What's In a Gene

by JW EQUINE

Dr. Danika Metallinos, a doctor of veterinary medicine with a bachelor's degree in genetics and a Ph.D. in molecular biology, addressed attendees at last year's Harris Ranch Seminar on the subject of "Genetics of Horse Breeding." As an assistant professor in genetics and part of the research team at U. C. Davis, Dr. Metallinos is working to develop more DNA tests that help eliminate undesirable traits and diseases.

"Inbreeding can cause problems when one individual piles up in a pedigree," she said, speaking of the 'popular sire effect' seen in breeds where artificial insemination is allowed. She expanded that to include a mare where embryo transfers are used. This effect, she said, weakens the gene pool of a particular breed and is magnified by the popularity of the offspring. (Impressive, progenitor of HYPP in Quarter Horses, would be an example.) She also mentioned Hyperelastosis Cutis (non-healing wounds under the saddle) as being an inheritable problem in cutting horse lines, which can be compounded in this manner.

At present, there is little fear of the popular sire effect occurring through artificial insemination or embryo transfer in the Thoroughbred industry. Neither is allowed, and during the 23rd International Thoroughbred Breeders meeting in Ireland, on Aug. 30, 2001, the status quo regarding these breeding methods was unanimously readopted. Protection, and control of the product so as to ensure a level playing field for all breeders, were given as the main reasons.

Getting more detailed about genetics, Dr. Metallinos used examples of coat color to define several genes. With autosomal dominant genes, the trait is not carried on a sex chromosome. It will be expressed, and half of the offspring will exhibit the trait regardless of gender when mated with a "normal" mare or stallion (i.e. tobiano coloring). Likewise, autosomal recessive genes are not carried on a sex chromosome, but both parents have to be carriers and both genes have to be present in order for the trait to be exhibited (i.e. white Thoroughbreds). Autosomal recessive traits are seen most often in inbred families. Semi-dominant genes are seen when some of the trait is displayed if one gene is present or more is displayed if both genes for the trait are present (i.e. buckskin and palomino coloring). In the case of polygenic traits, more than one gene affects the trait (i.e. the amount of white markings). Conformation traits are also influenced by polygenic or multiple genes.

Traits that are sex-linked are seen in sons of sons and daughters of daughters. Although the Y-chromosome is considerably smaller than the X-chromosome, research on the human genome has shown that some traits (i.e. hairy ears in men) are Y-linked, passing from father to son. On the female side, several things are suspected of being X-linked. Dr. Metallinos cited hemophilia in humans, where the daughters are carriers and sons are afflicted, as an example.

Non-chromosomal, mitochondrial DNA (mtDNA) is the stuff of female line (tail-female) inheritance, as mitochondria are passed almost exclusively through egg cells. In extremely rare cases sperm may contribute mitochondria, but in such small amounts that their mitochondria is overwhelmed by that of the egg. In essence, only females can pass a mitochondrial trait to the next generation even though both males and females can exhibit such a trait. Mitochondrial DNA has over a thousand copies per cell, is maternally inherited without recombination, and is not unique to that individual. It differs from nuclear DNA, which has two copies per cell, is inherited from both parents and is unique to that individual.

At first glance this maternal inheritance may look like a trait inherited in a different manner, except that all maternal offspring are affected instead of the 50 percent one would expect. Another telling characteristic is that even though males are as likely as females to be affected, their offspring will not exhibit the trait unless they were bred to a mare carrying the trait.

Mitochondrial DNA, although a minute percentage compared to nuclear DNA, serves an important role in cell respiration and nutrition. Mammalian mtDNA encodes fewer than 40 genes, 13 of which produce proteins, without which the main energy generation system used by cells would not function. The balance of these genes do the translating and are the support system. Dr. Ann Bowling, a leader in horse genetics at U. C. Davis before her death, referred to the mitochondrion as a "metabolic power station" in her book *Horse Genetics*.

Mitochondrial DNA has a different pattern of inheritance than nuclear DNA, and is currently being used to compare horse breeds, study populations of animals, assess genetic variation, trace the origins to the human race, and to solve crimes.

Regarding imprinted genes, Dr. Metallinos stressed that they are present in mammals only, and that all imprinted genes relate to growth and development (more on imprinted genes in an upcoming article).

Although genetic research usually focuses on diseases or disorders, tests have been developed for recessive traits (dominant and semi-dominant traits display themselves—therefore, tests are not imperative). So, as more and more questions are answered, an explanation for desirable characteristics and a way of predicting them, will likely emerge.