CFS Classification System Developed (AAEP 2012)

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Foals with contracted limbs, which can prevent them from standing or walking normally, often display a variety of other physical abnormalities that veterinarians haven't directly tied to the contractures. Understanding correlations between these characteristics could help veterinarians build a bank of knowledge to help guide treatment down the most effective path.

To this end, a group of researchers worked together to develop a uniform system for classifying cases of contracted foal syndrome (CFS). Jana Caldwell, MS, a PhD candidate at the Texas A&M University College of Veterinary Medicine and Biomedical Science, described the preliminary classification system she developed in a presentation at the 2012 American Association of Equine Practitioners convention, held Dec. 1-5 in Anaheim, Calif. Caldwell said she collaborated with colleagues—including Nathan M. Slovis DVM, Dipl. ACVIM, CHT, of Hagyard Equine Medical Institute, in Lexington, Ky.—on this project.

"A classification system will help make associations between certain phenotypes (expressed visible traits), optimal treatment approaches, and prognoses," she said.

Caldwell noted that CFS can affect any number of limbs and severe cases might require euthanasia. Mildly affected foals can receive medical and/or surgical treatment and lead productive lives.

The team reviewed 242 foal medical records from two large referral clinics and four Thoroughbred breeding farms. Foals were included in th study if they were born with a flexural deformity in at least one joint that required medical attention.

Caldwell and colleagues separated the foals into groups based on the number of limbs affected; they then further divided the groups by the nature of any additional clinical signs the foals displayed. The team then determined each case's disease severity based on treatment duration and angular measurements.

Preliminary classification groups for CFS included:

CFS IA, IB, and IC—Foals within these classifications possess flexion contractures only. "The literature indicates that there can be different underlying reasons for one limb involvement or multiple limbs," Caldwell explained. Also, she said, this is the least complex phenotype, meaning researchers would have different expectations from a molecular study seeking to tie together a variety of clinical signs at a molecular level than when looking at a more complex phenotype. She noted that the different subcategories are used to indicate the nature of limb involvement.

"For example, CFS IA are foals presenting with unilateral contractures (affecting only one limb but may include multiple joints (fetlock, knee or both)," Caldwell explained. "CFS IB are foals with bilateral contractures or when joints of the same leg are affected, both front knees, both front fetlocks, or all four."

CFS IIA and IIB—Foals classified in these groups have additional types of limb defects, Caldwell said. CFS IIA foals have angular limb deformities (valgus or varus deviations [i.e., toed out or toed in, respectively] in one or more joints), while IIB foals have ligament/tendon laxity. "These types of defects can be seen occurring at the same time, and so this infers that there may a common molecular pathway that has been disrupted during development that affects connective tissue of the limbs," she explained.

CFS IIIA, IIIB, IIIC—Foals considered IIIA have flexion contractures and entropion (lower eyelid inversion); IIIB foals have flexion contractures and patent urachi (failure of the urachus--a structure in the fetus which lies between the tip of the bladder and the umbilical cord and allows the excretion of urine during gestation--to obliterate, causing urine to come out the umbilicus); and IIIC foals have flexion contractures and both entropion and patent urachus, Caldwell explained.

"From medical records there was a relatively high incidence of these malformations occurring with flexion contractures and also occurring together," she said. Caldwell said this suggests a connection between these two structures and contractures and a possible shared molecular pathway during fetal development.

"They are all caused by (improper development) of common connective tissues," she continued, noting that many veterinarians might consider relating these defects to one another preposterous. However, she added, "there are many reports of one mutation in one gene that leads to seemingly unrelated developmental defects."

CFS IV—This group of foals possesses flexion contractures and defects of the axial skeleton (head, neck, and spine) including scoliosis, other vertebral defects, prognathism (underbite), and wry nose (a condition in which an affected foal's upper jaw and nose are deviated, or turned to one side), Caldwell said.

CFS V—This classification describes foals with flexion contractures and abdominal wall defects (herniations). "There are studies in other species that report an incidence of abdominal wall defects (in combination) with joint contractures," Caldwell explained. "Again, that suggest there is some common gene or pathway that is disrupted during development that only affects these structures."

Additionally, the research team suggested classifying disease severity by the numbers 1, 2, and 3 (with 1 being mild at 5-15 degrees in a flexed position from a normal limb angle, 2 being moderate at 15-30 degrees, and 3 being severe at more than 30 degrees), Caldwell said.

Caldwell noted that the system could be modified, if needed, once the equine veterinary community implements it in practice. Additionally, she said, changes could be made as scientists make research advancements and develop a better understanding of different phenotypes' biologic bases.

"We will never know for sure if these defects are related or are caused by independent factors until we can do more research, and we can't do good research until we have well-characterized phenotypes," Caldwell explained. A website is in the works that will allow veterinary professionals to add case data to the collection.