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Conference Report

Abstracts of the 25th International Colloquium on Animal Cytogenetics and Genomics (25th ICACG), 26–29 June 2024, Naples, Italy

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1. Introduction

The 25th International Colloquium on Animal Cytogenetics and Genomics is dedicated to the memory of Dr. James (Jim) Womack, a pioneer in gene mapping, especially in cattle. The meeting opened with an obituary presented by Prof. Penny Riggs, a former student at Texas A&M University (TAMU) and now a professor in the same department.

The meeting was organized into 10 sessions, beginning with General Opening Session 1, which featured three main lectures highlighting the fields of animal cytogenetics and genomics. As expected, among the 83 accepted abstracts for publication, those related to animal genomics were more prevalent than those focused solely on cytogenetics. However, several abstracts combined the two disciplines (Cytogenomics) to provide a deeper understanding of animal genomes and to better identify latent chromosome abnormalities related to fertility. Various genomic approaches were reported in several abstracts, aimed at improving the selection of animals for productive traits, disease resistance, and animal biodiversity.

Given the numerous abstracts on water buffalo (river type), a specific session was dedicated to this species, which is particularly important in Eastern, South American, and Mediterranean countries. Nonetheless, research on a wide range of animal species, including domestic and non-domestic animals, non-mammalian vertebrates, and invertebrates, was also presented. Special attention was given to the posters, which were displayed throughout the meeting. Additionally, 15 of the posters, selected by the chairpersons of the poster session, are presented and discussed on the final day. Five posters received awards. All abstracts underwent peer review, and only a few required corrections or modifications. In conclusion, the colloquium featured 13 lectures (L), 27 oral communications (O), and 43 posters (P). Each presentation was numbered according to the congress program. Special thanks to the editorial staff of the "Biology and Life Science Forum" journal for their assistance with the abstract's review and editing.

2. Dr. James (Jim) Womack Obituary

O1—Gene Mapping Is Good for You!—Remembering Dr. James E. Womack

Penny K Riggs and Womack Lab Former Students
Texas A&M University, College Station, TX, USA; riggs@tamu.edu (P.K.R.)

A true pioneer in the field of comparative animal genomics, Prof. Jim Womack (30 March 1941–13 August 2023) is remembered for his remarkable career, scientific achievements, and mentorship of 50 doctoral students and countless additional graduate students, post-doctoral scientists, and visiting scholars. Jim completed a Bachelor of Science degree at

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5.11. P18—Exploring the Genomic Inbreeding Level in Italian Mediterranean Buffalo Using Whole Genome Sequencing Data

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The water buffalo (*Bubalus bubalis*) is a domesticated species mainly farmed for milk production, with a European population of about 500,000 heads, of which approximately 88% are raised in Italy. Despite its economic importance, little research based on SNP arrays regarding genetic variability and the level of genomic inbreeding in the Italian Mediterranean Buffalo (IBM) breed has been carried out. To efficiently explore the unique characteristics of this breed, we sequenced 24 individuals belonging to 10 different farms in Caserta (Italy), focusing first on autozygosity estimates to infer whole genome selection signatures and possible candidate genes related to milk production. QC was performed with PLINK v.1.9, keeping only variants on autosomes and removing SNPs with call rates lower than 0.95 and MAF < 0.01, with a final dataset of 16,680,804 SNPs. The detection of ROHs was performed with the R package detect RUNS by applying the method "consecutive" and the following parameter settings: the minimum number of consecutive SNPs included was 100; the minimum length of ROH was 500 kb; a maximum gap of 100 kb; a maximum of five SNPs with missing genotypes and up to three heterozygous genotypes were allowed for the ROH to be called. Overall, our findings described genome-wide ROH patterns and identified potential selection hotspots containing genes that can be targets for future studies aimed at improving the performance of this breed.

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5.12. P19—High-Throughput De Novo Sequencing of Laser Microdissected Y Chromosome in the Mediterranean River Buffalo (2n = 50,XY)

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The sequencing and correct assembly of the Y chromosome sequences is still a challenge in mammals. In fact, apart from the pseudoautosomal regions (PARs), the Y chromosome lacks a homologous counterpart in the X chromosome. Therefore, in males, the use of genomic DNA as a template for NGS allows isolating the specific Y sequences only as a difference from the X sequences, with many potential gaps and assembly errors for the contemporary presence of X and Y. To overcome this problem, we present a high-throughput sequencing approach based on the direct isolation of Y-chromosomes by laser microdissection in the Mediterranean river buffalo.

Peripheral blood lymphocytes from 10 buffalo bulls were cultured in vitro for normal cultures. Fixed lymphocytes were spread on a polyethylene naphthalate membrane (PEN), which was attached to a 24 × 60 glass slide and treated for GTG-banding. Ten copies of the Y-chromosome from each bull were laser microdissected and collected in individual PCR tubes for DOP-PCR amplification and labeling. FISH confirmed the specific hybridization

of each Y-probe on lymphocyte metaphases before NGS. Library preparation was performed with the NEB Next Ultra II DNA Library Prep Kit for Illumina. High-throughput sequencing was performed by Illumina technology with the NovaSeq 6000 S4 Reagent Kit v1.5 (300 cycles). Raw data were processed by TrimGalore (v0.6.7), and de novo assembly was accomplished by SPAdes genome assembler v3.15.5. Gene sequences were predicted by AUGUSTUS (v3.5.0).

We generated about 40 Gb (90×) of Illumina short reads. Total assembly length was 2,260,027 bp, with an average GC% of 48.11%. A total of 861 genes were predicted, and 807 of them have a hit-to-reference, 210 are uncharacterized, and around 30 are without description. The total number of microsatellites identified was 552. Variant calling was conducted using the GATK4 pipeline, specifically employing the HaplotypeCaller tool for each sample separately and also in a multi-sample version. The total number of variants across all samples was 25,100.

Our approach yielded valuable insights into the genomic characteristics of the Y chromosome, and our results represent a milestone for the river buffalo.

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5.13. P20—A New Web Tool for Rapid Evaluation of Mammalian Genome Assemblies

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Quality assessment and multiple assembly comparisons are essential steps while assembling new genomes. Many tools for evaluating assemblies typically provide synthetic parameters representing assembly quality or overall features, while others provide long, detailed files where it is not always easy to identify and visualize the regions of correspondence and differences among different chromosome assemblies. A typical example is Quast, which is very effective in finding small as well as large similarities, but has to rely on Icarus to graphically display the mapped similarities.

Here we present a new web tool that uses Quast output to quickly identify and display similarities and differences between the compared assemblies, both in text and graphic modes.

The program uses a combination of PHP and R scripts to setup a web-accessible tool that takes as input one or more alignment results obtained by Quast or other similar tsv files in order to preprocess them and produce the many reports, summary tables, and plots.

The presented tool uses information about the alignment blocks, their start and end, in reference and query coordinates, together with additional annotations to represent the main alignment regions at the chromosomal or sub-chromosomal scale, highlighting similarities and colinearity between compared sequences, points of inconsistency, discontinuities, repeated regions, and interruptions in the assembled sequences. It provides a summary of genome coverage chromosome by chromosome and graphical alignment representations that highlight alignment blocks in detail. The program was developed while assembling the water buffalo genome and was found very handy and informative while evaluating the assembled genome sequence.

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