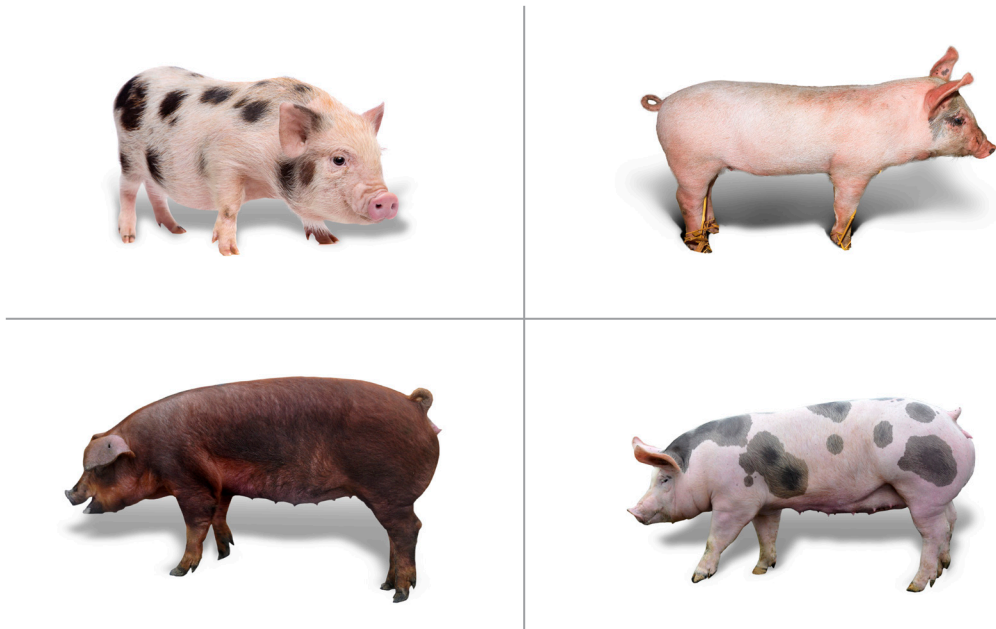


## Axiom<sup>®</sup> Porcine Genotyping Array

Powered by Axiom<sup>®</sup> Genotyping Solution, the array offers the highest data completeness, call rates, and reproducibility of any technology available today.



### Highlights

- Expert design: developed by key opinion leaders in the porcine community
- High resolution
  - 658,692 markers on the array
  - Equal spacing on chromosomes
  - Bias towards common variants
  - 56,000 of the most informative markers from an existing in-market 60,000 marker array, allowing compatibility with previous studies
- High diversity: includes markers from a diverse set of commercially relevant breeds, including rare/traditional breeds

### Applications

- Construction of high-resolution genetic maps
- Genetic improvement of pure lines
- Fine mapping of quantitative trait loci (QTL)
- Development of training populations for calculating breeding values with genomic selection models

## Comprehensive solution for pig genetics

Axiom® Porcine Genotyping Array (Axiom\_PigHDv1) for pig genotyping was designed through Affymetrix' Expert Design Program by Dr. Martien Groenen at Wageningen University, Netherlands, and Dr. Alan Archibald at The Roslin Institute, United Kingdom.

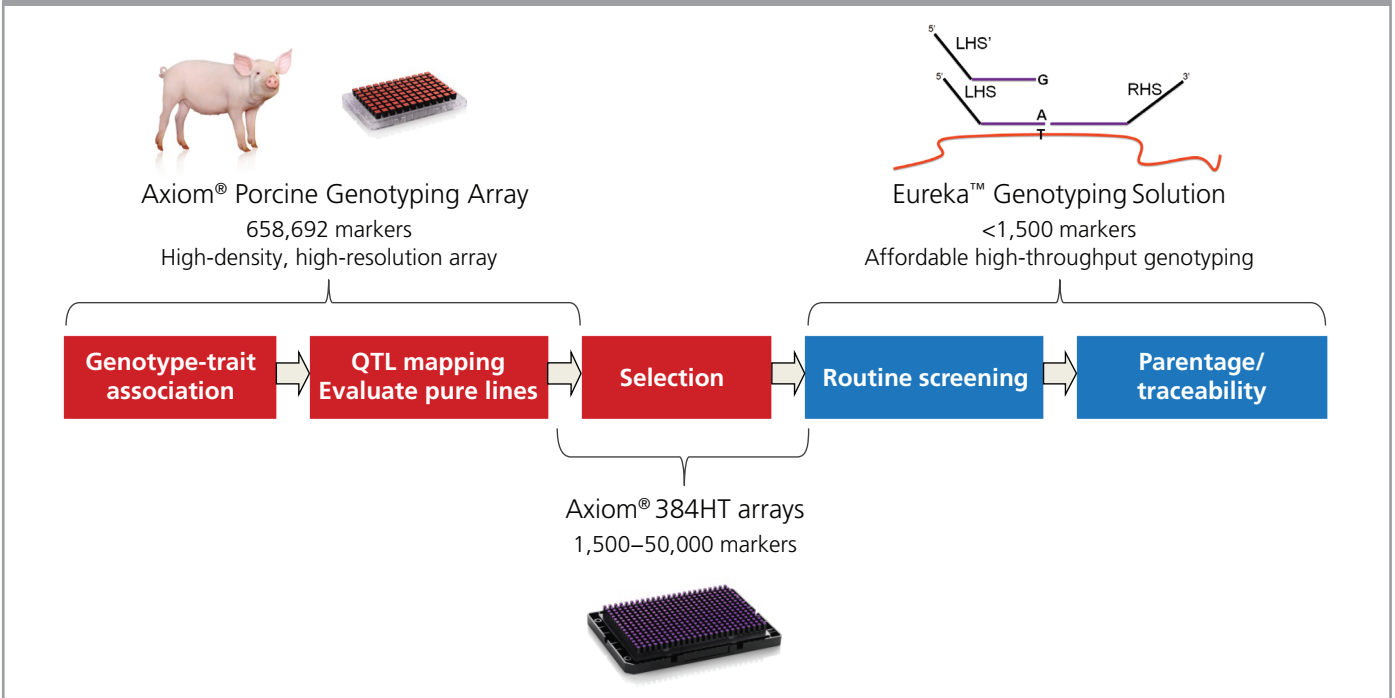
Axiom Porcine Genotyping Array is available in 96-array format and includes 658,692 markers discovered using whole-genome sequence data from 210 animals covering a broad range of commercial and non-commercial pig breeds that were selected for their relevance to world-wide breeding programs.<sup>1</sup>

This high-density array offers the power and resolution for a wide range of applications in pig breeding and genomics that include studying marker-trait association, evaluating pure lines, and identifying multiline reference populations, as well as research applications for genome-wide analysis and selective sweep analysis studies. A key benefit of using Axiom Porcine Genotyping Array is the ability to genotype samples without experiencing marker dropout or missing data, which have been observed when using other porcine genotyping products.

Markers that have been previously validated and associated with desirable traits are very important in maintaining and breeding elite commercial populations. The inclusion of the 56,000 most informative and confirmed markers from the existing in-market 60K array ensures that the new Axiom array can be used for comparison with data generated by previous studies. It also provides the ability to continue existing projects, while making use of the latest and most informative content, to extend the usefulness of the study.

The high-density array can be used to analyze samples from different populations and transfer markers of interest to Axiom® 384HT myDesign™ Custom Array. The lower-complexity Axiom 384HT custom arrays can be designed in less than six weeks and allow you to select a subset of markers of most interest to you. Axiom 384HT arrays can help achieve higher imputation accuracy in genomic selection in breeding programs at an extremely economical price point. The array manufacturing technology from Affymetrix guarantees 100% fidelity when transferring markers from the high-density array to the Axiom 384HT custom array.

**The continuum of the genotyping solution for pig genetics from Affymetrix.** Axiom® Genotyping Solution with the Axiom® porcine high-density array is ideal for applications such as genotype-trait association, QTL mapping, and pure line evaluation. In routine use applications, such as genomic selection in routine breeding where higher throughput is required, the Axiom® 384HT format offers the capability to genotype 50,000 markers and many thousands of samples per week. Applications such as traceability, parentage, and identification of recessive alleles can leverage the sequencing-based Eureka™ Genotyping Solution at a much higher throughput across targeted loci.



Genotyping is performed using Axiom™ Analysis Suite software. The convenient 96-array format and the one-click analysis of Axiom Analysis Suite software provides the least amount of hands-on-time for genotyping, minimizing costs and time to results.

## Array design

The markers on the array were identified from whole-genome data from 210 pigs sequenced at an average depth of 10X. After alignment against build 10.2 followed by single nucleotide polymorphism (SNP) calling and filtering, a total of 5.6 million putative markers were submitted to Affymetrix to calculate *in silico* design scores. Table 1 provides an overview of breeds from each group used for marker discovery.

All markers that were selected for synthesis on the array were prioritized using the following criteria: 1) High *in silico* design score; 2) Equal spacing along chromosomes; 3) Markers on unassigned contigs from build 10.2; 4) High minor allele frequency (MAF) >0.1 in European and Asian breeds; 5) Exclusion of any AT or GC markers that would have required more than one probe per marker; 6) Inclusion of 56,000 markers from the in-market 60K porcine array. A total of 658,692 markers were selected for synthesis on the array.

**Table 1: Major groups and the associated breeds used in discovery of markers.<sup>1</sup>**

Grouping	Breeds	Number of samples sequenced for marker discovery
European domestic, commercial	Large White, Landrace, Pietrain, Duroc, Hampshire	80
European domestic, rare/traditional	Angler Sattelschwein, British Saddleback, Bunte Bentheimer, Casertana, Cinta Senese, Gloucester Old Spot, Large Black, Linderodssvin, Mangalica, Middle White, Negro Iberico, Retinto, Tamworth	45
European wild boar	13 populations across Europe	38
Asian domestic	Jiangquhai, Jinhua, Leping Spotted, Meishan, Thai Native, Wannan Spotted, Wuzishan, Xiang, Zang	25
Asian wild boar	Breeds from China, Japan, Thailand	22

## Automated genotyping and classification

Axiom Porcine Genotyping Array was evaluated with samples from multiple commercial and domestic breeds including European wild boars to demonstrate the array performance across the different major groups. A total of 95 samples were processed and analyzed using Axiom Analysis Suite software, as per the *Axiom® Genotyping Solution Data Analysis Guide* (PN 702961 Rev. 3). The breeds and the number of samples representing the five different groups are shown in Table 2, along with the average MAF and approximate number of polymorphic loci for each group. A total of 532,130 markers (80.79%) were automatically identified as high-quality markers under the polymorphic high-resolution category. The call rate of markers in this category was greater than 99%. Of the 95 samples included in the evaluation, 63 had been sequenced previously. These previously sequenced samples were used to perform concordance analysis. The results from the data analysis are shown in Table 3. The data was automatically clustered, assigned genotypes, and classified into six categories for easy visualization. The resulting genotypes demonstrated high concordance with sequencing data and high call rates.

**Table 2: Samples and breeds used in the evaluation of Axiom® Porcine Genotyping Array.**

Grouping	Breeds	Number of samples	Average MAF	Approximate number of polymorphic loci
European domestic, commercial	Berkshire, Duroc, Landrace, Large White, Hampshire, Pietrain,	40	0.32	590,000
European domestic, rare/traditional	Angler Sattelschwein, British Saddleback, Bunte Bentheimer, Casertana, Cinta Senese, Gloucester Old Spot, Large Black, Linderodssvin, Mangalica, Middle White, Negro Iberico, Retinto, Tamworth	25	0.28	635,000
European wild boar	Nine different populations	18	0.30	600,000
Asian domestic	Meishan	4	0.72	400,000
Other	Nera Siciliana, Large White x[(LWxMeishan) x(LWxMS)] sire X Large White dam, <i>Sus cebifrons</i> , <i>Sus barbatus</i> , <i>Sus philipensis</i> , <i>Sus celebensis</i> , Large White x[(LWxMeishan)x(LWxMS)]	8	0.71	405,000

**Table 3: Results from Axiom® Porcine Genotyping Array assigned into six different categories.**<sup>1</sup> The third column displays the concordance observed between genotypes from sequence data and the Axiom array for each of the six categories. The markers in the recommended categories include 1) **PolyHighResolution markers**: markers demonstrating three clusters with good cluster resolution and at least two examples of the minor allele; 2) **NoMinorHomozygous markers**: markers exhibiting two clusters with no examples of the minor allele; 3) **MonoHighResolution markers**: markers demonstrating a single cluster; 4) **OffTargetVariant markers**: includes reproducible yet uncharacterized variants caused by double deletion, sequence non-homology, or DNA secondary structure.

SNP classification	Number	Percentage (%)	Concordance with sequencing (%)	Performance of markers from legacy 60K array (%)
<b>All markers</b>	658,692	100	93.3	
<b>Recommended markers</b>				
<b>PolyHighResolution</b>	532,130	80.79	97.6	73.7
<b>NoMinorHomozygous</b>	6,060	0.92	91.8	2.3
<b>MonoHighResolution</b>	4,551	0.69	56.4	3.8
<b>OffTargetVariant</b>	3,780	0.57	73.7	1.8
CallRateBelowThreshold (<97%)	40,261	6.11	93.3	6.0
Other	71,910	10.92	78.9	12.4

## References

1. Groenen M. A. M., Development of a high-density Axiom® porcine genotyping array to meet research and commercial needs. Plant & Animal Genome XXIII Conference, San Diego, CA (2015).

## Ordering information

Part number	Description	Details
550588	Axiom® Porcine Genotyping Array	Contains one plate with 96 arrays. Reagents and GeneTitan® MC consumables must be quoted separately
901606	Axiom® GeneTitan® Consumables Kit	Contains all GeneTitan® Instrument consumables required to process one array plate
901758	Axiom® 2.0 Reagent Kit	Includes all reagents (except isopropanol) for processing 96 DNA samples

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