

# GPE low coverage sequencing

## Sequenced bulls

Purebred	Bulls	Progeny	Grand-progeny
	<b>167</b>	<b>3409</b>	<b>10619</b>
Hereford	21	568	1526
Angus	21	478	1542
Simmental	19	452	1511
Limousin	19	413	1697
Charolais	19	373	1210
Gelbvieh	19	295	1294
Red Angus	16	276	1169
Salers	7	113	154
Shorthorn	6	112	98
Maine-Anjou	5	81	64
Santa Gertudis	5	76	81
ChiAngus	4	63	128
Braunvieh	3	59	85
Brahman	2	32	54
Brangus	1	18	6

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## Sequence variants observed

	Change rate
34,188,072 total variants	1 per 78 bases
30,687,789 bi-alleleic SNP	1 per 87 bases
1,566,589 deletions	
1,228,660 insertions	
705,034 multi-allelic variants	
217,798 tri-allelic SNP	
487,236 complex	

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## Predicted functional impact

High	6,513
Moderate	89,591
Low	109,171
Modifier	34,022,537
Total	34,227,812

# effects > # variants -  
some variants affect multiple genes

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## Genome coverage

99.6 % of BovineHD SNP detected in bull sequence

1.9x/bull mean coverage of BovineHD positions

## Genotype calls

$P(\text{another genotype}) < .25$

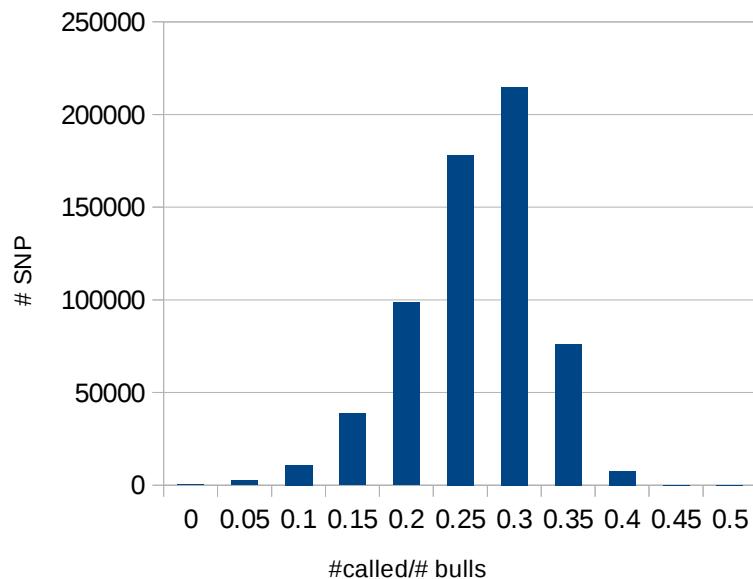
4.6x/bull mean coverage of complete genotypes  
(both alleles)

1.5x/bull mean coverage of partial genotypes  
(one allele)

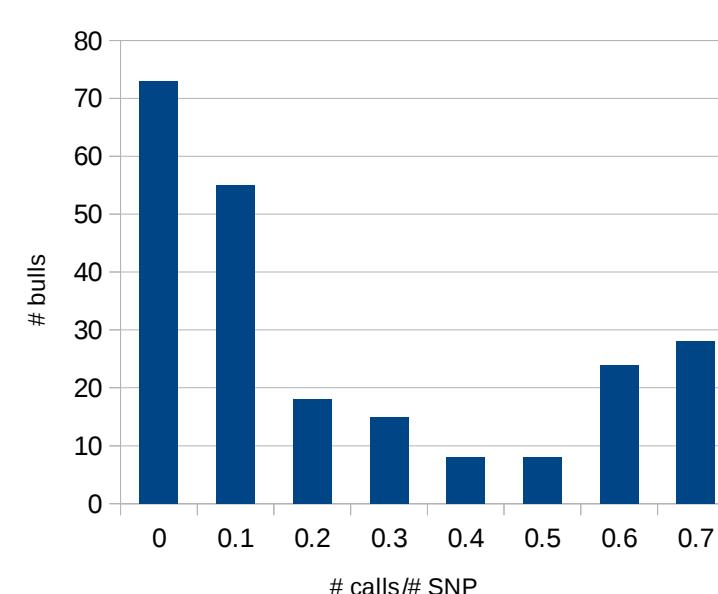
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## Sequence genotype call rates (BovineHD SNP)

28.9% by marker (0 to 83%)



29.3% by bull (0 to 97%)



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## Sequence-BovineHD genotype consistency

BovineHD	# genotypes	matching		opposite		% matching
		homozygous	heterozygous	homozygous	heterozygous	
homozygous	24177845	23182459		924799	70587	95.9
heterozygous	17894735			15796784	2097951	88.3
all	42072580	38979243				92.6

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## Interesting variant genotypes

95,401 High and moderate impact variants

21.8% call rate by bull (0 to 90%)

21.6% call rate by variant (0 to 98%)

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Interesting variant alternate allele frequency

	Bulls	Interesting	High Impact
		AAF	AAF
Hereford	21	0.16	0.21
Angus	21	0.18	0.24
Simmental	19	0.19	0.24
Limousin	19	0.19	0.24
Charolais	19	0.19	0.25
Gelbvieh	19	0.19	0.24
Red Angus	16	0.18	0.23
Bos taurus	220	0.19	0.26
Bos indicus X	12	0.27	0.29

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## Next steps

exome sequence – bulls and progeny

increased depth covering interesting variants

improve call rates, called and imputed genotype  
accuracy

transcriptome sequence (RNA-Seq)

gene expression levels

expressed variants

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## Next steps

RNA-Seq – 16 steers, high/low intake, gain, efficiency  
rumen papillae libraries

	differentially expressed genes	interesting variants bull and RNA-Seq	expressed variants bull and RNA-Seq
gain	9	70	92
intake	15	52	52
efficiency	6	39	55

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## Next steps

explain genetic and phenotypic variation

- need genotypes to determine associations with performance
  - impute sequence variant genotypes with pedigree and 50K/HD genotypes
  - identify 50K/HD haplotypes – estimate effects of haplotypes containing reference/alternate alleles
  - directly genotype specific variants - populations lacking sufficient reference genotypes

