ASVP DIAGNOSTIC EXERCISE NO. 29 **Roger Kelly**

(case kindly provided by Santiago Diab, of the San Bernardino CAHFS Diagnostic Laboratory, California, and circulated by the ASVP Mailing List on Feb.16, 2009)

History:

A blue-eyed white male American Paint foal showed signs of colic almost from birth. Euthanasia was performed at one day of age.

Description:

The small intestine is uniformly distended. The caecum is dark green, which suggests it contains mainly meconium. The large and small colon are uniformly pale, thin and empty, with the rectum apparently devoid of the meconium to be expected here in a colt foal.

Interpretation:

In such a young subject, developmental abnormality would have to be a likely basic disease process. Neoplasia doesn't get a look-in, nor is there evidence of acute or chronic inflammation, and any degeneration is likely to be secondary to the primary developmental problem.

Morphological diagnosis:

Congenital intestinal stasis (ileus)

Differential:

Intestinal atresia can cause congenital intestinal stasis and must be excluded by careful dissection of the bowel between the distended and empty segments. In atresia, proximal intestinal distension tends to be much more severe, with accompanying abdominal distension sometimes causing dystokia.

Aetiology:

Heritable ileocolic aganglionosis (Overo lethal white sybdrome; OLWS) due to the foal acquiring two copies of a mutated autosomal gene (endothelin receptor B mutation), designated as dominant but with variable expression. The abnormal gene affects development and migration of certain embryonic sem cells of neural crest origin, which in turn affects coat and eye color and development of intestinal ganglia.

Genetic testing of candidates for breeding can reveal their carrier (heterozygous) status. Samples from this foal tested homozygous for the mutated gene (Veterinary Genetics Laboratory, UC Davis).

Testing for this gene is available in Australia (see http://www.aegrc.uq.edu.au/index.html?page=30053)

Reference:

Santschi EM, Purdy AK, Valberg SJ, Vrotsos PD, Kaese H, Mickelson JR. Endothelin receptor B polymorphism associated with lethal white foal syndrome in horses. Mamm Genome 1998; 4:306–309.

Please advise me at roger-kelly@aapt.net.au if you want to discuss this case.