

OP-APR13-A15**Genetic screening in the clinical suspicion of autoinflammatory diseases**

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Autoinflammatory diseases (AIDs) are unprovoked activation of the innate immune system in the absence of autoantibodies, auto-reactive T-cells and pathogens. AIDs are characterized by recurrent inflammatory episodes and more than 30 genes are found to cause hereditary autoinflammatory diseases, to date. The most common AID is familial Mediterranean fever which is endemic to Mediterranean region. This heterogeneous group of diseases generally manifests overlapping signs and symptoms making diagnosis difficult in the clinic. Also, more than 50% of patients do not carry any pathogenic variant in known disease associated genes indicating complex inheritance affected by both genetic and environmental factors. In routine practice, mutation screening is performed mostly by hybridization based strip assays or as a gold standard technique: Sanger sequencing. In the last ten years, great advances in the sequencing technology enable clinical geneticists to screen more than one gene at the same time. Next generation sequencing (NGS) technology allows to screen many genes causing similar diseases in a short time. Thus, we are now able to detect disease causing mutations and eventually uncovering the genotype-phenotype relationship more effectively. We used multi-gene NGS panels to screen patients with AIDs and the data will be presented in the meeting.

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OP-APR13-B12**Human bone morphogenetic protein (hBMP)-2 characterization by physical chemical, immunological and biological assays**M.F. Suzuki¹, J.E. Oliveira¹, R. Damiani², E.R. Lima², K.C. Amaral¹, F.M. Silva², P. Bartolini^{1,*}¹ *Biotechnology Center, IPEN-CNEN/University of São Paulo*² *Biosintesis*

Commercial preparations of human-met-BMP-2 (GenScript) and of CHO-derived hBMP-2 (Infuse-Medtronic) provided a complete characterization of this protein, which belongs to the “transforming growth factors β ” superfamily, via SDS-PAGE, Western blotting, reversed-phase HPLC, high-performance size-exclusion chromatography and MALDI-TOF-MS.

E.coli-derived met-hBMP-2 has shown a large presence of dimer (MM = 26,054 Da), versus a theoretic value of 26,072 Da. More complex was the distribution of the CHO-derived product, whose exact MM has never been reported due to variable glycosylation: via MALDI-TOF-MS a dimer (28,732 Da) and a large amount of monomer (14,377 Da) were found. A novel method based on RP-HPLC was also validated for hBMP-2 qualitative and quantitative analysis directly in ongoing culture media. The classical “*in vitro*” bioassay, via alkaline phosphatase induction in murine myoblastic cells C2C12, confirmed that hBMP-2 bioactivity is mostly related to the dimer, being ~6-fold higher for the CHO-derived glycosylated form.

Considering that hBMP-2 is a highly effective osteoinductors, plays an important role during bone regeneration and repair, as

well as during embryonic development, and presents an extremely high aggregate value, we believe that these data pave the way to the characterization of this important factor when obtained by DNA recombinant techniques in different host cells.

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OP-APR13-C11**Challenges and upcoming developments in sperm sexing for cattle industry**D. Tapaloaga^{1,*}, P.R. Tapaloaga²¹ *Department of Animal Production and Public Health, University of Agronomic Sciences and Veterinary Medicine, Bucharest, Romania*² *Department of Animal Reproduction, University of Agronomic Sciences and Veterinary Medicine, Bucharest, Romania*

Artificial insemination (AI) was the most commonly used assisted reproduction technologies in livestock, revolutionizing the animal breeding industry during the 20th century. Nowadays, it also allows for faster and increased genetic improvement, allowing for improved herd performance and productivity. In contrast to medical use, where intra-uterine insemination is used in fertility treatments, AI is by far the most common method of breeding in intensively kept domestic livestock, such as dairy cattle (approximately 80% in Europe and North America), pigs (more than 90% in Europe and North America) and turkeys (almost 100% in intensive production) and it is continuously developing, being linked for example with sperm sexing technology. The other assisted reproduction technologies in animals, are generally confined to specialist applications or for research purposes, since the cost would be prohibitive for normal livestock breeding. That is why this study has as aim to encourage breeders to adopt the technology for better livestock production and reproductive efficiency of their livestock. The study has shown that, despite the fact that the basic principles of AI are the same, there is wide variation in the uptake of this biotechnology and the economic aspects should be deeply considered in all the cattle industry.

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OP-APR13-C12**Development of genomics in Belarus**

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Genomic studies in Belarus are carried out in several directions: structural and functional organization of genomes; medical and sports genetics; development of genomic technologies to improve the plant and animal breeding process and environmental protection activities. For Belarus's population, the influence of genetic factors on the formation of major socially significant diseases has been studied and research on longevity nutrigenomics and genomics is conducted. Studies on the search for genetic determinants of economic traits are conducted along with the DNA certification of the varietal gene pool of plants and livestock of animals to increase breeding efficiency. DNA marker sets for the marker-assisted selection of 16 crops (wheat, triticale, rye, potato, tomato, lupine, pepper, cabbage, canola, sunflower, soybean, flax,