## NEOGEN





Euroopa Maaelu Arengu Põllumajandusfond: Euroopa investeeringud maapiirkondadesse





## **NEOGEN Genomics**

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# Who am I?

Helene Hofeneder-Barclay, MSc **Business Development - EMEA Genomics** All Species for Academic and Research Accounts & Equine, Canine and Feline for **Breeders Associations** 

- Based in Ayr, Scotland, UK
- MSc in Molecular Biology from the University of Vienna •
- Working since 1997 in Molecular Biology / Genomics products and • services organisations
- 4 years with Neogen Europe ullet





#### **Short Introduction**

**GGP Arrays** 

Other service Affymetrix, Illumina, Agena (Massarray)

NGS, Whole Genome Sequencing

## **NEOGEN Europe, Ayr**













## **NEOGEN Genomics**

GGP Chips

- GGP Geneseek Genomic Profiler chips
- Customer content
- Proprietary design software
- Multiple species

























## **GGP** Arrays

**GeneSeek<sup>®</sup> Genomic Profiler<sup>™</sup> Custom Arrays** Neogen wants to produce affordable arrays, that have health traits and parentage SNPs

- GGP Bovine 100K– 100k SNPs
- GGP Bovine 150K 150k SNPs
- GGP F250 230K SNPs
- GGP *indicus* 50k and 80k SNPs  $\bullet$
- GGP Porcine 50k SNPs
- GGP Equine 80k SNPs lower price point
- GigaMUGA 150k SNPs
- MiniMUGA 10K SNPs
- Eucalyptus 60k SNPs  $\bullet$
- GGP Ovine 50K– 50k SNPs
- GGP Goat 70K 70k SNPs
- GGP Potato 35K 35k SNPs
- GGP Whiteleg Shrip– 50k SNPs

Chip design based on –

- Unique SNP select software and MOLO (Multiple Objective) Local Optimisation) algorithm.
- **Optimal SNP distribution**  $\bullet$
- Increased amount of SNPs at places where there are increased cross over events (e.g. telomeres)
- Even coverage across all chromosomes
- Inclusion of "must have" SNPs
- High Minor Allele Frequency (MAF) SNPs









## **Key Characteristics of Chip Design**

- Minor Allele Frequency (MAF) or Haplotype Frequency
- Optimal SNP distribution (not all uniform)
- inclusion of "must have" SNPs
- limited number and size of chromosomal gaps



MOLO – Centers on an objective function which maximizes adjusted information for a set of SNPs selected for multiple constraints.

## **GGP Bovine Upgrades**













## **GGP Bovine 50K**

- Covers majority of Bos Taurus and several Bos Indicus breeds
- Based on Angus, Red Angus, Beefmaster, Brangus, Charolais, Gelbvieh, Hereford, Limousin, Simmental, Holstein, Jersey, and Brown Swiss and Taurus x Indicus Cross Breeds
- Includes approximately 16,000 of the most informative SNPs from the original Illumina Bovine SNP50k and More than 44,000 SNPs overlap with the Illumina Bovine HD array
- 39,000 SNP overlap with the GGP Bovine 150K and greater than 12,000 SNPs from the previous version of the GGP-LD array are included
- Host of causative mutations for Dairy and Beef including SNPs that indicate disease susceptibility
- Commonly utilised USDA and ISAG parentage SNPs and hundreds of SNPs to enable conversion of ISAG microsatellite parentage data.
- Breed identification (Holstein, Jersey, Brown Swiss)

## **GGP Bovine 100K**

In addition to the original content from the GGP Bovine 50K, the new SNP content includes:

- 23,000 SNPs additional overlap to CDCB virtual evaluation and consolidates global dairy breeding evaluation
  - 66,000 SNP overlap to Illumina BovineHD
  - 100 to 200 Mitochondrial SNPs

MOLO Algorithm to calculate optimal SNP spacing, best coverage and highes MAF:

- Better Coverage Average SNP spacing of 36.5kb with a higher concentration on the telomeric regions of the chromosome.
- Even Distribution Average D-score of 0.92 and U score of 0.77
- Astounding Minor Allele Frequency weighted average MAF across 10 breeds of 0.3
- Increased accuracy of imputation to 600K array (>99%)
- Indicus, Beef and Dairy Traits





## **GGP Indicus 35K Brazilian Beef Industry**

- 40,092 animals genotyped on Bovine HD and GGP chps
- Pool of 788,879SNPs genotypes from Brahman, Droughtmaster, Guzerath, Gyr, Nellore (including polled Nellore) 69.32%, Santa Gertrudis and Tropical Composite
- 34,000 SNPs were chosen and 1,000 SNPs related to functional mutations and ISAG parentage SNPs were included
- 1,000 Y Chromosome SNPs
- MAF for SNPs used for SNP chip development 0.4318
- Imputation accuracy to Illumina Bovine HD in Nellore cattle 98.7%
- Reference: J.B.S. Ferraz, X. Wu, H. Li, J. Xu, R. Ferretti, B. Simpson, J. Walker, L.R. Silva, J.F. Garcia5, R.G. TaitJr., S. Bauck. Design of a low-density SNP chip for Bos indicus: GGP indicus technical characterization and imputation accuracy to higher density SNP genotypes.

## **GGP Indicus 50K Australian Beef Industry**

- Adding 20,000 additional SNPs to provide improved chromosomal coverage based on ARS 1.2 genome assembly
- MOLO to find the best SNPs from Illumina HD 770K to facilitate future imputation and research work and reduce gap sizes on each chromosome
- Currently imputes to 770K >96% accuracy for many Indicus breeds including cross influenced indicus breeds
- Improved overlap to Illumina 7K and previous Illumina Consortium Chip
- Added content for Holstein x native Indicus cross cattle
- Added dairy traits and conditions
- SNPs from Australian research work on genomic regions influencing economically relevant traits in indicus-taurus composite animals
- Application for synthetic breeds, like Girolando, Brangus, Braford and Composite cattle, like Montana
- Today we are delivering A2 results and can report milk protein markers **NEDGEN** from this chip





## GGP Equine V4 (80K) - Launch in Jan/Feb 2021





- **GGP Equine** upgraded to ~80,000 SNPs
- Add-on content Causative mutations
  - Health Traits, Coat Colours, Performance
  - Markers
- Parentage SNPs, ISAG Core Panel and Warmblood 200SNP panel
- Improved coverage of the Equine genome and increases average Minor Allele frequency
- Improved imputation to Affymetrix Equine HD
- Y and Mitochondrial Marker addition













### • **GGP Sheep** – upgraded to 50,000 SNPs

- Adding 35,000 additional SNPs to provide improved chromosomal coverage
- All SNPs from the Ovine HD (600K) chip
- Includes all 15,000 SNPs from the GGP Ovine LD chip
- Includes Parentage SNPs, Scrapie, Myostatin, Fertility Markers, ....













#### • GGP Goat 70K

- Overlap with the GoatSNP50 Illumina beadchip is around ~40,000
- Remaining 30,000 SNPs of content that does not overlap were selected to segregate in both fiber and dairy goat breeds with whole genome sequence data on nearly 300 animals
- Includes Parentage SNPs and Scrapie







## **Other Services**

#### **Platforms:** Illumina:

- Genotyping service at competitive prices on all commercially available Illumina chips.
- Neogen's own optimized GGP range of Illumina chips that combine genotyping for research purposes with parentage and health traits.
- Access to Consortia chips.

#### **Affymetrix:**

- Genotyping service at competitive prices on all commercially available Affymetrix arrays.
- Access to Consortia arrays.

#### **Custom Panels:**

Depending on SNP number and sample count we can develop any panel on an Agena, Illumina or Affymetrix platform.







## Whole Genome Sequencing

- WGS with Reference Genome Deliverables: FastQ and aligned BAM files
- De Novo Sequencing Deliverables: FastQ file
- Targeted Sequencing Still trialing different targeted GBS approaches and Amplicon Sequencing approaches.
- Skim Sequencing: 1x WGS and imputation thereafter Deliverables: FastQ file, imputed Variant Call File (vcf), imputed fixed array content. BAM file also possible.











Low-pass skim sequencing & imputation tool from NEOGEN® Genon

#### variant (SNP) discovery

• same cost and effort to sequence many individuals at low coverage as few individuals at high coverage

#### using low pass sequence data to genotype

- Relatively low direct call rate for any specific SNP
- imputation match low-coverage reads to *well characterized* reference haplotypes





	Concordance (Angus)	Concordance (Hereford)	Concordance (Simmental)
<b>4</b> x	99.8%	99.7%	99.8%
<b>1</b> x	99.5%	99.4%	99.5%
<b>0.4x</b>	99.3%	99.1%	99.2%

High concordance between imputed genotypes and fixed GeneSeek arrays in *B. taurus* breeds



## Skimseq pilot project – w/imputation

>50M variants identified

>1M *interesting* variants –

- 11K loss-of-function 1)
- 2) 220K nonsynonymous

#### SNPs

600K possible 1) regulatory variants

# SkimSEEK







# SkimSEEK



## Deliverables and timelines

- Raw sequence data (fastq)
- Variant call file (vcf)
- Fixed panel data report (e.g. GGP Bovine 100K)
- Available today!

Questions: hhofenederbarclay@neogen.com





## Why Neogen

#### **TRUSTED LEADER**

NEOGEN Genomics has the worlds most innovative animal genomics laboratories with unparalleled capacity and flexibility.

#### **ELITE TECHNOLOGY**

Our chips include custom content for highly accurate predictions.

#### **QUICK TURN AROUND TIME**

Time is money and we strive to get every sample through our lab in 21 days.

#### **SUPERIOR SUPPORT**

Programs like Igenity Dashboard and personal assistance from the NEOGEN customer service team are there to help you every step of the way. Support is also provided through training and presentations















## Thank You For Listening

#### **Get In Touch**

www.NEOGEN.com

