

# COPY NUMBER VARIATIONS AND QUANTITATIVE TRAIT LOCI IN SOUTH AFRICAN BRAHMAN CATTLE

M.D. Wang<sup>1</sup>, F.C. Muchadeyi<sup>2</sup>, M. Makgahela<sup>1,3</sup>, S. Mdyogolo<sup>1</sup> & A. Maiwashe<sup>1,3</sup>

<sup>1</sup> Agriculture Research Council-Animal Production Campus, Private Bag X2, Irene, 0062, South Africa. <sup>2</sup>Agriculture Research Council-Biotechnology Platform, Private Bag X1, Onderstepoort, 0110, South Africa. <sup>3</sup> University of the Free State, 205 Nelson Mandela Drive, Park West, Bloemfontein, South Africa

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11-16 February 2018

11<sup>th</sup> WORLD CONGRESS  
ON GENETICS  
APPLIED TO  
LIVESTOCK PRODUCTION  
[wcgap.com](http://wcgap.com)



# Background

# GENETIC VARIATION

Single base pair



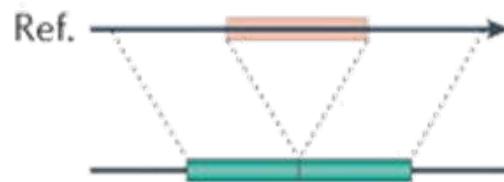
Large chromosomal events



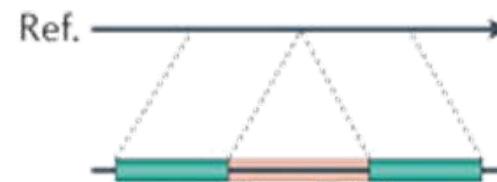
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# STRUCTURAL VARIATION

**Deletion**



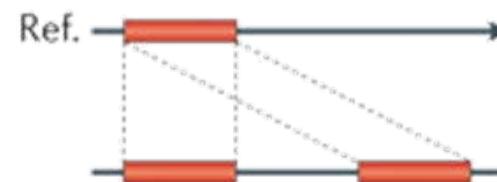
**Novel sequence insertion**



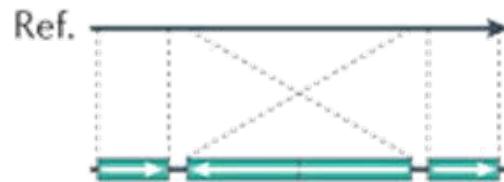
**Tandem duplication**



**Interspersed duplication**



**Inversion**



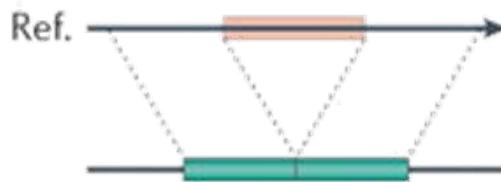
**Translocation**



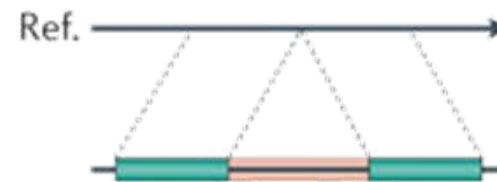
Nature Reviews | Genetics

# COPY NUMBER VARIATION

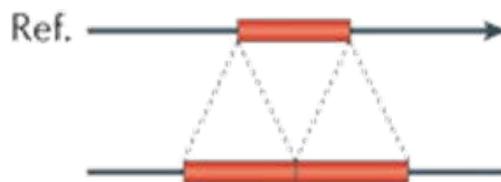
**Deletion**



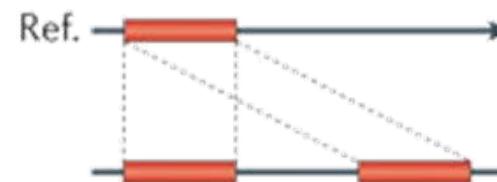
**Novel sequence insertion**



**Tandem duplication**



**Interspersed duplication**



# COPY NUMBER VARIATION

**CNVs**

**Cattle**

- Disease
- Adaptation
- Production
- Breed formation



# BRAHMAN CATTLE IN SOUTH AFRICA



- 1954
  - 8 bulls
  - 10 cows
- 2018
  - 60 000 +

<http://www.vidiani.com>



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# BRAHMAN CATTLE IN SOUTH AFRICA



➤ Adaptation



# BRAHMAN CATTLE IN SOUTH AFRICA



- Adaptation
- Maternal line



# Materials & Methods

**40 RSA Brahman Bulls**



**Extract DNA**

**Bovine HD**

**GenomeStudio**



**40 RSA Brahman Bulls**



**Extract DNA**

**Bovine HD**

**GenomeStudio**



**Plink**

**SNP QC**

**LogR ratio  
B Allele frequency**

**PennCNV**



**40 RSA Brahman Bulls**



**Extract DNA**

**Bovine HD**

**GenomeStudio**



**Plink**

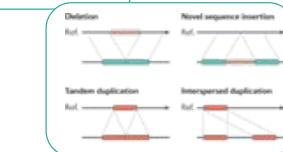
**SNP QC**

**LogR ratio  
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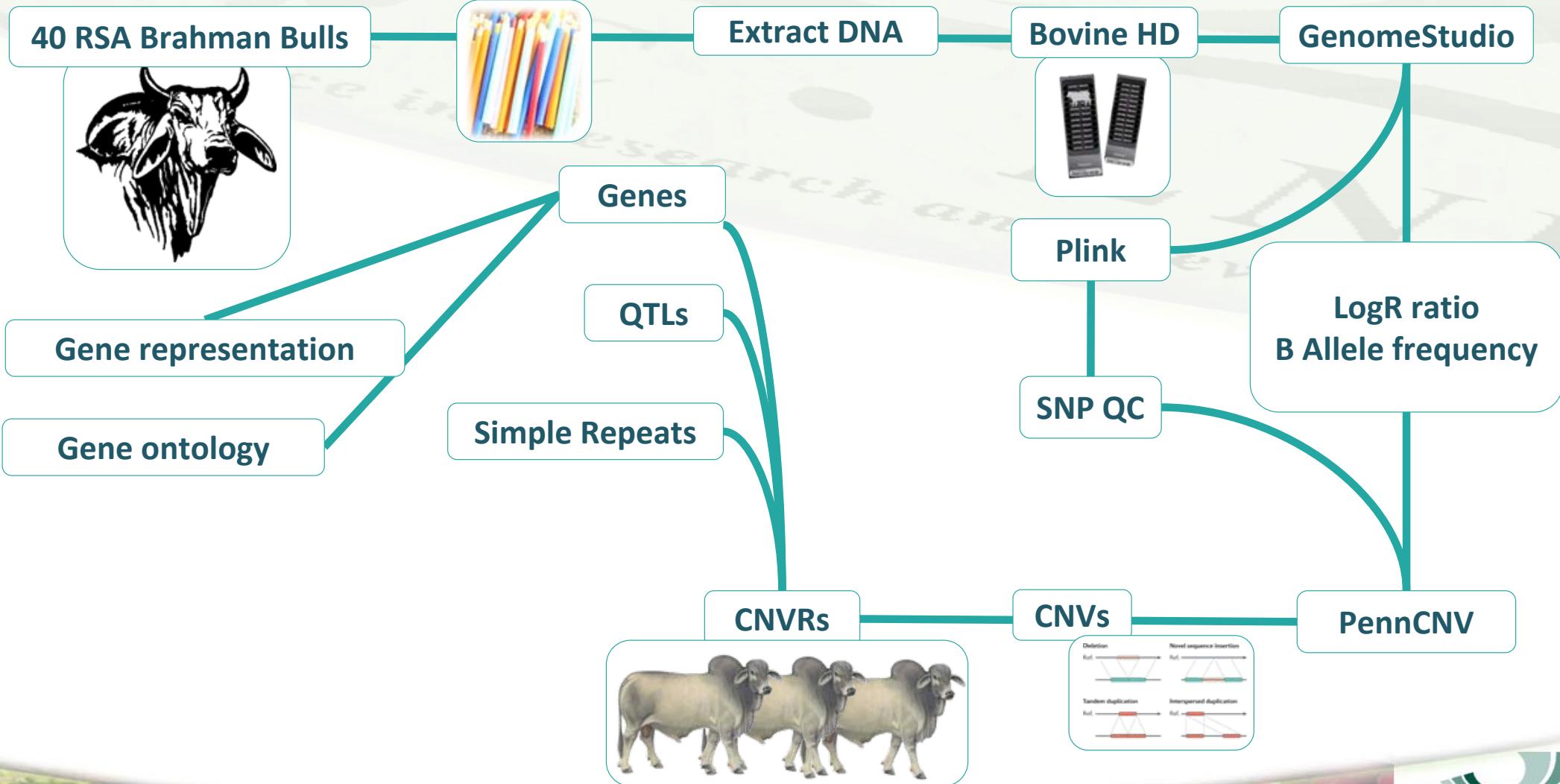
**CNVRs**



**CNVs**



**PennCNV**



# Results

# GENOME WIDE CNVR DETECTION

CN	CNVR	MinL	AvL	MaxL	SumL
DEL	22	2 566	56 879.46	34 300	1 290 500
DEL/DUP	19	2 965	259 546.26	506 110	4 931 379
DUP	10	25 983	87 832.10	276 838	878 321
<b>Total</b>	<b>51</b>	<b>2 566</b>	<b>138 451.92</b>	<b>506 110</b>	<b>7 061 048</b>



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<b>DEL</b>	22	2 566	56 879.46	371 870	1 251 348
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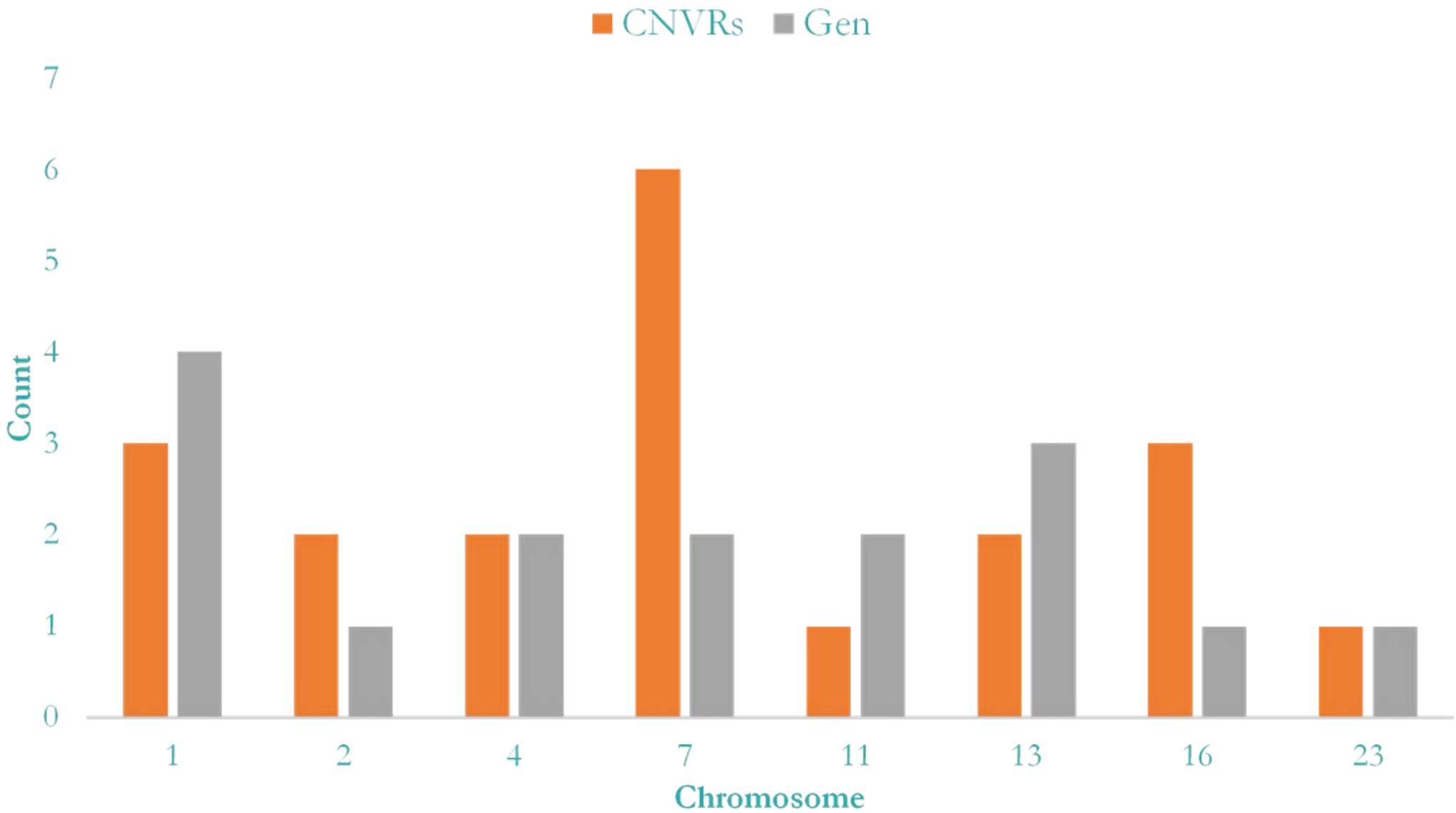
Chr	Start(bp)	End(bp)	Wang ☺	Jiang et al.(2012)	Hou et al.(2012)	Zhang et al.(2015)
1	103 158 887	103 180 729	Del	Dup	Dup	
3	54 522 367	54 968 058	Del/Dup	Del	Del	Dup
4	113 696 929	113 757 613	Dup	Dup	Del/Dup	Dup
5	73 185 738	73 190 311	Del		Del	Del
6	3 190 744	3 217 181	Dup		Dup	Dup
7	42 736 530	42 861 486	Del/Dup	Dup	Del/Dup	
9	53 896 584	53 899 150	Del		Del/Dup	Del
10	22 492 853	22 968 477	Del/Dup	Del/Dup		Del
12	72 419 043	72 713 303	Del	Del/Dup	Del/Dup	Del/Dup
12	70 376 431	70 809 607	Del/Dup	Del/Dup	Del/Dup	Del
12	71 617 637	72 123 747	Del/Dup	Del/Dup		Del
12	75 462 926	75 946 247	Del/Dup	Del/Dup		Dup
15	45 475 858	45 554 117	Del		Del	Del
16	7 666 301	7 713 008	Dup		Del/Dup	Dup
18	61 722 654	61 810 826	Del/Dup	Del	Del/Dup	Del
23	25 417 035	25 443 589	Del	Dup	Del/Dup	Del

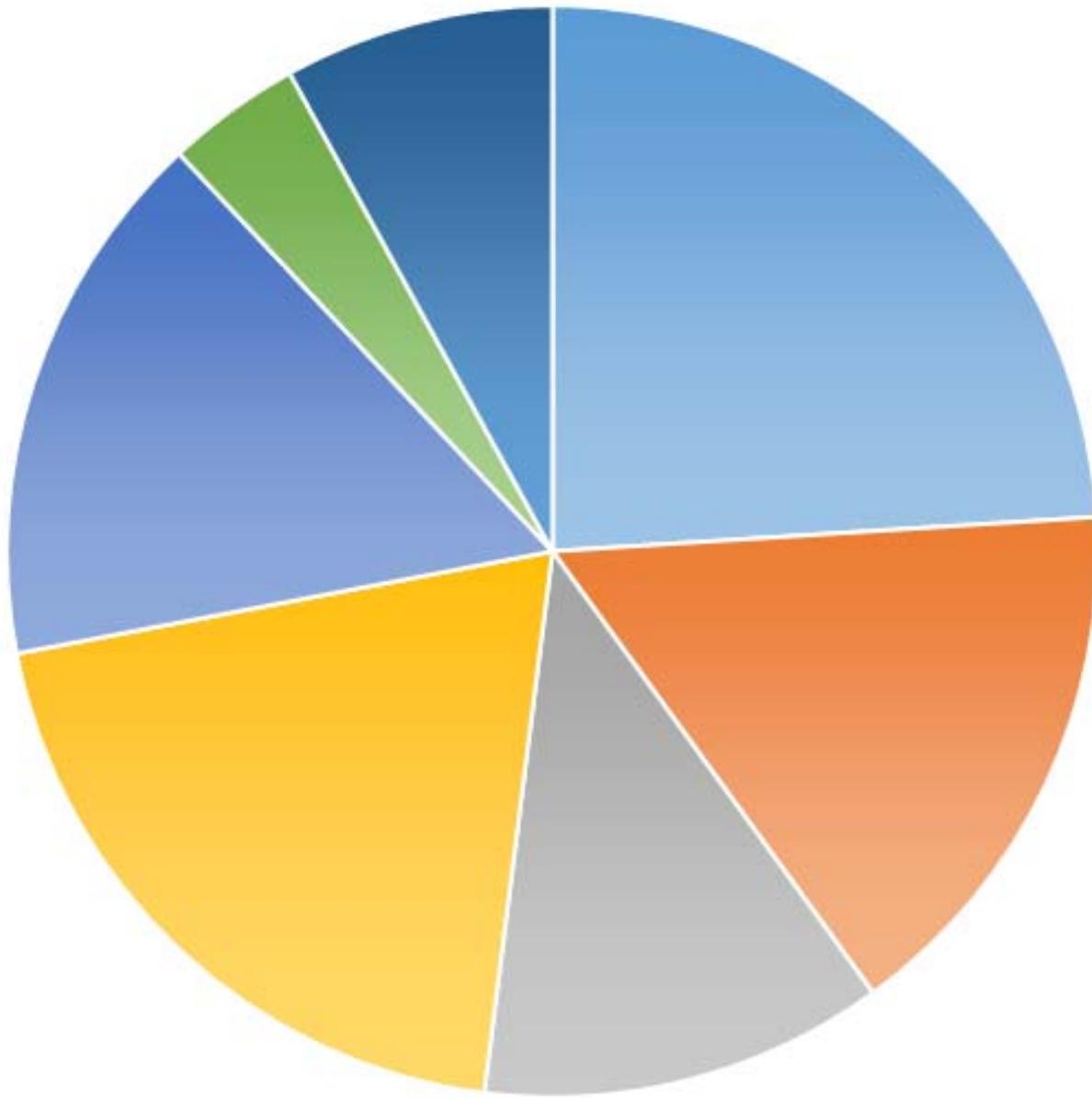
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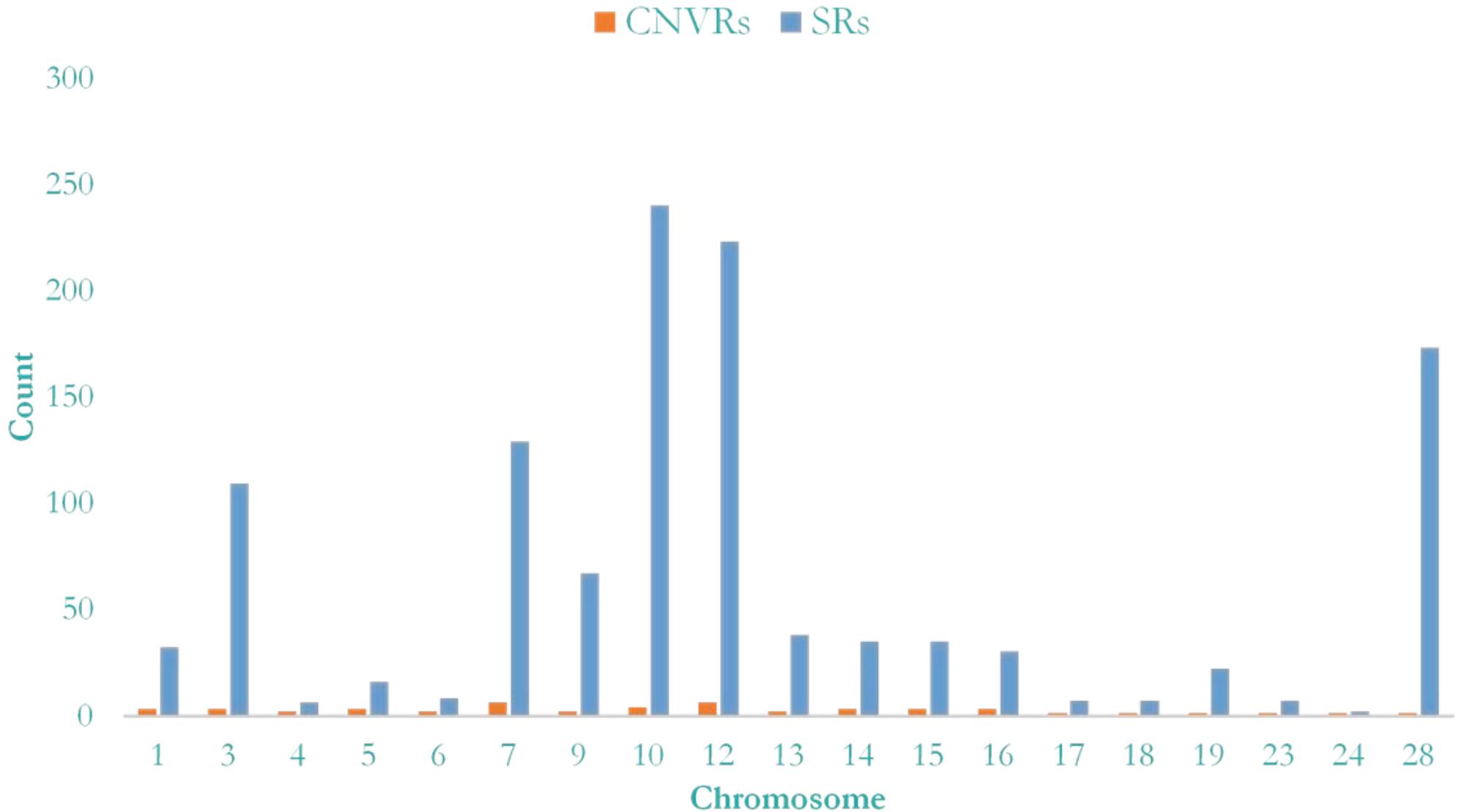
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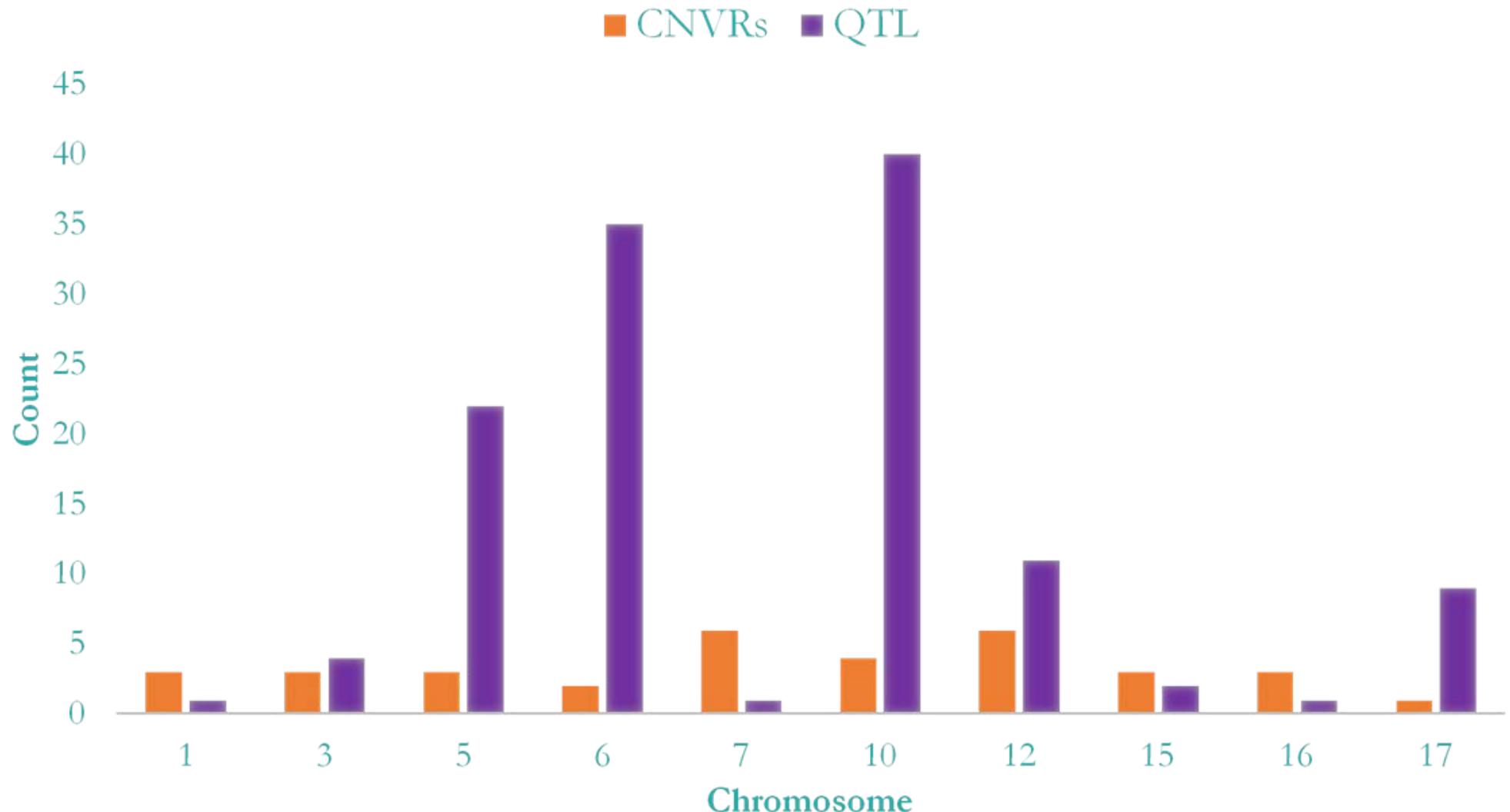
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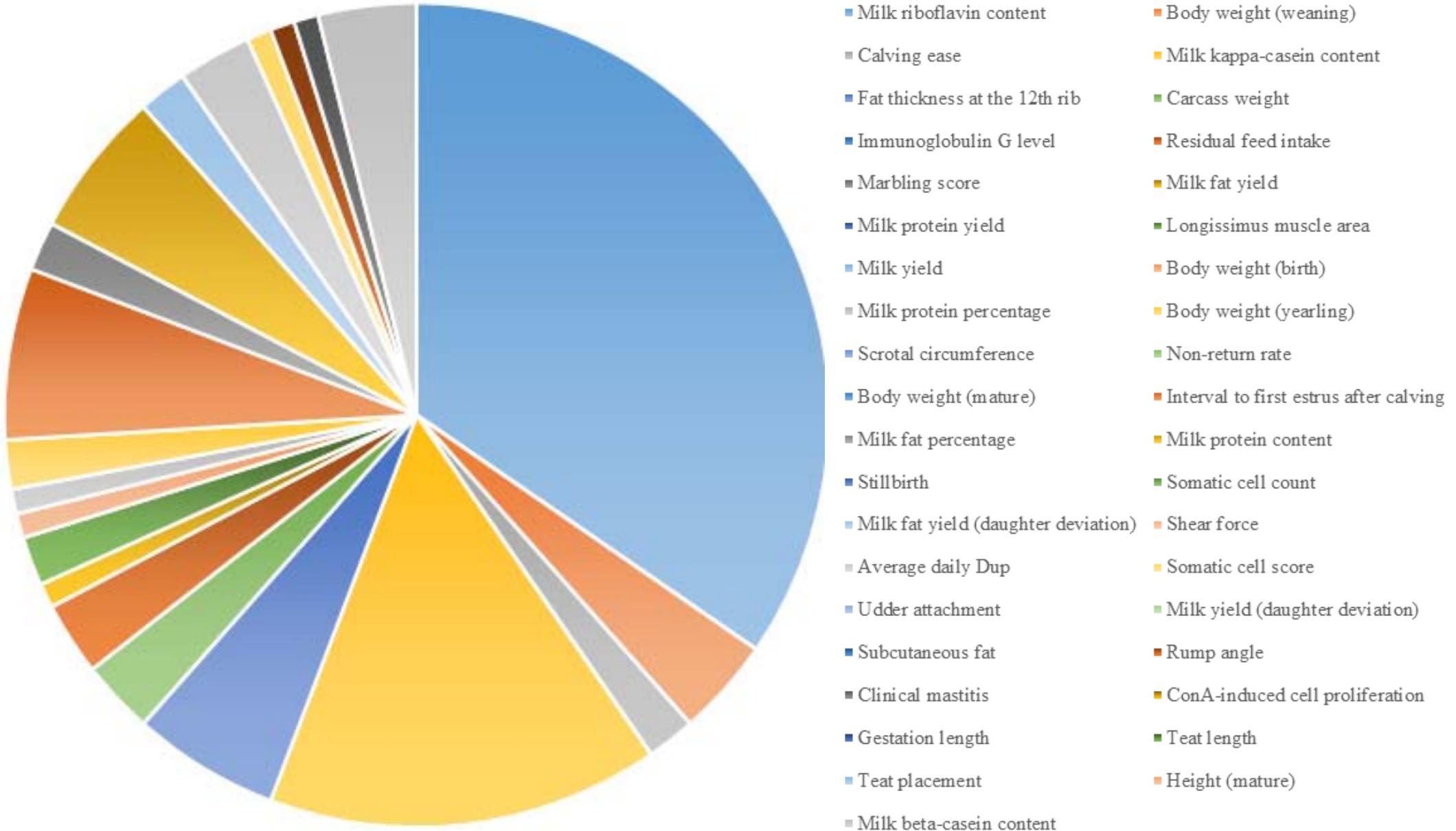




- 1 response to stimulus (GO:0050896)
- 2 immune system process (GO:0002376)
- 3 cellular process (GO:0009987)
- 4 metabolic process (GO:0008152)
- 5 biological regulation (GO:0065007)
- 6 cellular component organization or biogenesis (GO:0071840)
- 7 localization (GO:0051179)







# Conclusions & Recommendations



# CONCLUSIONS & RECOMMENDATIONS

**40 RSA Brahman Bulls**



# CONCLUSIONS & RECOMMENDATIONS

## 40 RSA Brahman Bulls



- 51 common CNVRs using Bovine HD beadchip
  - 29 = Jiang *et al.*, (2012), Hou *et al.*, (2012) and/or Zhang *et al.*, (2015)
- Simple repeats, genes and QTLs
- CNVR genes primarily involved in 7 biological processes
- Future studies
  - Formation
  - Inheritance
  - Effect



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Thank you!! 😊

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