



Equine Veterinary Journal



Supplementary Item 4: Diseases/traits in the horse with unknown modes of inheritance and genetic mutations not yet identified. Diseases and traits with active research into the genetic basis of the disease or functional genomic studies are **bolded** and references include these studies. Other traits/diseases are included based upon comparative information in other species and references include case descriptions. QH = American Quarter Horse, TB = Thoroughbred, WB = Warmblood. References can be found either in the main article or in Supplementary Item 5.

Disease/trait	Breed(s)	Reference(s)
Alopecia areata	Multiple; QH and Appaloosa most common	[107]
Arthrogyposis	Multiple; Arabian, QH, TB reported	[108]
Occipitoatlantoaxial malformation; occipitalisation of atlas and atlantalisation of axis	Primarily Arabian; also reported in Appaloosa, QH, Friesian, Miniature horse	[109; 110]
Atresia coli	Multiple	[111; 112]
Atrial fibrillation, lone	Standardbred	[113]
Body size	Multiple	[27; 34]
Bone fracture, traumatic	Thoroughbred	[114]
Brachygnathia	Multiple	[115; 116]
Choanal atresia	Multiple; reported in Standardbred, Thoroughbred, American Miniature, Oldenburg horse	[117-122]
Cleft palate	Multiple	[123; 124]
Coloboma	QH	[125-127]
Common Variable Immunodeficiency	Multiple	[128]
Coxofemoral luxation	Miniature horse	[129; 130]
Cribbing (crib-biting)	Multiple	[131]
Cryptorchidism	Multiple	[132]
Curly coat	American Bashkir Curly Horse	[133]
Cushing's disease	Multiple	[134]

Deafness, sensorineural	American Paint horse	[135]
Degenerative joint disease (osteoarthritis)	Multiple; genetic studies in Thoroughbreds and Icelandic horses	[114; 136]
Degenerative myeloencephalopathy; also see neuroaxonal dystrophy	Multiple	[137-139]
Dwarfism	American Miniature, Friesian	[140]
Leukoencephalomyopathy, spongiform	QH	[141]
Ectopic ureter	Multiple	[142-144]
Endocardial fibroelastosis	QH	[145]
Entropion	Multiple	[146; 147]
Epilepsy, juvenile idiopathic	Arabians	[148; 149]
Exercise-induced pulmonary haemorrhage	Multiple	[150]
Glaucoma	Multiple	[151]
Guttural pouch tympany	Multiple; genetic studies in Arabians and German warmblood horses	[35; 152]
Hemangiosarcoma	Multiple: TB overrepresented	[153]
Heart defects including: Ventricular septal defect Atrial septal defect Persistent ductus arteriosus Tricuspid atresia Tetrology of Fallot Pentalogy of Fallot Persistent right aortic arch Persistent truncus arteriosus	Multiple; Arabian overrepresented	[154-157]
Hernia, diaphragmatic	Multiple	[158]
Hernia, inguinal	Multiple	[159-161]
Hernia, scrotal	Multiple	[162]
Hernia, umbilical	Multiple	[161; 163]
Holoprosencephaly	Morgan (1 report)	[164]
Hydrocephalus	Multiple, including Miniature horse, Friesian, Thoroughbred, Standardbred	[165; 166]
Hyperbilirubinemia	Thoroughbred	[167]
Hyperornithinemia, hyperammonemia and homocitrullinuria	Morgan	[168]

Hyperparathyroidism, primary	Multiple	[169; 170]
Hyperthyroidism	Multiple	[171; 172]
Hypoadrenocorticism	Multiple	[173]
Hypoparathyroidism	Multiple	[174-176]
Hypospadias	Friesian	[177-179]
Hypothyroidism	Multiple	[180; 181]
Hypotrichosis	Arabian, Appaloosa	[182]
Immunoglobulin M deficiency, selective	Arabian	[183]
Impaired acrosomal reaction	Multiple	[184]
Insect bite hypersensitivity	Multiple	[37; 185-189]
Intersex	Multiple	[190]
Knobbed acrosome	Multiple	[191]
Laminitis (research primarily focused on functional genomic studies)	Multiple	[42; 192-196]
Laryngeal neuropathy/paralysis, recurrent	Multiple; often taller breeds	[33; 197]
Linear epidermal nevi	Belgian Draft	[198]
Lymphangiectasia	Standardbred (1 case reported)	[199]
Lymphedema, chronic progressive (chronic pastern dermatitis)	Draft breeds	[200-202]
Megaesophagus	Multiple, high prevalence in Friesian horse	[203; 204]
Metabolic syndrome (i.e. insulin resistance)	Multiple	[205; 206]
Motor neuron disease, lower	Multiple; QH at higher risk	[207; 208]
Narcolepsy	Familial form: Miniature and Lippizan horse, Shetland and Suffolk pony	[209; 210]
Navicular disease	Multiple; genomics research in Hanoverian warmblood horses	[211-214]
Neuroaxonal dystrophy (see also degenerative myeloencephalopathy)	Multiple	[139; 215]
Neuronal ceroid lipofuscinosis	Icelandic x Peruvian Paso crossbred (3 distantly related cases)	[216]
Squamous cell carcinoma, ocular	Multiple; Draft breeds and Appaloosa overrepresented	[217]
Omphalocele	Arabian (1 report)	[218]
Osteochondrosis/Osteochondritis dissecans	Multiple; genetic studies in Warmblood breeds, Trotters, Standardbreds,	[219-230]
Osteopetrosis	Peruvian Paso	[231]
Patellar luxation	Miniature horse	[232-234]

Pemphigus foliaceus	Multiple	[235; 236]
Polycystic kidney disease	Multiple	[237; 238]
Polydactyly	Multiple	[239]
Polysaccharide storage myopathy, type 2	Multiple	[240]
Portosystemic shunt	Multiple	[241; 242]
Prekallikrein deficiency	Belgian Draft, Miniature horse	[243; 244]
Recurrent airway obstruction	Multiple; genetic studies in European Warmblood breeds	[245-250]
Recurrent exertional rhabdomyolysis	Multiple; Thoroughbred and Standardbred overrepresented	[251-255]
Recurrent uveitis	Multiple; Appaloosa and German Warmblood breed overrepresented	[36; 256; 257]
Renal dysplasia	Reported in QH, TB, American Miniature	[258-261]
Sarcoid	Multiple	[262; 263]
Schistosomus reflexus	Multiple	[264]
Self-mutilation syndrome	Multiple	[265]
Sex-reversal: XY female, SRY-positive	Multiple	[84]
Shivers	Multiple: TB, WB, Draft horses overrepresented	[266]
Situs inversus	Standardbred (1 report)	[267]
Stringhalt	Multiple	[268]
Superficial digital flexor tendon injury	Multiple; genetic studies in TB	[114; 269]
Lordosis (swayback)	Multiple; genetic studies in American Saddlebred	[270]
Syringohydromyelia	Multiple	[271]
Tendons, contracted	Multiple	[272]
Testicular regression syndrome	Oldenburg (1 report)	[273]
Wryneck / ± torticollis	Multiple; Draft horses overrepresented	[274]
Urolithiasis	Multiple	[275]
XX testicular disorder of sexual development	American Saddlebred, Arabian, Lusitano, Paso Fino	[132; 276; 277]
Yellow fat disease (nutritional panniculitis or steatitis)	Appaloosa (1 report)	[278]