UCDAVIS VETERINARY MEDICINE
Veterinary Genetics Laboratory

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Case of the Quarter

A VGL research study lead by Dr. Rebecca Bellone identified a *de novo* genetic variant in the microphthalmia-associated transcription factor (*MITF*) gene of a Thoroughbred stallion with splashed white coat pattern. The variant was passed on from the investigated stallion to his nearly all-white offspring that has blue eyes and was clinically confirmed to be completely deaf. The offspring was also compound heterozygote for two dominant white variants in the *KIT* gene (W22/W20). Additionally, the study was able to determine that this novel variant, designated as SW8, originated in the maternal chromosome of the stallion's mother. This study was the first to identify the parental origin of one of the four known *de novo MITF* variants reported in the horse and clinically confirm deafness in one horse with this

SW8 variant.

SW8 has now been added to the VGL's Splashed White test and you can read more about this case on our website.

Explore the hundreds of genetic tests available from the VGL for 20+ species

Communication and Outreach

Horse Genetics

The American Paint Horse Association released a new continuing education course that was developed in collaboration with VGL director, Dr. Rebecca Bellone. This incredible resource explains the basics of genetics and the six main genetic health conditions that affect stock horses. The course is available to subscribers of HorseIQ.

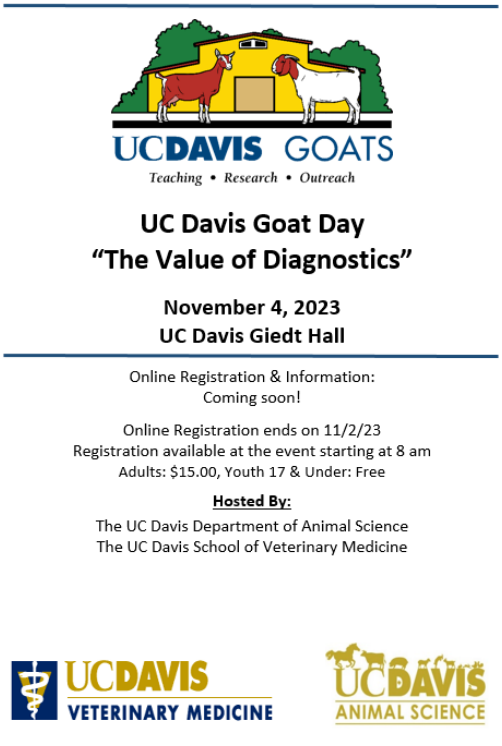
Forensics

In partnership with CalAnimals, VGL's Quality Manager and Forensics scientist, **Christina Lindquist**, led a "Furensics Animal DNA Workshop" to increase awareness of what animal forensics can accomplish. In addition to providing examples of cases that VGL helped investigate, Christina also explained how officers can help

Upcoming Events

UC Davis Goat Day

UC Davis Goat Day is changing dates! The next event will be **November 4, 2023,** and include various talks and demonstrations for goat owners and breeders. VGL will be there to provide information about the available genetic tests and their applications.



grow our Canine CODIS database to help support dog fighting cases.

The Canine CODIS database is similar to the FBI's human CODIS database that is used in criminal and missing person investigations. The Canine CODIS contains individual DNA profiles from dogs that are seized during dog fighting investigations as well as profiles from unknown samples collected at suspected dog fighting venues. DNA can then be used to identify relationships between dogs and thereby allow officers to expand their investigations to those who breed and train dogs for fighting.

Mammalian Ecology and Conservation

Dr. Ben Sacks from the Mammalian Ecology and Conservation Unit participated in an International Union for Conservation of Nature (IUCN) workshop held in September at the Royal Zoological Society of Scotland in Edinburgh, Scotland, to develop a framework for revising the taxonomy

Research Highlights

VGL researchers have contributed 4 new publications since July.

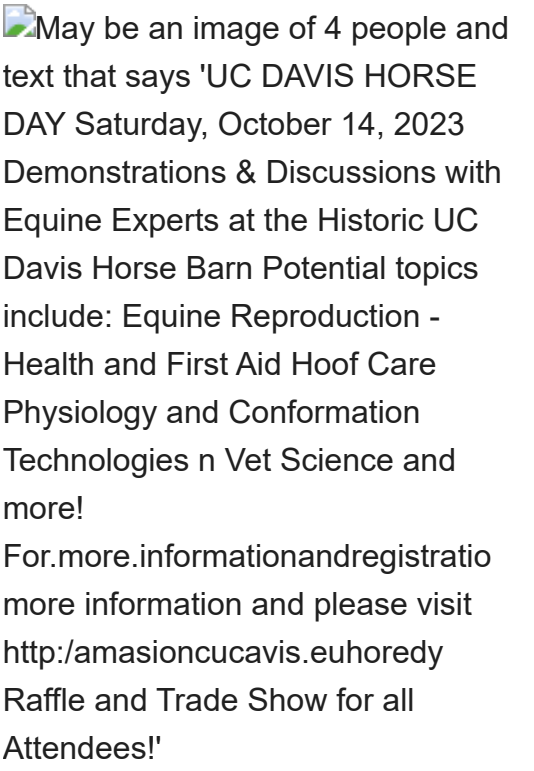
A study by VGL research team led by **Dr. Rebecca Bellone** identified a novel Splashed White variant (SW8) in a Thoroughbred horse. Highlighted in our Case of The Quarter, the SW8 is a *de novo* variant in the *MITF* gene that was passed on to one offspring who had a nearly all-white coat pattern and blue eyes and was clinically confirmed to be completely deaf. This is the first study to identify the parental origin of one of the four known *de novo MITF* variants reported in the horse and clinically confirm deafness in one horse with the variant.

Another study by VGL researchers **Elizabeth Esdaile** and **Dr. Felipe Avila**, led by **Dr. Rebecca Bellone**, provided further evidence that the *CSNB2* allele causes congenital stationary night blindness (CSNB) in the Tennessee Walking

of the Family Canidae, which includes various dog-like species referred to as canids.

UC Davis Horse Day

UC Davis' Horse Day offers a day of continuing education in the form of workshops, demonstrations, and lectures that are dedicated to horse owners and enthusiasts. This year's Horse Day took place on **October 14, 2023,** and VGL was there to talk to horse owners, breeders and students attending the event. We had our coat color activity and brochure to share some fun genetics with our visitors!


May be an image of 4 people and text that says 'UC DAVIS HORSE DAY Saturday, October 14, 2023 Demonstrations & Discussions with Equine Experts at the Historic UC Davis Horse Barn Potential topics include: Equine Reproduction - Health and First Aid Hoof Care Physiology and Conformation Technologies n Vet Science and more! For.more.informationandregistratio more information and please visit http:/amasioncucavis.euhoredy Raffle and Trade Show for all Attendees!'

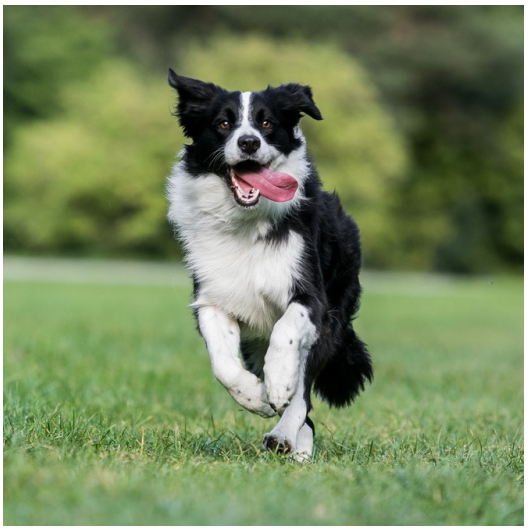
New VGL Tests

Horse. The researchers also detected the allele in nine other horse breeds, confirming blindness in horses homozygous for *CSNB2* (*CSNB2*/*CSNB2*) in the Standardbred and the Missouri Fox Trotting Horse. This is the first report with clinical confirmation of CSNB in Standardbreds and Missouri Fox Trotting Horses, suggesting the condition has been previously underdiagnosed in these and other breeds. Given recent findings, the VGL recommends testing horses from breeds that have the *CSNB2* allele to allow for informed breeding decisions, particularly Tennessee Walking Horses, Standardbreds, Missouri Fox Trotting Horses, and closely related breeds. The study was supported in part with funding from the United States Trotting Association (USTA) and from the UC Davis Center for Equine Health.

Testing for *CSNB2* can be ordered through the VGL.

Dr. Sophie Preckler-Quisquater, a postdoctoral scholar in the Mammalian Ecology and Conservation Unit, published a paper that was featured in the cover of Molecular Ecology. The study used multiple genome sequencing techniques to study two morphologically similar lineages of North American grey foxes.

Rod-cone dysplasia type 2 (rcd2)

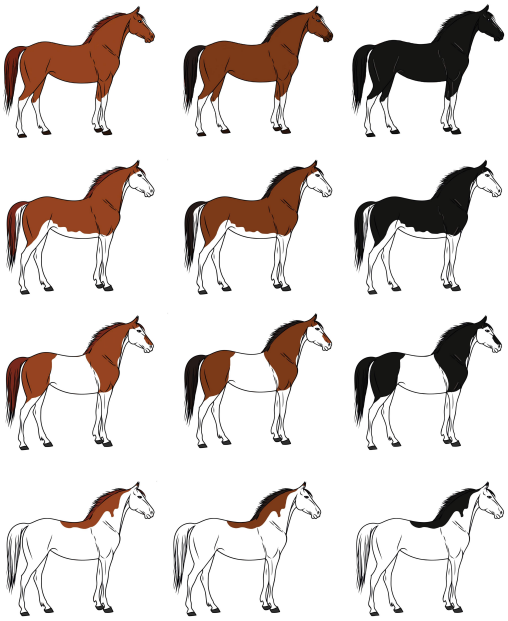


Rod-cone dysplasia type 2 (rcd2) is a type of early-onset retinal degeneration that affects collie breeds and typically leads to blindness. Affected dogs begin showing signs of night blindness as early as 6 weeks of age and can be completely blind by 6-8 months.

DNA testing can determine the genetic status of dogs and help inform breeding decisions to avoid producing affected puppies.
Visit our website to learn more about this disorder.

Splashed White - Test Update

Dr. Ben Sacks and **Stevi Vanderzwan** from the Mammalian Ecology and Conservation Unit, collaborated on the development of an integrated spatial capture–recapture model that was used to estimate the abundance, density, and distribution of the cougar (*Puma concolor*) in a remote protected area of Yosemite National Park. Noninvasive survey methods such as the one described in the study can be useful to support conservation efforts in situations where the capture and handling of animals would be challenging and expensive.



The equine Splashed White test has been updated to include two new alleles: **SW7** and **SW8**.

Splashed white is a variable white spotting pattern characterized by a broad blaze, extended white markings on the legs, variable white spotting on the belly, and often one or two blue eyes.

SW8 was recently discovered by VGL research highlighted above in our Case of the Quarter.