



## Genetic Disorders Clinical Signs Overview

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This material is meant to provide a quick list basic overview to help owners become familiar with and recognize commonly observed clinical signs frequently associated with several genetic disorders affecting the Arabian horse. This list is not intended to provide detail on every potential clinical sign nor will an affected foal/horse necessarily show all of the signs listed for a specific disorder. Additionally, these signs may be indicative of other conditions. Links for more detailed information on each of these disorders are included below. Owners should consult with their veterinarian for additional information, evaluation and diagnosis.

### **SCID/CID: Severe Combined Immunodeficiency** (AKA “Bubble boy” disease in humans)

- Diarrhea
- Difficulty breathing
- Fever
- Blood work indicators:
  - Lack of immunoglobulin M (IgM)
  - White blood cell count <1,000/mm<sup>3</sup>

[Click here for more detailed information](#)

### **LFS: Lavender Foal Syndrome** (AKA Coat Color Dilution Lethal)

- Arched back/extended neck
- Difficult birth
- Odd coloration of hair
- Paddling legs when lying down
- Seizures
- Unable to stand

[Click here for more detailed information](#)

### **CA: Cerebellar Abiotrophy**

(AKA Cerebellar Cortical Abiotrophy)

- Head tremor
- Hyperreactivity
- Jerky movement of the head when moving
- Lack of awareness of where feet are placed
- Lack of balance
- Poor depth perception
- Problems judging speed and distance
- Stiff movement with exaggerated action of front legs
- Wide-legged stance

[Click here for more detailed information](#)

[Click here for a research update](#)

### **GPT: Guttural Pouch Tympany**

- Difficulty swallowing
- Noisy or difficult breathing
- Persistent upper respiratory infections
- Severe non-painful swelling in area behind jowl

[Click here for more detailed information](#)

### **JIE: Juvenile Idiopathic Epilepsy**

(AKA Juvenile Epilepsy Syndrome)

- Seizures ranging from twitching to loss of consciousness with rapid muscle contractions
- After seizures blindness and depression

[Click here for a research update](#)

### **OAAM: Occipitoatlantoaxial Malformation**

- Inability to stand
- Poor coordination/weakness in legs
- Paralysis in legs

[Click here for more detailed information](#)

#### **ARABIAN HORSE ASSOCIATION<sup>SM</sup>**

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