A fine genetic analysis of congenital diseases in pig

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and sperm, with a tendency for upregulation in IAR stallions, and suppression of A-alleles in AC and AG heterozygotes. Two novel FKBP6 splice variants were found in testis and sperm. FKBP6 was considered as a susceptibility gene for IAR in stallions and a candidate gene for male subfertility in other mammals. A simple genotyping test is recommended for the detection of IAR susceptible individuals among potential breeding stallions. Successful use of sperm as a source of DNA and RNA propagates non-invasive sample procurement for fertility genomics in animals and humans.

Key Words: acrosome, stallion, FKBP6

P5054 DNA methylation analysis in sperm from infertile/subfertile boars. H. Acloque[1], A. Pinton[1], A. Congras[1], C. Delcros[1], F. Vignolles[1], S. Ferchaud[2], J. Riquet*[1], and M. Bouissou-Matet Yerle[1], *[1]INRA, Cell Genetics Laboratory, Castanet-Tolosan, France, [2]INRA, UEICP, Ruillé, France.*

Male infertility is an increasing health challenge for our societies, either for human or livestock's populations. As diffusion of the genetic progress goes through sires, male infertility consequently slows the improvement of animal selection schemas and farms' productivity. In the french pig sector, more than 35% of boars selected on agronomical criteria and to be diffused through Artificial Insemination Centers (AICs) are culled for bad sperm quality (including concentration, morphology and motility). We propose here to study the epigenetic marks in the sperm and somatic DNA from fertile and subfertile/infertile boars at two different levels. First, we started the production of genomewide methylome maps from fertile boars. Then, taking advantage of our collection of sperm and somatic cells from fertile and infertile boars, we compared DNA methylation levels at specific loci reported to be altered in infertile humans. Combining these studies, our aim is to define specific or common epigenetic signatures of farm animals' infertility. These epigenetic signatures will be an additional parameter to evaluate the sperm quality and will finally help to lower the number of potentially infertile males introduced in the selection schemas. Moreover acquisition of epigenetic information on germ cells may help to introduce epigenetic factors within genomic selection models.

Key Words: infertility, epigenetics, sperm DNA methylation

P5055 A fine genetic analysis of congenital diseases in pig. S. Rousseau[1], N. Iannuccelli[1], E. Pailhoux[2], B. Servin[1], and J. Riquet*[1], *[1]INRA, Cell Genetics Laboratory, 31326 Castanet-Tolosan, France, [2]INRA, Developmental Biology and Reproduction, 78352 Jouy-en-Josas, France.*

The most important congenital genetic defects that occur in piglets are hernias (umbilical hernia and inguinal or scrotal hernias), cryptorchidism and splay legs, and to a lesser extent intersexuality, hermaphrodism and anal atresia. They affect on average 3% of the commercial pig populations worldwide. It is important to stress out that, besides the direct economic loss, these defects have a serious impact on animal welfare and health. Some of the defects cause direct piglet mortality, while others lead to culling of piglets showing the disorder. For most of these defects there are strong indications for a genetic component. In SwAn project, funded by ANR (the French national agency of research) we propose to focus on the identification of genes underlying congenital / hereditary disorders in pigs. We propose (1) to construct a collection of affected samples obtained in collaboration with private breeding companies, (2) to use genome-wide association studies (GWA) with a SNP panel comprising 60 000 markers covering the entire genome at a density which is appropriate for population-based association studies, (3) to identify some of the causal mutations and genes of these congenital diseases and (4) to propose effective marker assisted selection (MAS) against genetic defects. First results obtained for intersexuality, scrotal hernia and cryptorchidism will be presented.

Key Words: pig, GWAS, congenital diseases

P5056 Genome-wide association study for cryptorchidism in dogs. X. Zhao, S. Onteru, D. Garrick, and M. Rothschild,* *Dept. Animal Science, Iowa State Univ., Ames, IA, USA.*

Cryptorchidism is a condition in which one or both testes fail to descend into the scrotum. The incidence in purebred dogs varies between 1 and 14%. The mode of inheritance of cryptorchidism is not known. To determine genes or genome regions responsible for cryptorchidism, DNA samples from 204 male Siberian Husky dogs (105 cases and 99 controls) collected from USA, Canada and UK were genotyped with the CanineHD BeadChip. The K-means clustering algorithm (K = 2) was applied to a difference matrix based on additive genetic correlations to separate dogs into 2 clusters to reduce population structure. Case-control analyses were performed separately for each cluster using PLINK and Bayes-B in Gensel. A 0.85Mb region on CFAX accounted for 1.06% of the genetic variance in cluster 1 and contained several genes involved in extracellular matrix (ECM) remodeling. It is known that ECM remodeling of the gubernaculum is involved in testicular descent. A genomic region covering 2Mb on CFA27 explained 3.21% of genetic variance in cluster 2. Fifty genes including 16 members of Keratin subfamily are located in that region. It appears that mutations on CFA27 and