

Programme & Abstracts

12th International Conference on

Canine and Feline Genetics and Genomics



ICCFGG2024
Helsinki – Finland

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Conference Ethics

The International Canine and Feline Genetics and Genomics Conference encourages scientists to present and discuss preliminary data prior to scientific publication. Presentation of research findings is a major factor for the success of the conference. In order to maintain conference integrity and participation, we stress that information presented here should be treated as preliminary work and out of respect for the authors, should only be used with the authors' permission.

In order to maintain the high quality of the conference we would like to inform all attendees of the following policies:

- We ask that new information obtained at this conference will not be used, for either research or commercial purposes, without permission from the authors.
- The abstracts in the conference program booklet cannot be cited.
- Publication of studies presented here in peer-reviewed journals is of course highly encouraged.

Breaches of confidentiality and common sense ethics will ultimately interfere with the pre-publication free flow of information and discussion expected of our conference and endangers the spirit of the conference.

We all thank you for your understanding and attention to this matter.



Conference committees

Scientific organizing committee

Hannes Lohi, University of Helsinki, Finland
Jennifer Meadows, University of Uppsala, Sweden
Jeffrey Schoenebeck, Roslin Institute, Scotland, UK
Christophe Hitte, University of Rennes, France
Greg Barsh, Hudson Alpha, US
Adam Boyko, Cornell University, US
Eva Furrow, University of Minnesota, US
Leigh Anne Clark, University of Georgia, US
Claire Wade, University of Sydney, Australia
Qinghong (Johnny) Li, Nestle Purina Research, US

Local organizing committee

Hannes Lohi, University of Helsinki and Folkhälsan Research Center
Marjo Hytönen, Folkhälsan Research Center and University of Helsinki
Mikko Peltonen, Faculty of Veterinary Medicine
Nanna Mourujarvi-Rosenlöf, University of Helsinki
Confedent International

Presentation awards

The scientific organizing committee will select six graduate or postdoctoral student talks and posters (3+3) for best presentation awards (250€ each) in the conference.

#2

Novel rare variants found in English Cocker Spaniel with early retinal degeneration

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Poster session 2

Progressive retinal atrophy (PRA) is a hereditary condition observed in dogs, including the English Cocker Spaniel (ECS) breed, where a recessive variant in PCRD accounts for most of the cases.

A male blue roan ECS developed blindness at two years of age. Both parents were tested clear for 26 known variants associated with retinopathies. The retina of both eyes appeared devoid of blood vessels and hyperreflective, leading to the diagnosis of early-onset retinal degeneration. No other clinical signs were detected. The genome of the case and the two parents were sequenced and the polymorphisms, small indels, and large structural variants called compared against the Dog10k database.

We found 24 exclusive indels and SNPs. Of these, two were coding variants heterozygous in the parents and homozygous in the case: one AA exchange and one frameshift, both involving genes with a degree of expression in the retina. Additionally, we detected one non-coding exclusive structural variant.

This study underscores the complex genetic underpinnings of PRA in ECS, highlighting the potential involvement of novel genetic variants beyond PCRD mutations. Genotyping of a large score of controls is necessary to confirm the segregation of either the exclusive variants with the phenotype.

Detection of novel variants is crucial for preserving breed health and assisting breeders confronted with the challenge of producing blind dogs despite their diligent testing efforts.

