The revolution in PRE equine genomics: new Axiom EQUIGENE medium-density array

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INTRODUCTION

The **Axiom EQUIGENE SNP array** (ThermoFisher) enables a wide range of applications in equine genomics, including **parentage analysis, heritable diseases detection, and analysis of economically important traits**, as well as **genomic selection** research. Developed under the GO EQUIGENOM innovation project, funded by the Spanish Ministry of Agriculture, Fisheries and Food, this optimized medium-density array is designed for the Purebred Spanish Horse (PRE) and related breeds.

MATERIALS AND METHODS

Animals genotyped and sequenced

4,490 representative horses (1,907 males and 2,583 females) belonging to 1,718 stud farms were selected for **genotyping**.

284 PRE horses were **sequenced** to add Y chromosome and mitochondrial markers.

Imputation analysis

A reference population of animals genotyped at high density (HD) (2,359) and medium density (MD) (1,781) were used to select the markers that best imputed at HD and MD.

Genomic markers related to traits of economic importance

GWAS analysis was implemented for the search of SNPs associated with important diseases for PRE. Data from animals diagnosed and genotyped with HD (867), MD (738) arrays, as well as from animals with whole genome sequencing (284) were used.

All the **markers related to** traits of economic importance such as **diseases, coat colour, conformation, sportive performance…** that have been published in scientific journals or in international databases (OMIA, Ensembl genome browser, ISAG…) were added.



RESULTS

The Axiom EQUIGENE SNP array includes 90,926 SNPs markers, evenly distributed across the equine genome with an average distance of 26 kb (between 15k and 35k), mapped to the EquCab3 reference genome.

Figure 1. *Total number of SNPs distributed on each chromosome*

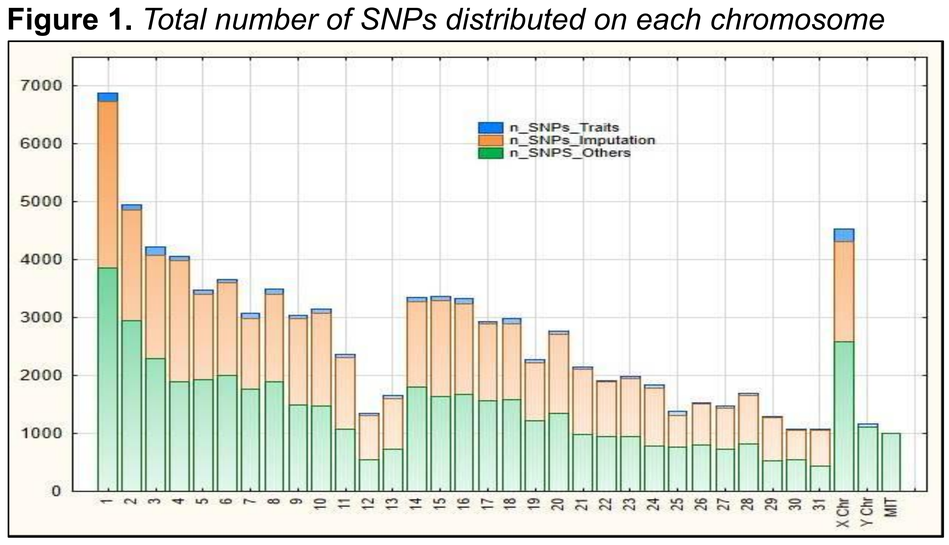

Table 1. *Total number of SNPs distributed on each chromosome*

SNPs related to	number SNPs
Imputation: more informative, useful and high-performing SNPs from existing arrays of different densities	26,017 for HD imputation and 15,705 for MD imputation
Parentage testing	861 SNPs recommended by ISAG
ECAY markers and SRY region	1,115 + 50
PAR region markers	136
Mitochondrial markers	1,000
Traits	1,240
GWAS analyses for diseases	257

Table 2: *SNPs markers related to traits of economic importance in the horse*

TRAITS	Examples	SNPs
Diseases, malformations, and hereditary defects	**Monogenic diseases, Polygenic diseases, Hereditary syndromes, X-linked disorders…** **Musculoskeletal Diseases**: Degenerative joint disease, Congenital Myotonia, Immune-Mediated Myositis, Myopathies, Malignant hyperthermia, Navicular disease… **Integumentary System Diseases (Skin, Hair, Mucosa, Hooves):** Incontinent pigment, Overo Lethal White Syndrome, Hoof wall separation disease, Naked foal syndrome, Fragile Foal Syndrome, Equine anhidrosis, Junctional Epidermolysis Bullosa, vitiligo … **Malformation and defects**: Occipitoatlantoaxial Malformation, Prognathism… ❑Up to 50 types	447
Coat Colour	**Grey coat colour** **Basic coat colours:** Bay, Black, Chestnut **Dilutions:** Cream (Palomino, Buckskin, Perlina, Cremello), Pearl, Snowdrop, Sunshine, Dun, Champagne, **White Patterns:** Dominant White, Sabino, Roan, Overo, Leopard complex, Macchiato, Appaloosa, Tobiano, Splash White... ❑Up to 21 types	63
Conformation	Body Size…	9
Performance	Speed, endurance, Racing performance, Racing distance, Gait laterality, trot gait (rhythm and stride pattern), jumping ability…	66
Behaviour	Temperament, manageability, reactivity to stimuli, docility, intelligence, trainability,…	62
Other productive characters	Molecular markers associated with fertility, Impaired acrosomal reaction, Androgen insensitivity syndrome…	40

Advantages and Benefits of Using the EQUIGENE Array: High resolution and precision, improved genetic selection, early diagnosis of hereditary diseases, development of genomic selection in horse breeds (improve the **accuracy of genetic values** and **increase the reliability** of evaluations in animals at early ages...), **wide applicability across breeds and populations.**

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