



## Animal Genetic Testing Standardization

**Organised by a Standing Committee:**      YES      NO

First meeting of *ad hoc* group developed from Applied Genetics of Companion Animals workshop.

### NEW ISAG WORKSHOP ANNOUNCEMENT Animal Genetic Testing Standardization

Please join us for the inaugural ISAG workshop focusing on the genetic testing of diseases and traits in all species – the **Animal Genetic Testing Standardization**. As the number of genetic tests for diseases, phenotypes and traits in our animal species rapidly increases and more and more direct-to-consumer laboratories come online around the world, like our needs to standardize genetic testing for parentage and identification, the need for standards for diseases and phenotypic traits is becoming critical. Please help the committee to identify the needs of the commercial testing community. We hope to focus on requirements for data sharing, common nomenclature and verbiage for variant identification and reporting, standard criteria for the determination of pathogenicity of DNA variants and more. Please help us identify the common concerns for all species and the specific needs of individual animal groups. All are welcome, expertise is needed, to help launch focused improvements and standards for the genetics of animal health and management!

#### Meeting information

Date: Tuesday, 04 July 2023
Time: 17:30 – 19:30
Number of participants: 56

#### Chair

Name: Leslie A. Lyons (organizer)
Affiliation: Department of Veterinary Medicine & Surgery, College of Veterinary Medicine, University of Missouri, Columbia, MO USA
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## Agenda

<p><b>Animal Genetic Testing Standardization</b>          Chair(s): Leslie A. Lyons (organizer)          Location: Cape Town, South Africa          Date &amp; Time: Tuesday, 04 July 2023 17:30 – 19:30</p>	
17:30	<p><b>Welcome and history of workshop development &amp; need</b>          Leslie Lyons – University of Missouri</p> <ul style="list-style-type: none"> <li>- USA/European statements on Direct-to-Consumer testing for health</li> <li>- Pet genomics medicine runs wild – concerns of accuracy/relevance</li> <li>- American College of Medical Genetics &amp; Genomics criteria</li> <li>- Pathogenicity working group – Bart Broeckx – HCM variants plus</li> </ul>
<p><b>Role of OMIA in Standardization of Animal Genetics</b>          Imke Tammen &amp; Frank Nicholas, University of Sydney (OMIA)          Introduction/summary of scope of OMIA</p> <ul style="list-style-type: none"> <li>- Nomenclature for diseases/traits (synonyms can be listed, links to OMIM and MONDO) linked to unchanging <b>OMIA IDs</b></li> <li>- Standardised nomenclature and IDs for breeds based on Vertebrate Breed Ontology (<b>VBO</b>, OLS: <a href="https://www.ebi.ac.uk/ols4/ontologies/vbo">https://www.ebi.ac.uk/ols4/ontologies/vbo</a>; Ontobee: <a href="https://ontobee.org/ontology/VBO">https://ontobee.org/ontology/VBO</a>; access <b>VBO</b> via API: <a href="https://www.ebi.ac.uk/ols4/help">https://www.ebi.ac.uk/ols4/help</a>)</li> <li>- Standardised nomenclature for variants: unique and unchanging <b>OMIAvariantID</b> linked to coordinates based on Human Genome Variation Society (HGVS) recommendations and European Variation Archive (EVA) ‘rs IDs’ if available</li> <li>- Possibility for further improvement (in collaboration with this ISAG committee)</li> <li>- Clinical significance of variants: proposed to review if ClinVar terminology would be useful and to review what evidence is needed for different categories:             <ul style="list-style-type: none"> <li>o For Mendelian disease (<a href="https://pubmed.ncbi.nlm.nih.gov/25741868/">https://pubmed.ncbi.nlm.nih.gov/25741868/</a>)</li> <li>o For Low penetrance variants and risk alleles (<a href="https://clinicalgenome.org/site/assets/files/4531/clingenrisk_terminology_recommendations-final-02_18_20.pdf">https://clinicalgenome.org/site/assets/files/4531/clingenrisk_terminology_recommendations-final-02_18_20.pdf</a>)</li> </ul> </li> </ul>	
<p><b>World Small Animal Veterinary Association (WSAVA) Concerns</b>          Jerold Bell, Tufts University – WSAVA Hereditary Disease Committee (chair)</p> <ul style="list-style-type: none"> <li>- Vets do not understand what tests is done or to request</li> <li>- Information needs to be better for lay public – breeder</li> <li>- Genetic tests need to be validated within a breed before reporting</li> <li>- Better reporting / counselling is required</li> </ul>	
<p><b>Perspectives from Commercial Laboratories and Comparison Testing</b>          Leanne van de Goor, VHL Genetics</p>	
<p><b>Discussion</b> – identification of tasks, goals, and prioritization</p>	



## INTERNATIONAL SOCIETY FOR ANIMAL GENETICS

**Working groups** - Develop with participants by email

### **Summary of the meeting**

#### **Workshop Intention**

- Develop standards for diseases and phenotypic trait testing
- Improve understanding by clients and registries
- Same interpretations, regardless of commercial service
- Improve data sharing between laboratories
- Requirements for data sharing, common nomenclature and verbiage for variant identification and reporting
- Standard criteria for the determination of pathogenicity of DNA variants
- Identify the common concerns for all species & specific needs of individual animal groups

#### **Discussion Topics**

1. Defined set of alleles / nomenclature positions for presenting phenotypic and disease alleles, maybe parentage too?

#### Follow human and mouse standards

<http://varnomen.hgvs.org/recommendations/DNA/variant/alleles/>)

HGNC database (<https://www.genenames.org/>)

Mouse Genome Informatics (MGI) ([www.informatics.jax.org/](http://www.informatics.jax.org/))

Vertebrate Gene Nomenclature Committee (VGNC, <https://vertebrate.genenames.org/>)

2. Standard verbiage for the presentation of phenotypic and disease traits to be used by all labs
3. Ways to share data across labs
4. Standard requirements for sample submission forms (i.e., vet signature??)
5. Defining which tests should be available
6. How/if to include information about DNA testing methodology in reports
7. Defining the pathogenicity of variants
8. Different priorities for different species
9. If DNA testing for genetic conditions should be included in comparison testing – current conducted for cats
10. WSAVA point of view
11. Try to provide DNA for labs for validation – investigator effort
12. Can genotypes from different labs be collated and presented on a site like OMIA?
13. IPFD DogWellNet efforts – see website: <https://dogwellnet.com/> (Commercial labs should support OMIA as did IPFD!)

#### **Summary of Comments and Suggestions**

- ISAG committee could make recommendations on nomenclature/standardised IDS for disease/variant/breed
- ISAG committee could recommend nomenclature for clinical significance and evidence needed to support causality
- ISAG committee species groups could classify existing variants in regard to clinical significance/ resolve disagreement relating to clinical significance of variants



## INTERNATIONAL SOCIETY FOR ANIMAL GENETICS

Create 3 subgroups to address different issues throughout the next two years (people in attendance at workshop will be invited to join). Subgroups provide reports at ISAG2025 and we discuss at ISAG2025 if a permanent standing committee should be formed/if species specific subgroups should be formed

- **Subgroup 1** - Nomenclature: Recommendations on how to standardize nomenclature for diseases/traits/variants/breeds to allow data sharing. (Lyons et al group established, more welcome)
- **Subgroup 2** - Reporting: Recommendation on what information should be included in reports/ made publicly available and how to improve communication to various end users.
- **Subgroup 3** - Comparison testing: Recommendations relating to comparison testing for causal variants – is it needed/how should it be run
- **Subgroup 4** – Recommendations on how to report clinical significance of variants/identify what evidence is needed for each classification (review usefulness of clin var nomenclature). (Perhaps Dr. Broeckx et al group to lead, others welcome)

### Election of committee members:

#### Other business.

The new Standing Committee will move ahead with the proposed goals including making the necessary arrangements to communicate these goals with the Society and communication with email list.

### New Committee chair

Chair:

Term of service (*add years of first and second term of service*):

Affiliation:

E-mail address:

### New Committee co-chair (optional)

Chair:

Term of service (*add years of first and second term of service*):

Affiliation:

E-mail address: \_\_\_\_\_

*Note: One term runs for two bi-annual conferences (i.e., four years)*

**New Committee members**

Other committee members	First term of service (from year to year)	Second term of service (from year to year)	Email address

**SIGNATURE**

\_\_\_\_\_

**Chair**

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**Duty laboratory (if applicable)**