

Equine Genetic Disorders Reviewed

What do sex-reversal syndrome, junctional epidermolysis bullosa, and autosomal trisomy have in common? They're all equine genetic diseases. But these conditions also have distinct differences that can help practitioners evaluate and diagnose suspect cases.

At the Equine Endocrine and Genetic Disorders Symposium, held Nov. 21 in Lexington, Ky., Teri Lear, PhD, equine genetics researcher at the University of Kentucky's Gluck Equine Research Center, delivered a presentation reviewing how practitioners can recognize genetic diseases in everyday practice.

Lear explained that there are two types of genetic disease: chromosome abnormalities and inherited disease. The former can include chromosome or DNA mutations (termed disorders of sex development, or DSD) or variations in chromosome structure or number. The latter includes diseases passed genetically from a parent to an offspring.

She reviewed how veterinarians can examine chromosomes and DNA before describing how these disorders appear in clinical practice.

Chromosome Abnormalities

Equine Turner syndrome The first DSD Lear described was equine Turner syndrome, in which a mare is missing one X chromosome (XO instead of XX; in other words, she has 63 chromosomes, not 64). Lear said this defect can occur in all breeds and is the most common chromosome abnormality. These mares typically present with a small body size, poor conformation, angular deformities, and small and inactive ovaries with an underdeveloped reproductive tract. XO mares are also infertile.

Sex-reversal syndrome Next, Lear described sex-reversal syndrome, in which a horse has the outward appearance of a mare, but is genetically male (XY instead of XX). This defect has been identified sporadically in Thoroughbreds, Standardbreds, Quarter Horses, and in families of Arabian horses. These infertile horses might be bigger bodied than most mares and have small inactive ovaries and a non-tubular uterus.

Sex-reversal syndrome can also go the

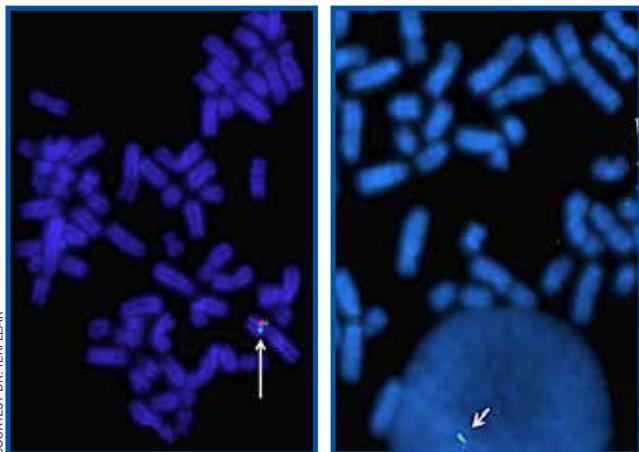
other way: Outwardly male horses can have an XX sex chromosome instead of an XY. Lear said these horses have malelike external genitalia, but lack sperm; ovarian tissue; increased anogenital length; an enlarged clitoris or small penis; and a closed vagina. Most exhibit stallion-like behavior, Lear said, and some have high testosterone levels. XX males are also infertile.

Autosomal translocations Another type

of chromosome abnormality is autosomal translocation, which occurs when there is an interchange or transfer of chromosomal segments between two or more different chromosomes. Lear said translocations are known to cause repeated early embryonic loss (generally within the first 45 days of pregnancy), but some affected mares can produce foals. Affected horses appear normal, she said.

Autosomal trisomy The last chromosomal abnormality Lear described was autosomal trisomy, a condition similar to Down syndrome in humans in which there are three copies of a particular chromosome, instead of the normal two. Lear said affected horses can have clinical signs including abnormal genitalia, an overbite, angular limb deformities, scoliosis, a domed skull, neurologic deficits, and metabolic disorders.

Some horses are less severely affected than others, she noted. She described one filly that was considered a dummy foal (a foal that suffered from lack of oxygen during delivery), but later developed progressive neurologic and musculoskeletal deficits, inappropriate social responses, a dull



Left image: Two genes mapping to the horse X chromosome (red and green dots). Right image: A gene from the horse Y chromosome mapping to the Y chromosome (green dots). Chromosomes are stained blue.

Articles of Interest

Nutrition's Role in Enhancing Aging Horses' Immunity

Role of Nutrition in Geriatric and EMS Horses

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Equine Genetic Disorders

mentality, a protruding tongue, abnormal joint growth, and a history of falling in her stall. After evaluation, Lear said the filly was diagnosed as having 40% normal chromosomes and 60% chromosomes with trisomy.

Inherited Disorders

Lear closed her lecture with a discussion on inherited genetic disorders. Many of these are breed-specific, she said, and practitioners should keep that in mind when faced with a possible case. She touched briefly on:

Junctional epidermolysis bullosa (or JEB), a condition found in American Saddlebred and Belgian foals that causes skin lesions over pressure points of the body in newborn foals and results

in large areas of skin loss. The disease is a lethal condition, and affected foals typically are euthanized shortly after birth.

Lavender foal syndrome is a fatal neurologic disease found in Arabian horses. Common clinical signs in affected foals include seizures; opisthotonos (severe hyperextension of the head, neck, and spinal cord); stiff, paddling leg movements; nystagmus (involuntary movement of the eyeballs); and, often, a soft lavender color to the foals' coats.

Dwarfism in Miniature Horses is an area recently researched by one of Lear's graduate students, John Eberth. He confirmed four types of dwarfism in Miniatures, caused by mutations in a specific gene important for cartilage development. Eberth's research on the topic is ongoing.

Lear noted that her current and future

research goals involve evaluating:

- Contracted foal syndrome in Thoroughbreds;
- Parrot mouth in Thoroughbreds and other breeds; and
- Lordosis (sway back) and congenital cataracts in American Saddlebred horses.

Take-Home Message

While researchers like Lear have developed a good understanding of equine genetic disorders, there is still much to learn. But for now, equine practitioners can use the solid knowledge base Lear and many others have developed to evaluate and diagnose horses with chromosomal abnormalities and inherited disorders. [UK](#)

Erica Larson is the News Editor for TheHorse.com.

Nutrition's Role in Enhancing Aging Horses' Immunity

Over the past century, improvements in health care and advancements in biology, chemistry, and medicine have extended the average lifespan of humans and companion animals, including horses. However, we are now facing new challenges with the paradox of an older population with increased longevity, while confronted with the potential for many years of poor health. A better understanding of the mechanisms leading to a decline in physiologic function with age would provide new predictive biomarkers and potential therapeutic targets.

It has been well-documented that the aged, including horses, have increased susceptibility to and prolonged recovery from infectious diseases, poor responses to vaccination, and increased incidence of various cancers. Furthermore, it is now



ANNE M. EBERHARDT

accepted that chronic inflammation (inflamm-aging) is a major underlying condition of many age related diseases, such as atherosclerosis, arthritis, cancer, diabetes, osteoporosis, dementia, vascular diseases, obesity, and metabolic syndrome.

In anti-aging research, much attention is focused on nutritional interventions as practical, cost-effective approaches to mitigating this

age-related breakdown in immune function. These natural dietary compounds found in a variety of fruits, vegetables, nuts, and seeds are promising candidates in helping to combat the effects of aging. They possess broad biological activities: anti-oxidative, anti-inflammatory, detoxification, regulating signaling pathway, and modulation of enzyme activities (see sidebar on the following page).

Since aged horses (those 20 years and older) have increased levels of inflammation, and treatment with long-term use of non-steroidal anti-inflammatory drugs (NSAIDs) such as flunixin meglumine and phenylbutazone can pose health problems, we are interested in nutritional interventions to counteract this inflamm-aging process.

Flavonoid (quercetin) and polyphenolic compounds (curcuminoids, resveratrol, pterostilbene, and hydroxypterostilbene) were compared to phenylbutazone and flunixin meglumine to determine differences in equine cytokine production in cell culture. White blood cells from aged horses were isolated and incubated overnight with each compound of NSAID at multiple concentrations. Inflammation production was measured when white cells were stimulated.

At varying doses (measured in micromolar units [μM]), each of the compounds and NSAIDs significantly reduced cellular inflammation: curcuminoids (20 μM), hydroxypterostilbene (40

Aging Horses' Immunity

Natural Dietary Compounds with Anti-Inflammatory Mechanisms

Group	Compound
Carotenoids	Beta-carotene, lycopene, lutein
Flavonoids	Quercetin, catechin, epigallocatechin-3-O-gallate, theaflavin
Isothiocyanates	Sulforaphane, phenethyl, isothiocyanate
Terpenoids	Limonene, retinoic acid
Proanthocyanidins	
Omega-3 fatty acids	Eicosapentaenoic acid, docosahexaenoic acid
Polyphenolic compounds	Curcumin, gingerol, resveratrol, pterostilbene

μM), pterostilbene (80 μM), quercetin (160 μM), resveratrol (160 μM), flunixin meglumine (40 μM), and phenylbutazone (>320 μM). Interestingly, curcuminoids at a concentration of 20 μM reduced inflammation to the same level as higher doses of flunixin meglumine (40 μM) and phenylbutazone (>320 μM). All natural compounds outperformed phenylbutazone by being effective at lower doses.

This preliminary research has led into two studies using aged horses to determine, 1) if a relationship exists between circulating vitamin and fatty acid levels to systemic inflammation and muscle mass, and 2) if anti-inflammatory supplementation affects immune responses to vaccination. These are preliminary steps to identify effective nutritional intervention regimens to improve function of the immune system in the aged horse. [UK](#)

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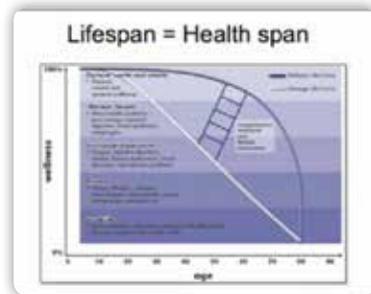
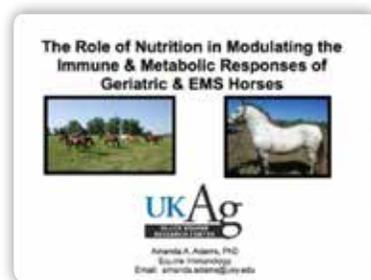
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Role of Nutrition in Geriatric and EMS Horses

Dr. Amanda Adams, assistant research professor at the University of Kentucky's Gluck Equine Research Center, shares information about how nutrition can impact the immune responses of geriatric horses and horses with equine metabolic syndrome.



View the video at TheHorse.com/32879



UK College of Agriculture, Food, & Environment Has New Dean

Citing a compelling vision for the future of the college and a national reputation for leadership, University of Kentucky (UK) Provost Christine Riordan, PhD, announced Nov. 26 that Nancy Cox, PhD, has accepted the position as dean of the UK College of Agriculture, Food, and Environment.

Cox will replace Scott Smith, PhD, who has led the college since 2001. He plans to return to the faculty, where he has been a member of 35 years. Cox is currently associate dean of research in the college, a position she has held since 2001 when she came to UK from Mississippi State University.

Cox is widely regarded as a leader in the field, serving on several national policy boards and organizations and has close ties to both civic and agricultural leaders throughout the commonwealth, which enthusiastically supported her candidacy for dean.

President Eli Capilouto praised both Cox and Smith, saying, "UK College of Agriculture, Food, and Environment is widely known as one of the leading colleges in the country. That's, in large part, because of the leadership of Scott Smith and his senior team, including Nancy Cox.

We Need Your Feedback!

We want to give you, our valued readers, the opportunity to share your insights about the Bluegrass Equine Digest newsletter. We have created a brief online survey to collect this information, and we would appreciate your help. The information that you share with us will be very valuable as we plan for the future.

This quick survey asks you to share your perceptions of the Bluegrass Equine Digest as well as some information about yourself. Rest assured, your responses are completely anonymous. Your answers simply help us to better understand our readers and what we can do to provide improved content.

The survey should only take a few minutes to complete, but will provide us with better information about our readers and what you would like to see in the Bluegrass Equine Digest. [Click here to access the survey now.](#)

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"I want to thank Dean Smith for his long service and leadership," he continued. "I am excited about the prospect of Nancy's continued leadership both on our campus and in the Commonwealth,

where she is so respected."

Riordan added, "We are fortunate to have found someone with the rare combination of skills, intellect, and reputation of Dr. Nancy Cox. She is widely known and highly regarded throughout Kentucky for her knowledge and relationships with agriculture, political, and civic leaders. At the same time, she is renowned nationally in helping lead the research and administrative efforts of one of the country's premier colleges of agriculture, including an extension service treasured throughout the commonwealth."

Among many duties, Cox oversees the college's research portfolio, which includes \$25 million in external awards in fiscal year 2012. She currently represents the college on most Kentucky agricultural commodity boards, was the founding administrator of the university's growing Ag Equine Programs, and has been the key administrator in innovative alliances with private industry such as Alltech.

On the national level, she serves or has served on key federal policy boards such as the American Society of Animal Science and the USDA's National Advisory Board for Research.

"I am very honored to represent, serve, and support the UK Ag community, including its outstanding students, faculty, staff, and an extension system known throughout the commonwealth for its commitment to service," Cox said. "During interviews for this position, I was impressed once again by the pivotal role the College of Agriculture, Food, and

UK Researchers Receive 2013 Horse Call Grant Award

The Zoetis Horse Call Grant review committee announced the award of a Horse Call Grant for the study proposal, "Transabdominal Ultrasonography: A Monitoring Tool for *Parascaris equorum* Burdens in Foals," by Martin Nielsen, DVM, PhD, EVPC, assistant professor of parasitology at the University of Kentucky (UK) Maxwell H. Gluck Equine Research Center, and Jill Stowe, PhD, Dickson Professor of Equine Science and Management in UK's Department of Agricultural Economics and in the Department of Economics.

"We believe that the most productive way to advance veterinary research will be through an open model of discovery," said Rob Holland, DVM, PhD, director of Zoetis Outcomes Research. "There is a real economic value imposed on horse owners when their horse becomes sick and a real value when disease is prevented from occurring. We would like to help establish what the intrinsic value is when this is done and to determine how a practice achieves this. This collaboration is grounded

in Zoetis' mission to continuously improve the health and care of animals worldwide and our commitment to advance veterinary science and education."

The Zoetis Horse Call Grant Award is an annual program that helps fund one or more individuals through a grant for innovative, clinically relevant research proposals that determine the economic impact of equine encephalitic, parasitic, respiratory or other infectious diseases or the impact of preventing these diseases. Another area this grant supports is determining the economic impact of practice management business practices that emphasize prevention of equine infectious or parasitic diseases or prevention of horse-related injuries to clients, technicians or veterinarians of these practices.

"By focusing on the economic aspect of equine care, we're hoping this call for collaborative research will help set a new standard in equine research," said Sabina Gasper, senior director of Zoetis Outcomes Research. "This is one of many types of partnerships we're exploring at Zoetis, and we hope working with partners we will not only advance science but help to promote excellence in veterinary economic research." UK

New Dean

Environment and its stakeholders play in the economy of the Commonwealth. I look forward to building on that legacy of engagement.”

Riordan also thanked Smith for his long tenure as dean, which included leading the college in confronting pivotal issues such as mare reproductive loss syndrome and the end of the tobacco price support program.

“UK is fortunate that Dean Smith will continue in our faculty ranks, providing his guidance to students, faculty and staff in the college and throughout the university,” Riordan said.

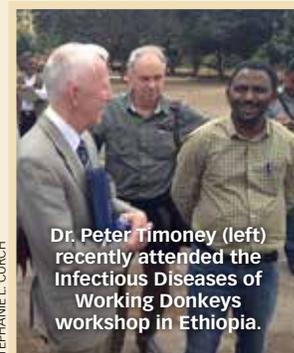
Department of Entomology Chair John J. Obrycki, PhD, who co-chaired the search committee that helped select Cox, said, “Dr. Nancy Cox impressed the members of the search committee with her breadth of knowledge, administrative experience, and understanding of the land-grant mission of the College of Agriculture, Food, and Environment at

First Annual Havemeyer Workshop on Infectious Diseases of Working Donkeys

The first annual Havemeyer Workshop on Infectious Diseases of Working Donkeys was held November 2013 in Ethiopia. The University of Kentucky was represented by Peter Timoney, PhD, FRCVS, professor and former department chair and director of the Maxwell H. Gluck Equine Research Center.

“The intent of this workshop is to bring together experts from around the world, with the aim of identifying future strategies and solutions needed to reduce the burden of infectious diseases on working equids worldwide,” noted the event’s organizing committee. “Through sharing of knowledge and experience, it is hoped we can arrive at solutions to some of the problems that affect the health, welfare, and productivity of working equids.”

Presenters at the meeting covered everything from viral, bacterial, fungal, and parasitic pathogens to diagnostics, surveillance, and disease control strategies. **UK**



Dr. Peter Timoney (left) recently attended the Infectious Diseases of Working Donkeys workshop in Ethiopia.

STEPHANIEL CURICH

the University of Kentucky. She stressed the need for a shared vision of the future of the newly renamed college and how we will address the diverse needs

of the citizens of the commonwealth of Kentucky.”

Riordan said Cox begins her tenure as dean on Jan. 1, 2014. **UK**

Kentucky 2013 Farm Cash Receipts Could Approach, Exceed \$6 Billion

Kentucky agricultural cash receipts could approach or possibly exceed \$6 billion in 2013, boosted by exceptionally strong equine, poultry, and cattle markets, according to agricultural economists from the University of Kentucky College of Agriculture, Food and Environment.

PhDs Will Snell, Kenny Burdine, Cory Walters and Tim Woods, all from UK’s Department of Agricultural Economics, shared their agricultural economic outlook for 2014 and an overview of 2013 during the Kentucky Farm Bureau Federation conference Dec. 5 in Louisville.

Official U.S. Department of Agriculture 2013 cash receipts won’t be released until summer 2014. But based on recent sales figures, it appears the upward trajectory in equine receipts experienced in 2011 and 2012 continued in 2013.

“The increase in sales figures during the fall sales indicate the market is continuing to rebound from post-recession lows,” Burdine said. “Continued strong sales and likely increases in stud fees next year should support the market. Assuming demand remains strong, 2014 receipts could post another year-over-year increase.”

The agricultural outlook for 2014 is mixed, with expectations of continued strong prices for livestock, coupled with lower feed costs and strong international demand.

“The equine industry is hoping momentum will continue,” Snell said.

A copy of the outlook publication, including information on individual farm sectors, can be found at www2.ca.uky.edu/cmsspubsclass/files/esm/Outlook2014.pdf. **UK**

>Carol Lea Spence is a UK Agricultural Communications Specialist.

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