N16-180 ‘RJ’

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Initial Presentation

Signalment:
• 1 day old colt
• Quarter horse

History:
• Primiparous mare
• Premature umbilical cord rupture
• Foal unable to stand or nurse post-delivery
Initial Assessment

- Obtunded
- Bradycardia
- Injected mucous membranes
- Hypoxemia
Hospitalization

- Supportive care
- Oxygen therapy
- Colostrum
- Ultrasound
- Blood culture
Hospitalization

- Initial improvement
- Diarrhea
  - Diarrhea panel
- Spontaneous death
Necropsy

- Mild serosanguinuous peritoneal effusion
- Mildly reddened small intestinal serosa
- Mild, multifocally reddened descending colon
- Bright green, liquid to pasty feces
Gross Findings

- Peritoneal effusion, serosanguinous, mild
- Enterocolitis, multifocal, minimal to mild
Morphologic Diagnoses

Glycogen-like inclusions, intrasarcoplasmic, multifocal, moderate to marked, heart

Glycogen-like inclusions, intracytoplasmic and extracellular, multifocal, mild to moderate, brain
Ancillary Testing

DNA Testing:
- Glycogen Branching Enzyme Deficiency (GBED)
  - Homozygous for GBED gene mutation
Glycogen Branching Enzyme Deficiency (GBED)

Glycogen storage disease IV

- Autosomal recessive
  - Gene mutation in GBE1 gene
  - Nonsense mutation in codon 34

- Glycogen branching enzyme
  - $\alpha$1,6 branch point formation during glycogenosis

- Fatal hereditary condition in Quarter horses

- Death in utero or early life
Thank you!
Recommended Resources

Image Credits & Sources

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