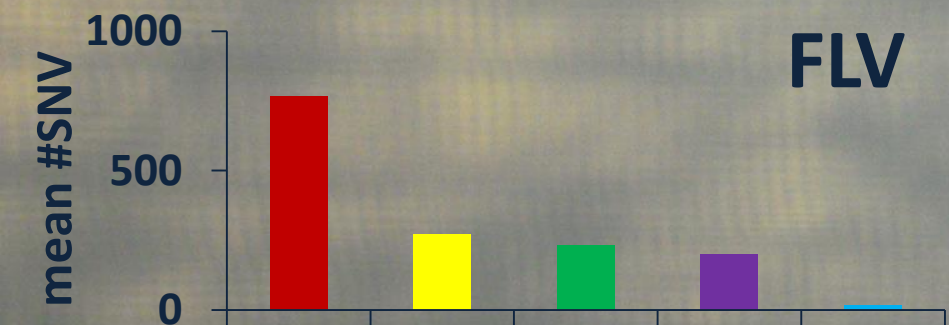
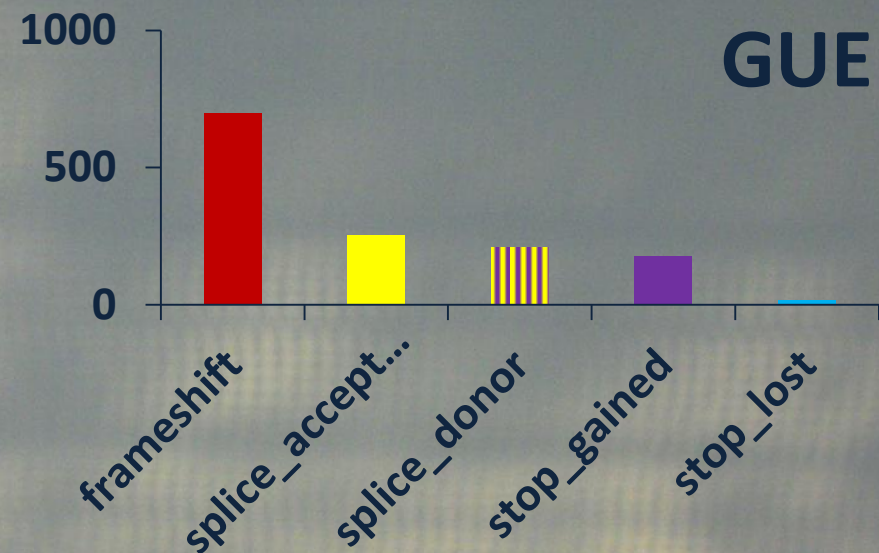
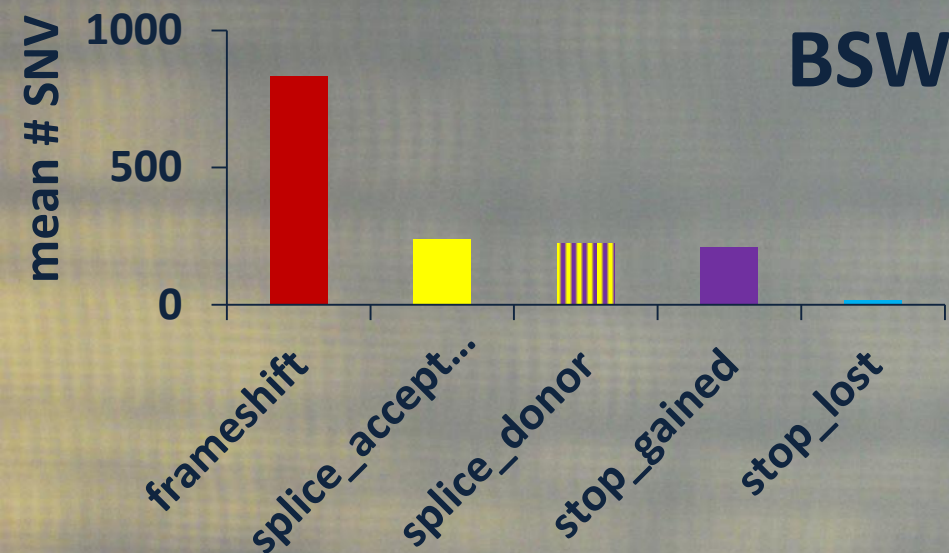
	HIGH	LOW	MODERATE	MODIFIER
HIGH	1.00	1.00	1.00	1.00
LOW		1.00	1.00	1.00
MODERATE			1.00	1.00
MODIFIER				1.00

→ GUE

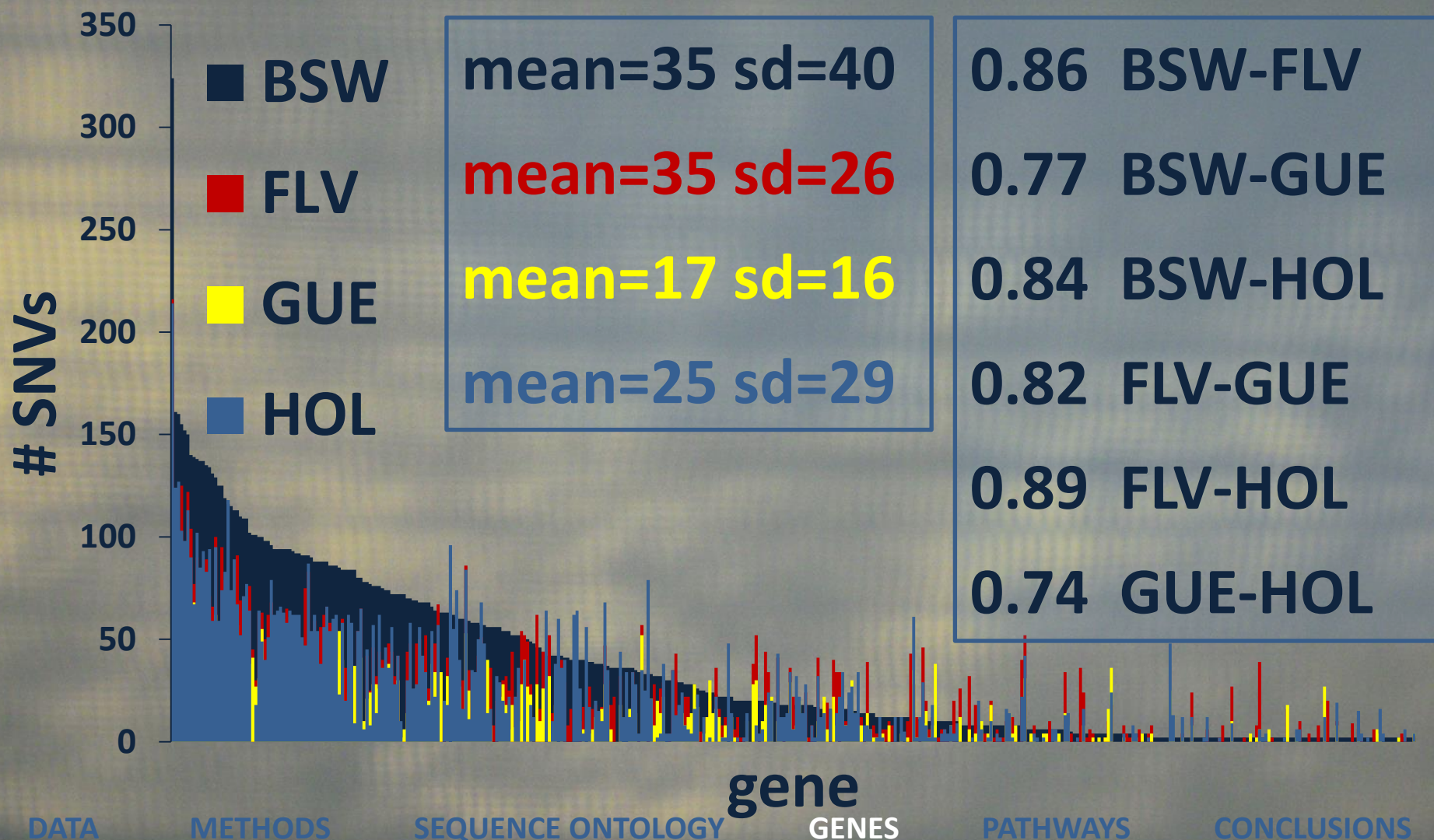
	HIGH	LOW	MODERATE	MODIFIER
HIGH	1.00	0.97	0.97	0.91
LOW		1.00	0.99	0.89
MODERATE			1.00	0.91
MODIFIER				1.00

→ HOL

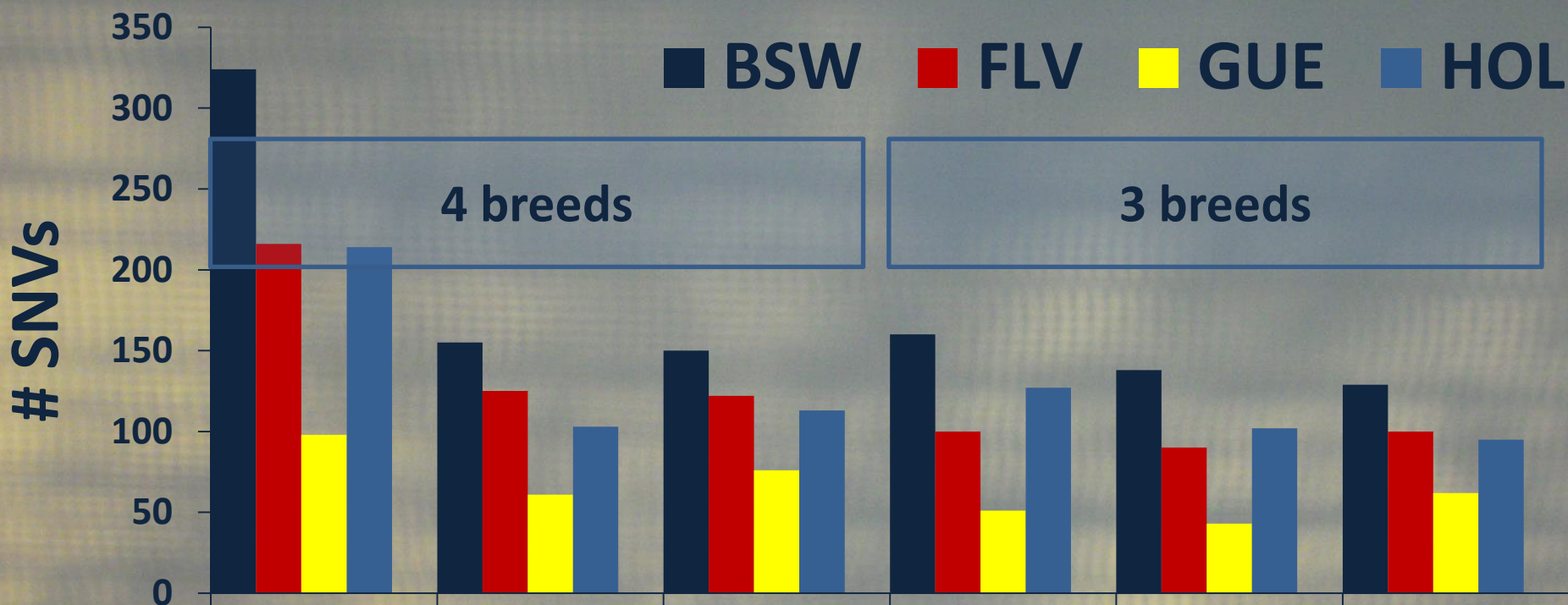
## variation in SNV number between HIGH SO



## HIGH SO SNV distribution within genes



## top 10 genes with the most SNVs in HIGH class



CACNA1H

00000015666

SIPA1L3

00000010463

TNFRSF10D

SLIT3

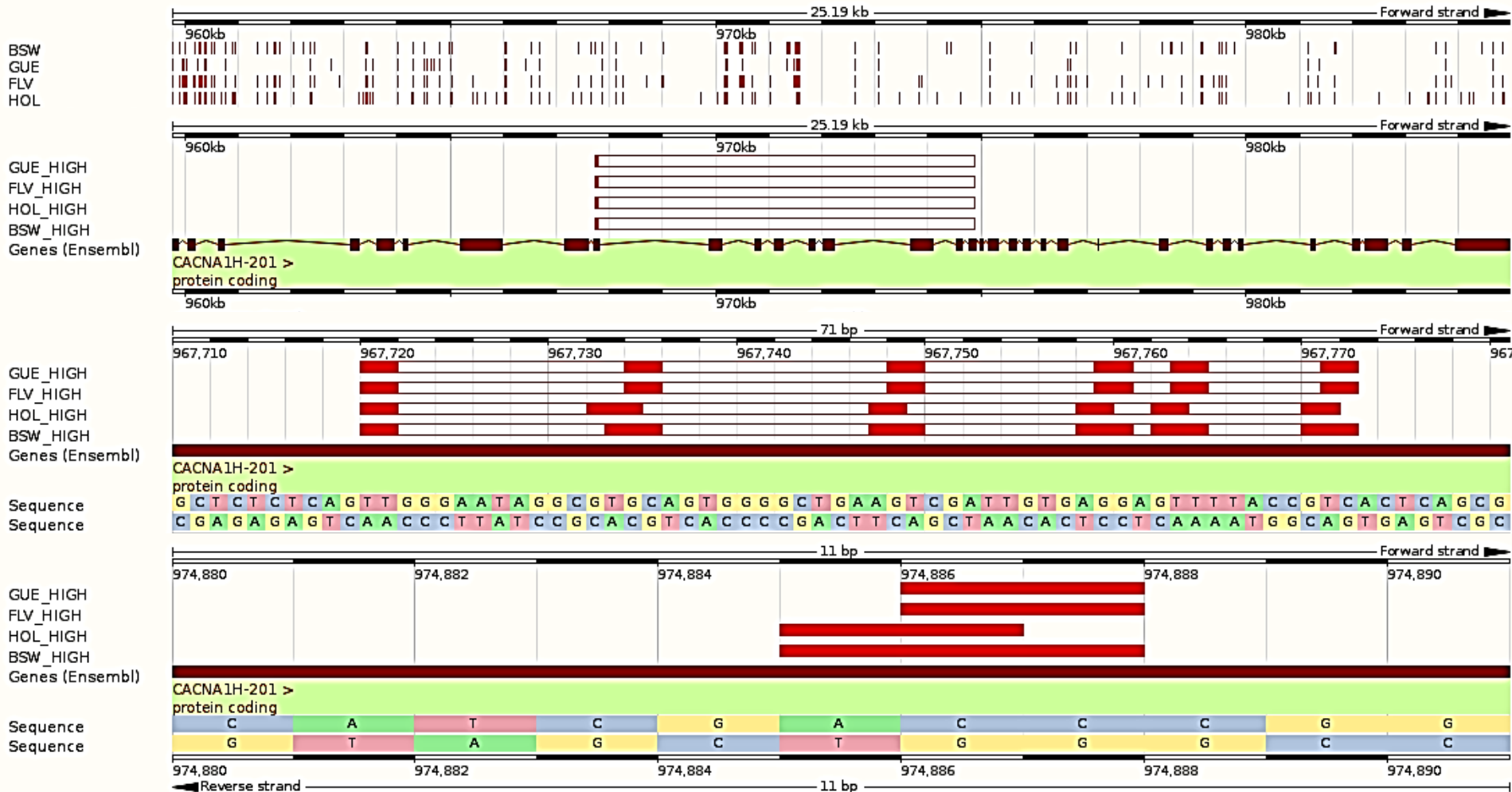
**bta25**  
33 exons  
1 transcr

**bta18**  
23 exons  
1 transcr

**bta08**  
10 exons  
2 transcr

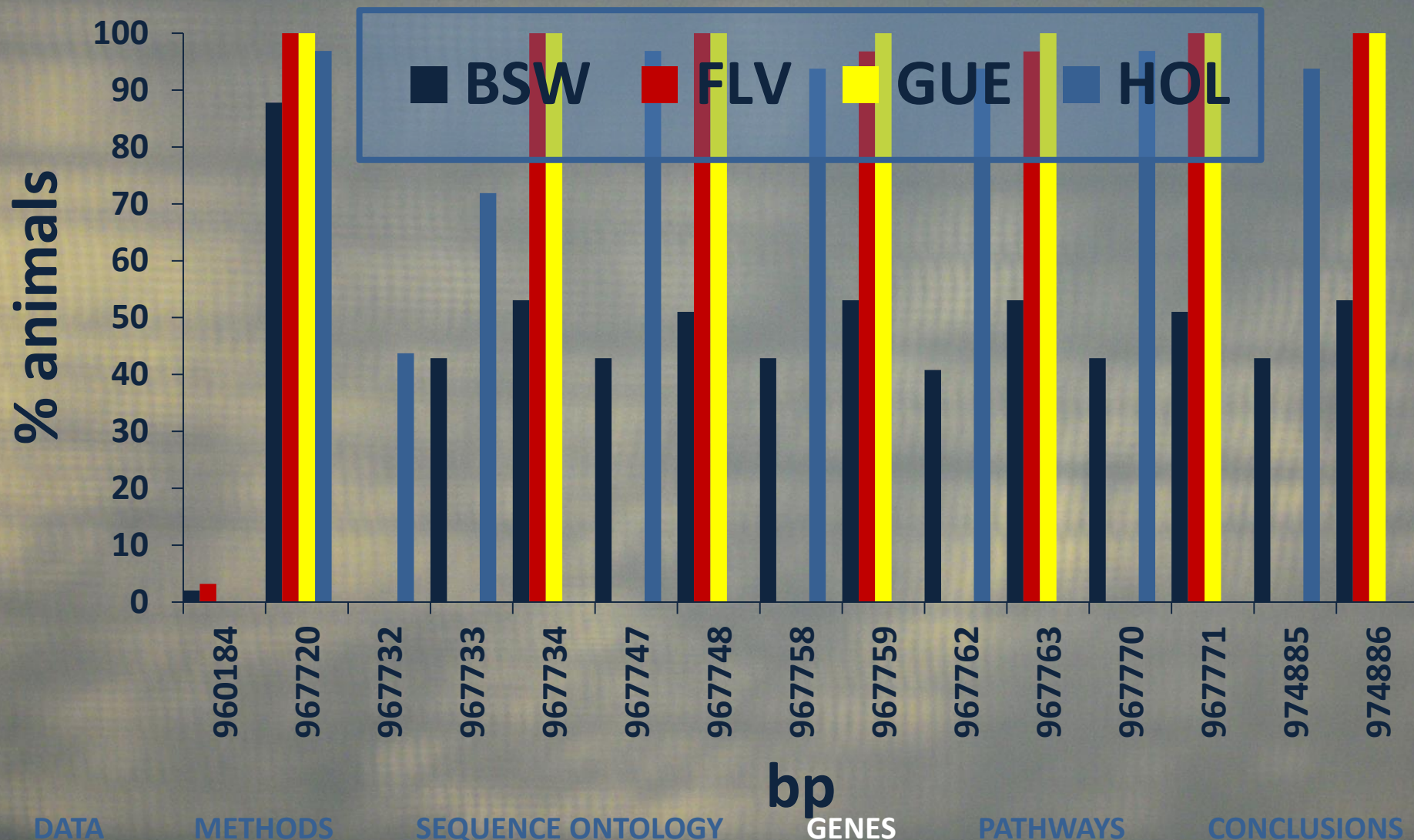
**bta20**  
34 exons  
1 transcr

## high SO SNV distribution across CACNA1H

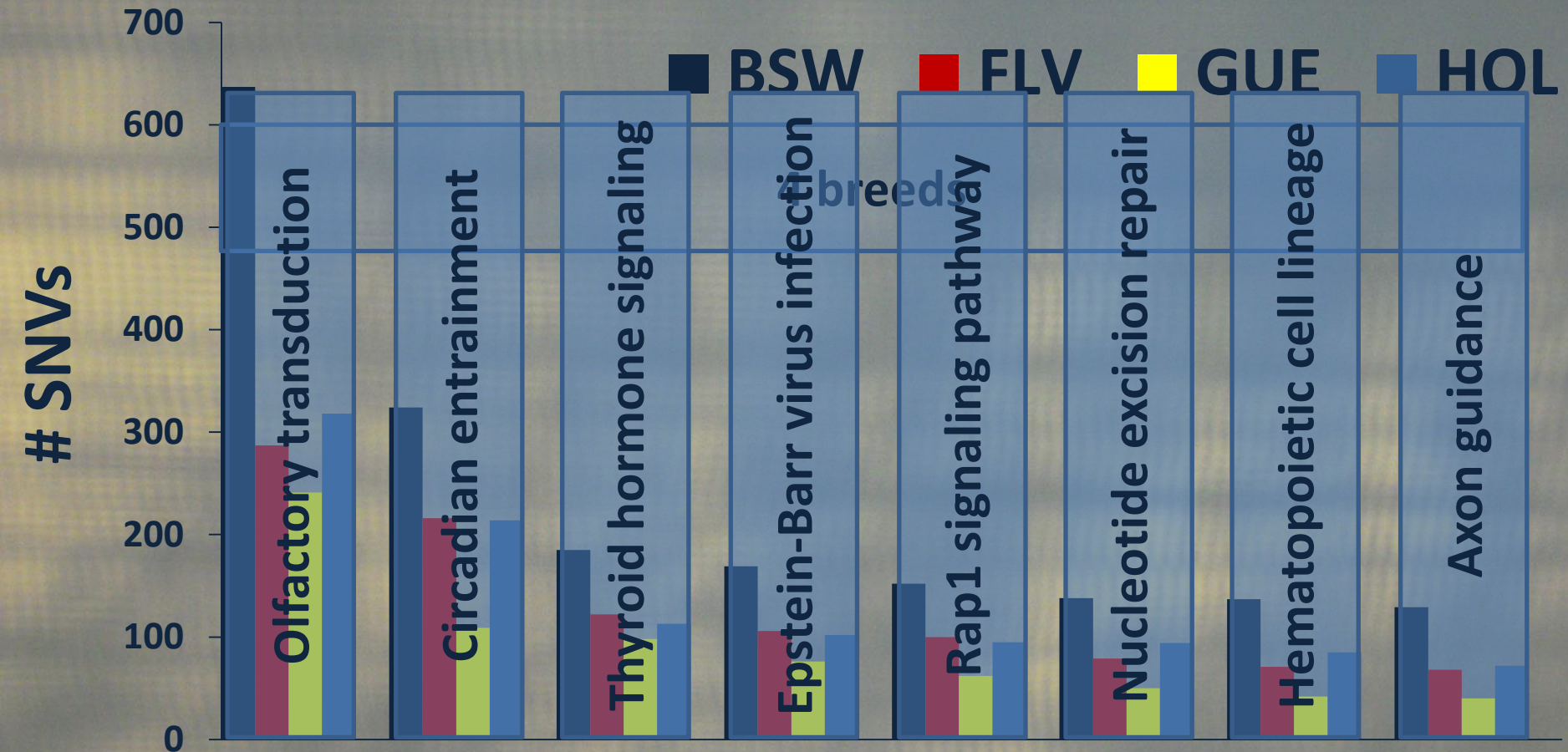


There are currently 63 tracks turned off.  
 Ensembl Bostaurusversion 81.31 (UMD3.1) Chromosome 25: 974,880 - 974,890

## % of animals with high SO variants in CACNA1H



## top 10 KEGG with the most SNVs in HIGH class



## 1. CACNA1H

- Calcium channels are located in cell membranes → responsible for communication between cells → Icelandic population *Nature Genetics 2015*
- Functional SNVs → indels
- True variants or reference genome artefacts ?



## 2. KEGG

- **poor annotation in cattle**
- **Most variable pathway categories:**
  - Organismal Systems, Diseases, Environmental Information Processing, Genetic Information Processing
- **Most variable pathways:**
  - Not directly related to traits under selection
  - Related to reaction to environment (infection, response to stimuli, DNA repair, nervous and hormone signaling)
- **No excess of variation in pathway categories:**
  - Metabolism → diminished by selection ?
  - Cellular Processes → conserved ?
  - Drug Development → no annotation in cattle

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Riccardo Giannico**

**Daniele Vicario**

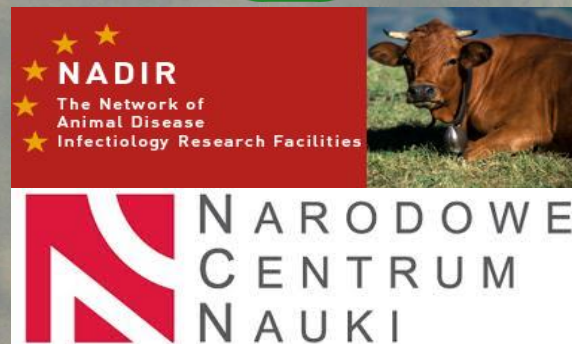


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Universidad  
de Zaragoza

Qualitas



Associazione Nazionale Allevatori  
Bovini della Razza Bruna

GENO

ZuchtData

Roslin BioCentre

**Thank you ...**

## SNV density

CACNA1H

Intergenic region on BTA25

