The role of rare variants in genomic evaluation

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Missing heritability

rare variants

- 33.4% of sequence variants have a MAF<0.05
- Neither on the currently available commercial SNP arrays, nor well tagged by the SNPs on the arrays
- Big impact on individual phenotype
- Selection makes causal variants with a low MAF

~5000 Nordic Holstein bulls

~15 million imputed genetic variants

Contribution of rare and low-frequency variants in the variation of different traits

Trait	Variance explained		
	Whole genome sequence	Rare variants	Low-frequency variants
Fertility	0.60	0.00	0.13
Longevity	0.63	0.08	0.14
Health	0.51	0.13	0.12

RLFVs contributed a larger proportion of the explained DRP variances (>13%) for Fertility, Longevity and health traits

Impact of rare and low-frequency variants on reliability of genomic prediction

Marker sets	Fertility	Health	Longevity
50k	0.39	0.32	0.29
50k + All RLFVs from genes (1,600,000)	0.40	0.33	0.28
50k + RLFVs in genes with significant association (30,000)	0.39	0.32	0.28
50k + RLFVs with annotations (26,000)	0.38	0.32	0.26

RLFVs can slightly affect the ranking of bulls in the selection especially the top bulls