

# \* *Breed Specific Diseases in Horses*

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## Acknowledgements

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\* *Breed Specific Diseases* ó

Defined as Specific to that öFamilyö of horse  
 meaning this disease can be inherited and, thus,  
 has a genetic basis

*Some diseases may even be found primarily  
 within certain bloodlines within the breed*

Genetics ó inheritance of traits

Autosomal dominant

Autosomal recessive

Sex-linked

\* *Diseases are passed from parents to  
 offspring through DNA*

\* *Autosomal Dominant* trait means the abnormal gene from one parent is needed in order for offspring to inherit the disease. One parent may often have the disease.

\* *Autosomal Recessive* trait is expressed only in the absence of a dominant gene. Both parents must carry the mutated gene and each parent must pass this copy of the mutated gene. Affected offspring, thus, inherit two copies of the mutated gene and the associated disorder is then expressed. When two carriers of a recessive disorder are bred, they have a 25% chance of producing an affected foal.

\* *Sex Linked* traits are carried by either sex chromosome

\* *Breed Specific Diseases in Horses  
 Modes of inheritance*

*Horse Genome Project*

\* Mapping of the Equine Genome continues to aid the understanding of the genetics of many equine diseases

- 1 - Importance ó reduce disease incidence through genetic testing and careful breeding
- 2 - Gene therapy may be employed in the future as a method to treat some diseases

**\* *Breed Specific Disease  
Process of Discovery***

\* Arabians

\* Quarterhorses

\* Paints

\* Appaloosas

\* Thoroughbreds

\* Drafts

\* Miniature horses

**\* *Breed Specific Diseases  
Popular Horse Breeds***

- \* SCID (Severe Combined Immunodeficiency)
- \* CA (Cerebellar Abiotrophy)
- \* LFS (Lavender Foal Syndrome)

*Other Arabian diseases*

- Juvenile Idiopathic Epilepsy
- Guttural Pouch Tympany (GPT)
- Occipitoatlantoaxial Malformation (OAAM)
- Strangulating Lipoma
- ESPA (Equine Systemic Proteoglycan Accumulation)



*\* Arabian Specific Diseases*

*Severe Combined Immunodeficiency Disease (SCID)*

- \* inherited disease seen in pure and part-bred Arab horses
  - Autosomal recessive
- \* results in very weakened immune system
  - Enhanced susceptibility to infection and first show signs of disease at between two days and eight weeks of age.
- \* symptoms - raised temperature, respiratory complications, diarrhea
  - typical of new-born foals with a range of infections.

*Foals affected by SCID always die from the disorder within the first six months of life*

*\* Arabian Specific Diseases*

### *Cerebellar Abiotrophy (CA)*

- \* inherited as an Autosomal recessive trait
- \* foals seem normal at birth
  - symptoms typically noticeable after four months
  - maybe observed shortly after birth,
  - sometimes develop after the first year
- \* clinical signs - head tremors, a lack of balance, difficulty rising from a reclining position
  - May be confused with Wobbler's syndrome, Equine Protozoal Myeloencephalitis (EPM) and trauma

### \* *Arabian Specific Diseases*

CA affected foals are often euthanized or restricted to life as pasture pets

- \* never coordinated enough to be ridden safely
- \* danger to themselves because the condition predisposes these horses to accidents and injury.

### \* *Arabian Specific Diseases*

Lavender Foal Syndrome (LFS)-

- \* proposed as autosomal recessive inheritance
  - Foals born with unusual pale lavender, pale pink or even silvery appearance = lavender foal
- \* neurologic disease
  - Often misdiagnosed as "dummy" foals or misdiagnosed as having some type of spinal cord injury
  - LFS foals unable to stand, cannot attain sternal recumbency
  - partial seizures/episodes of tetany may be present
  - foals usually euthanized within a few days of birth on humane grounds, since these foals are incapable of survival.

\* *Arabian Specific Diseases*

\* Genetic Testing

Animal Genetics, VetGen, UC Davis

- \* SCID
- \* CA
- \* LFS

\* *Arabian Horse Specific Disease*

- \* JIE (Juvenile Idiopathic Epilepsy)  
a brain disorder - self-limiting - not usually fatal
- \* GPT (Guttural Pouch Tympany)  
affected guttural pouch distended w/ air to form characteristic nonpainful swelling
- \* OAAM (Occipitoatlantoaxial Malformation)  
malformation of occipital bone and the first two cervical vertebrae (the atlas and axis) ó atlas fused to base of the skull
- \* *Strangulating Lipoma*
- \* *ESPA/DSL D (Equine Systemic Proteoglycan Accumulation/Degenerative Suspensory Ligament Desmitis)*

### \* *Other Arabian Specific Diseases*

#### *Strangulating Lipoma*

- \* benign mass of fatty tissue that forms into a circular ball.
- \* stimulus or cause for the growth of this fatty tissue unknown
- \* Arabians (and Quarter Horses) are predisposed
- \* strangulating - cut off blood flow to the intestinal tract
  - \* causes painful problem for the horse and an emergency for an equine surgeon
- \* not all lipomas need surgery
  - might hang out in the horse's abdomen for a lifetime without ever causing signs of colic

### \* *Arabian Specific Diseases*

*DSL* (*Degenerative Suspensory Ligament Desmitis*)/ *ESPA*  
(*Equine Systemic Proteoglycan Accumulation*)

- \* debilitating disease thought to only affect the ligaments of legs
  - \* recently been proven to be a systemic disease
    - affects being found throughout the organs, eye and muscle tissue of the affected equine
    - failure of tendons and ligaments to maintain, remodel, and repair in a normal fashion.
    - fetlocks drop down - becomes coon footed
- If severe, euthanasia is often the only option

*However, some positive results reported anecdotally with diet and exercise*

### \* *Arabian Specific Diseases*

- \* HYPP (Hyperkalemic Periodic Paralysis)
- \* PSSM (Polysaccharide Storage Myopathy)
- \* HERDA (Hereditary Equine Regional Dermal Asthenia)
- \* GBED (Glycogen Branching Enzyme Deficiency)
- \* Malignant Hyperthermia
- \* Malignant Melanoma (more gray horse specific as opposed to breed)



### \* *Quarterhorse Specific Diseases*

\* *Impressive*

(Image from  
[www.ponybox.com](http://www.ponybox.com))

HYPP ó Linked to QH  
sire



\* *HYPP – “Hyperkalemic Periodic Paralysis”*

Autosomal dominant inherited muscle disorder

Only one copy of gene needed to express disease

Too much potassium ( $K^+$ ) in blood

Sporadic attacks of muscle contractions/tremors

Can progress to full body collapse and  
inability to breathe

\* *Quarterhorse Specific Diseases*

\*Prevention of HYPP

\* *Dietary Management is Critical*

Low K+ feeds (e.g., Timothy, Bermuda, oats, wheat, barley, beet pulp)

*Couple with exercise and/or turnout*

\* Medical Management ó decreases potassium levels

Acetazolamide

Hydrochlorthiazide

\* *Quarterhorse Specific Diseases*

PSSM (Polysaccharide Storage Myopathy)

\* Common cause of exertional rhabdomyolysis, i.e.ö in Quarterhorses

Different from tying-up in Thoroughbred

PSSM ó abnormal accumulation of normal sugars (polysaccharides in the muscle) and accumulation of abnormal sugars

Two forms ó Type 1 and Type 2

\* *Quarterhorse Specific Diseases*

## PSSM

### Management

#### \* Dietary

Low NSC hay

Fat sources óe.g., soybean oil, canola oil, flaxseed, peanut

#### Exercise

\* no more than 5 min walk/trot to start

\* ensuring that exercise is gradually introduced and consistently performed

\* minimizing any days without some form of exercise

## \* *Quarterhorse Specific Diseases*

### *HERDA (Hereditary Equine Dermal Asthenia)*

#### *Quarterhorse Specific Diseases*



\* Poco Bueno identified as the sire

*(Image from Legends: A Western Horseman Book – John A. Stryker of Fort Worth)*

### **HERDA**

- \* called Hyperelastosis Cutis
- \* lack of adhesion within the skin layers due to a collagen defect
- \* causes the outer layer of skin to split from the deeper layers, to even tear off completely.
- \* saddle areas seem to be most prone to these lesions.
- \* autosomal recessive trait



[http://mytevisjourney.blogspot.com/2013\\_08\\_27\\_archive.html](http://mytevisjourney.blogspot.com/2013_08_27_archive.html)

### ***Quarterhorse Specific Diseases***

#### \* **Glycogen Branching Enzyme Deficiency (GBED)**

##### ***Quarterhorse Specific Diseases***

- \* Autosomal recessive
- \* Inability to store sugar ó fatal
- \* Low energy, weakness
- \* Low body temperature, seizures ó sudden death



*Image from*  
<http://www.about-horses.net/quarter-horse/>

Malignant Hyperthermia

- \* Autosomal dominant
- \* Triggered by anesthesia
- \* High temperature, sweating, acidosis, muscle rigidity
- \* Testing recommended for QHs undergoing general anesthesia

\* *Quarterhorse Specific Diseases*

\* Malignant Melanoma

Not Quarterhorse specific as a disease

-- but gray QHs ó significantly lower prevalence

2-fold lower incidence



\* *Quarterhorse Specific Diseases*

\* *Recurrent Equine Exertional  
Rhabdomyolysis (RER) –  
“tying up”*

- \* Muscle cramps, pain
- \* Elevated muscle enzymes
- \* Resembles PSSM but different cause
- \* RER d/t defect in muscle calcium regulation not sugar



*Thoroughbred  
Specific Diseases*

- \* RER 3-4 times more likely in fillies  
Autosomal dominant sex-linked disorder
- \* RER ~6 times more likely in nervous horses
- \* Most common in Thoroughbreds 2-3 years old
- \* Management is nutritional
  - Low sugar/higher fat/higher fiber
  - No more than 20% NSC daily
  - Vitamin E/Selenium supplementation

\* *Thoroughbred Specific Disease*

Exercise Induced Pulmonary Hemorrhage

- \* Post race "Bleeders"
- \* Stallions that bleed more likely to have offspring that bleed post race

Laryngeal Hemiplegia

- \* "Roarers" d/t paralysis of laryngeal muscles 2ndary to nerve degeneration
- \* More common in taller horses
- \* Surgical correction "Tie-back"
  - \* Abducts arytenoid cartilage that obstructs the airway

**\* Thoroughbred Specific Disease**

Coat color associated deafness

- \* Splashed white or frame overo
- \* Strongly associated with
  - Extensive white on head, limbs
  - One heterochromic iris
  - Two blue eyes



**\* American Paint Horse Specific Diseases**

Frame Overo



Image from  
<http://press.apha.com/pdfs/guidebooks/ColorGenGuide.pdf>

*\* American Paint Horse Specific Diseases*

Overo Lethal White Syndrome (OLWS)

- \* Autosomal recessive
- \* Most common carriers are frame overos
- \* Fatal condition in foals few days old
  - underdeveloped, contracted intestine
  - Surgical correction not possible
  - Fatal constipation
  - Opt for euthanasia since death inevitable

*\* American Paint Horse Specific Disease*

Squamous Cell Carcinoma (SCC)

- \* thought to be from solar radiation, especially in light pigmented horses
  - (no genetic basis except light-colored skin)
- \* malignant tumor arising from skin cells
- \* SCC is the second most common tumor of the horse
- \* most common tumor of the eyelid and external genitalia.
  - Tumor is locally aggressive, but slow to metastasize.

**\* American Paint Horse Specific Disease**

Equine Recurrent Uveitis

- \* Moon blindness
- \* one of the oldest known ocular diseases in the horse
- \* most common cause of blindness worldwide
  - Appaloosas have a reported 8 fold higher incidence*

Treatment

Systemic NSAIDS (Banamine), topical corticosteroids to control the ocular inflammation, atropine for pain

**\* Appaloosa Specific Diseases**

### Congenital Stationary Night Blindness

- \* Inherited disorder
  - present from birth
  - does not get worse with age
  - Affected horses suffer from impaired night vision
  - Day vision typically is normal
- \* Management of horses with CSNB
  - Easy and straightforward
  - Since the condition present since birth, horses are already adapted.
  - Provide safe places to eat, drink and sleep during the night
  - Understand when the horse is faced with a dark area that limits their vision

## *Appaloosa Specific Diseases*

- \* Shivers
- \* Scratches/Grease Heel
- \* Chronic Progressive Lymphedema (CPL)



## *\* Draft Horse Specific Diseases*

Shivers

- \* neuromuscular condition most prevalent in draft horses and draft crosses
  - symptoms sudden jerking or trembling of the hindquarters, leg flexes toward the horse's belly, tail lifts and trembles in a pumping motion during backing
  - horse unable to back because hind legs get stuck in an upward, flexed position.
- \* speculated to be inherited
- \* not caused by PSSM

**\* *Draft Horse Specific Disease***

Grease Heel (Scratches or Mud fever)

- \* dermatitis on the back of the pastern
  - persistent with drafts (breeds w/ lots of feathering)
    - most often found in the hind legs
  - result of long hair on pastern coupled w/ constant moisture and poor hygiene
  - inflammation and oozing fluid that becomes crusty and scabby
- \* Treatment
  - cut away the long hair, wash w/ warm water and mild soap, scrub to remove the scabs
  - home remedy - 1 part Neosporin, 1 part cortisone cream and 2 parts zinc oxide

**\* *Draft Horse Specific Disease***

*Chronic Progressive Lymphedema (CPL)*

- \* affects large draft breeds with heavily feathered lower extremities primarily in Shires, Clydesdales and Belgians
- \* painful, disfiguring disease
- \* formation of large, nodules - cause permanent skin ulceration and lameness
  - may eventually lead to the animals' early demise d/t massive secondary infections
- \* initially appears to be a therapy-resistant condition - over time, vascular and lymphatic-vessel changes develop

**\* *Draft Horse Specific Disease***

*CPL good husbandry practices*

- \* blow-dry lower limbs and keep stall bedding clean and dry.
- \* effective management more successful if feathering is clipped
  - poor circulation, decreased lymph drainage, hyperkeratosis and occlusion by dense, long feathering provides the perfect environment for opportunistic infections
  - feathering can interfere with topical treatment as well as with keeping the pastern dry.
- \* other recommendations - keep legs of affected horses clean, dry and pristine, treat any infections, provide daily exercise

**\* *Draft Horse Specific Diseases***

*CPL good husbandry practices*

- \* wrap legs - short-stretch bandages developed for humans with lymphedema
  - positive influence on these horses
- \* wrap must be well padded - applied with relatively high pressure
- \* wrapping legs for three months reported to diminish swelling, nodular lesions and ulcers
- \* manual lymph drainage
  - used successfully with more acute lymphedema
  - expected to keep CPL horses in better health.

**\* *Draft Horse Specific Diseases***

- \* Fecoliths
- \* Tracheal Collapse
- \* Hyperlipemia



**\* *Miniature Horse Specific Disease***

### Fecoliths

- \* high risk of developing feed impactions and fecoliths (rock-like fecal balls)
- \* most likely due to disproportional small intestinal diameter compared to feed particle size
- \* tend to produce drier feces and coupled with the challenges of providing good dental care to ensure food is chewed properly
  - thus, more likely to get impactions
- \* feed impactions and fecoliths account for greater than 60% of all colic cases

### \* *Miniature Horse Specific Diseases*

### Tracheal Collapse

- \* most common in minis.
- \* common signs - respiratory distress, inspiratory honking noises, increased abdominal expiratory effort, exacerbated by stressful events, pregnancy, exercise, dusty environment, eating
- \* Radiographs or endoscopy typically show the collapsed trachea flattened top to bottom

Treatment is supportive

- poor prognosis - very high (up to 75%) mortality rate
- better alternative treatment may be nasotracheal intubation with a foal-size nasogastric tube
  - supplement with exogenous oxygen may temporarily stabilize affected animals

### \* *Miniature Horse Specific Diseases*

Hyperlipemia

- \* most common in ponies, miniature horses, and donkeys
- \* primary disease process associated with obesity, pregnancy, lactation, stress, or even transportation
- \* poor feed quality or decreased feed intake during a period of high-energy requirement (e.g., pregnancy, systemic disease), causes disturbance of fat metabolism
- \* accumulation of large amounts of triglycerides (break down products of fat) in the blood
- \* Diagnosis
  - gross blood sample - milky appearance d/t to high levels of fat in the blood, develops a thick white layer on top of the red blood cells

\* *Miniature Horse Specific Diseases*Hyperlipemia

## Treatment

rapid correction is necessary

- \* IV fluids to correct dehydration and other electrolyte imbalances, provide energy, as dextrose (simple sugar)
- \* nutritional therapy
  - high carbohydrate feeds as a gruel administered via a stomach tube - vital aspect of hyperlipaemia treatment.

*corrects the negative energy balance, increases glucose in the blood stream, promotes insulin release to inhibit further break down of fat*

\* *Miniature Horse Specific Diseases*

Summary

\* In closing “*breed specific equine diseases*,” have a genetic origin, and are, thus, inherited by a particular variety of horse. Should you own one of the breeds covered in this presentation, or if you have concerns about your horse in general, please contact your veterinarian for more information. I know that I would be only too happy to help you in any way I can. Thank you for your time and courtesy.

*I would like to take the next few moments to answer any of your questions now.*

\* ***Breed Specific Diseases***