Electronic Supplementary Material - Appendices

Appendix 1. Full breed list, listed alphabetically.

Breeds searched (* denotes those identified with inherited disorders)											
#		Breed	#		Breed	#		Breed	#		Breed
1	Ab	Abyssinian	31	BF	Black Forest	61	Dul	Dülmen Pony	91	HP	Highland Pony*
2	Ak	Akhal Teke	32	Boe	Boer	62	DD	Dutch Draft	92	Hok	Hokkaido
3	Al	Albanian	33	Bre	Breton*	63	DW	Dutch Warmblood	93	Hol	Holsteiner*
4	Alt	Altai	34	Buc	Buckskin	64	EB	East Bulgarian	94	Huc	Hucul
5	ACD	American Cream Draft	35	Bud	Budyonny	65	Egy	Egyptian	95	HW	Hungarian Warmblood
6	ACW	American Creme and White	36	By	Byelorussian Harness	66	EP	Eriskay Pony	96	Ice	Icelandic*
7	AWP	American Walking Pony	37	Cam	Camargue*	67	EN	Estonian Native	97	Io	Iomud
8	And	Andalusian*	38	Camp	Campolina	68	ExP	Exmoor Pony	98	ID	Irish Draught
9	Anv	Andravida	39	Can	Canadian	69	Fae	Faeroes Pony	99	Jin	Jinzhou
10	A-K	Anglo-Kabarda	40	Car	Carthusian	70	Fa	Falabella*	100	Jut	Jutland
11	Ap	Appaloosa*	41	Cas	Caspian	71	FP	Fell Pony*	101	Kab	Kabarda
12	Arp	Araappaloosa	42	Cay	Cayuse	72	Fin	Finnhorse*	102	Kar	Karabair
13	A	Arabian / Arab*	43	Ch	Cheju	73	Fl	Fleuve	103	Kara	Karabakh
14	Ard	Ardennes	44	CC	Chilean Corralero	74	Fo	Fouta	104	Kaz	Kazakh
15	AC	Argentine Criollo	45	CP	Chincoteague Pony	75	Fr	Frederiksborg	105	KPB	Kerry Bog Pony
16	Ast	Asturian	46	CB	Cleveland Bay	76	Fb	Freiberger*	106	KM	Kiger Mustang
17	AB	Australian Brumby	47	Cly	Clydesdale*	77	FS	French Saddlebred	107	KP	Kirdi Pony
18	ASH	Australian Stock Horse	48	CN	Cob Normand*	78	FT	French Trotter	108	KF	Kisber Felver
19	Az	Azteca	49	CRH	Colorado Ranger Horse	79	F	Friesian*	109	Kis	Kiso
20	Ba	Balearic	50	Com	Comtois*	80	Gal	Galiceño	110	Kla	Kladruby
21	Bal	Baluchi	51	CoP	Connemara Pony*	81	GP	Galician Pony	111	Kna	Knabstrup
22	Bei	Ban-ei	52	C	Criollo*	82	GL	Gelderlander	112	Kus	Kushum
23	Ban	Banker	53	Cri	Crioulo	83	Gid	Gidran	113	Kust	Kustanai
24	Bar	Barb	54	DalP	Dales Pony*	84	GAS	Golden American Saddlebred	114	Lat	Latvian
25	Bas	Bashkir	55	Dan	Danube	85	Got	Gotland*	115	L	Lipizzan*
26	BC	Bashkir Curly	56	DarP	Dartmoor Pony	86	Gro	Groningen*	116	LHD	Lithuanian Heavy Draft
27	BP	Basotho Pony	57	DEL	Deliboz	87	Gua	Guangxi	117	Lok	Lokai
28	B	Belgian*	58	Dj	Djerma	88	Hac	Hackney	118	Los	Losino
29	BhP	Bhirum Pony	59	Do	Døle	89	Haf	Haflinger*	119	Lus	Lusitano
30	BhoP	Bhotia Pony	60	Don	Dongola	90	Han	Hanoverian*	120	Mal	Malopolski

Breeds searched (* denotes those identified with inherited disorders)

#		Breed	#		Breed	#		Breed	#		Breed
121	Man	Mangalarga	147	NF	Norwegian Fjord*	173	RHD	Russian Heavy Draft	199	TWH	Tennessee Walking Horse*
122	Mare	Maremmano*	148	Ob	Ob	174	RT	Russian Trotter	200	Ter	Tersk
123	Mar	Marwari	149	Old	Oldenburg*	175	Sb	Saddlebred*	201	Thes	Thessalian
124	Mba	M'Bayar	150	OT	Orlov Trotter	176	Sa	Sanhe	202	TB	Thoroughbred*
125	MP	Mérens Pony	151	P	Paint*	177	SHD	Schleswiger Heavy Draft	203	Tok	Tokara
126	Mes	Messara	152	Pal	Palomino*	178	Sfu	Schwarzwälder Fuchs	204	Tor	Tori
127	M	Miniature*	153	Pant	Pantaneiro	179	SF	Selle Francais	205	Trak	Trakehner*
128	Mis	Misaki	154	PF	Paso Fino*	180	Sha	Shagya	206	US	Ukrainian Saddle
129	MFTH	Missouri Fox Trotting Horse	155	Perc	Percheron*	181	SP	Shetland Pony*	207	Vlaa	Vlaamperd
130	Miy	Miyako	156	PP	Peruvian Paso*	182	SH	Shire*	208	Vlad	Vladimir Heavy Draft
131	Mon	Mongolian	157	PPo	Pindos Pony	183	SFH	Single-Footing Horse	209	Vy	Vyatka
132	Mob	Morab	158	Pin	Pinia	184	SkP	Skyros Pony	210	WeP	Welara Pony
133	Morg	Morgan*	159	Pinta	Pintabian	185	SoP	Somali Pony	211	WP	Welsh Pony and Cob*
134	Moy	Moyle	160	Pint	Pinto	186	Sor	Sorraia	212	WAB	West African Barb
135	Murg	Murgese*	161	PK	Polish Konik	187	SGC	South German Coldblood*	213	WSP	Western Sudan Pony
136	Must	Mustang*	162	PA	Pony of the Americas	188	SoHD	Soviet Heavy Draft	214	Wie	Wielkopolski
137	NSH	National Show Horse	163	Po	Pottok	189	SpM	Spanish Mustang	215	X	Xilingol
138	NFP	New Forest Pony	164	Prz	Przewalski	190	SpB	Spanish-Barb	216	Yak	Yakut
139	NK	New Kirgiz	165	PT	Pyrenean Tarpan	191	SpN	Spanish-Norman	217	Yan	Yanqi
140	NP	Newfoundland Pony	166	Qa	Qatgani	192	Stb	Standardbred*	218	Yi	Yili
141	Nom	Noma	167	Qu	Quarab	193	SCB	Sudan Country-Bred	219	Yo	Yonaguni
142	Noo	Nooitgedacht Pony	168	QH	Quarter Horse*	194	Su	Suffolk*	220	Z	Zaniskari Pony
143	NoL	Nordland	169	QP	Quarter Pony	195	SW	Swedish Warmblood	221	Zh	Zhemaichu
144	Nor	Noric	170	RH	Racking Horse	196	Tai	Taishuh			
145	NSwH	North Swedish Horse	171	RMH	Rocky Mountain Horse*	197	Tar	Tarpan			
146	NE	Northeastern	172	RD	Russian Don	198	Taw	Tawleed			

Appendix 2. Full list of inherited disorders listed by name of the disorder. Common abbreviations appear in brackets and common synonyms in curly brackets. Total papers found on each disorder, and the number of breeds affected, are shown ('/' denotes none known). The organ system affected and C or D and S or B category are listed. Organ system was abbreviated as follows: Beh = behavioural, CV = cardiovascular, GI = gastrointestinal, Imm = immune system, Int = integument, MS = musculoskeletal, N/S = nervous or sensory, RS = respiratory system, UG = urogenital, Mult = multiple systems affected.

#	Abbr	Condition	Total papers	No breeds affected	Organ system	C or D	S or B
1	Agam	Agammaglobulinemia	2	3	Imm	D	S
2	Alb	Albinism	1	/	Int	D	S
3	AFLD	Angular and flexural limb deformity - dwarfism	1	1	MS	C	S
4	ASD	Anterior segment dysgenesis (ASD)	2	1	N/S	D	S
5	ASD/M	Anterior segment dysgenesis (ASD) /Multiple congenital ocular anomalies (MCOA)	1	1	N/S	C	B
6	BilA	Bilateral aniridia	1	1	N/S	D	S
7	Cat	Cataracts	2	7	N/S	D	S
8	CCD	Central core disease (CCD)	1	/	MS	D	S
9	CCDL	Coat colour dilution lethal (CCDL) {lavender foal syndrome}	2	1	Mult	D	S
10	Cere	Cerebellar abiotrophy	4	3	N/S	D	B
11	Cerv	Cervix abnormalities	1	2	UG	D	S
12	ChLA	Chronic LAD	2	/	RS	D	B
13	CP	Cleft palate	1	/	MS	D	S
14	Col	Colic	2	3	GI	D	B
15	CHA	Complex heart anomalies	1	1	CV	D	B
16	Conf	Conformation defects	3	/	MS	C	B
17	ConfB	Conformation defects - back at the knee		/	MS	C	S
18	A/SCere	Congenital atresia or stenosis of the cerebral aqueduct	1	1	N/S	D	S
19	Mios	Congenital miosis	1	1	N/S	C	S
20	Ncat	Congenital nuclear cataracts	1	1	N/S	D	S
21	CPL	Chronic progressive lymphedema (CPL)	2	3	Int	D	S
22	Cryp	Cryptorchidism	3	1	UG	D	S
23	CSNB	Congenital stationary night blindness (CSNB) {equine night blindness}	5	1	N/S	C	S

#	Abbr	Condition	Total papers	No breeds affected	Organ system	C or D	S or B
24	CVCN	Cervical vertebral compressive myelopathy (CVCM) {Cervical vertebral malformation (CVM), wobbler syndrome}	4	3	MS	D	S
25	Deaf	Deafness associated with pigmentation - white coat or blue eyes	1	1	N/S	C	B
26	Irid	Degeneration of the iridocorneal angle	1	/	N/S	D	S
27	DentOJ	Dental disorders -overjet lesion		1	MS	C	S
28	Dent	Dental disorders – congenital	2	/	MS	C	B
29	DJDhj	Degenerative joint disease (DJD) - hock joint {bone spavin}	2	1	MS	C	S
30	DJD	Degenerative joint disease (DJD) - distal and proximal inter-phalangeal, fetlock and hock joints {bone spavin}	5	2	MS	C	B
31	DOD	Developmental orthopaedic disease (DOD)	1	1	MS	D	B
32	White	Dominant white coat colour	1	/	Int	C	S
33	DSLD	Degenerative suspensory ligament desmitis (DSLD) {equine systemic proteoglycan accumulation (ESPA)}	2	1	MS	D	S
34	Dwar	Dwarfism	2	2	MS	C	S
35	EDM	Equine degenerative myeloencephalopathy (EDM) - predisposition	2	1	N/S	D	S
36	EI	Equine epitheliogenesis imperfecta (EI)	1	/	Int	D	S
37	EIPH	Exercise induced pulmonary haemorrhage (EIPH) {epistaxis}	2	1	RS	D	S
38	Ent	Enterolithiasis	2	4	GI	D	S
39	EPSSM	Equine polysaccharide storage myopathy (EPSSM)	31	22	MS	D	S
40	MelDz	Equine melanotic disease	1	/	Int	C	S
41	ERU	Equine recurrent uveitis (ERU)	2	1	N/S	D	S
42	Eryth	Erythrocyte FAD Deficiency	1	1	CV	D	S
43	FLD	Flexural limb deformity	1	/	MS	C	B
44	Reje	Foal rejection	1	1	Beh	D	S
45	FPIS	Fell pony immunodeficiency syndrome (FPIS) {Fell pony foal syndrome}	9	1	Imm	D	S
46	G6P	Glucose-6-phosphate dehydrogenase deficiency	1	1	CV	D	S
47	GBED	Glycogen branching enzyme deficiency (GBED) {Glycogen storage disease IV, Transglucosidase deficiency, Andersen disease, amylopectinosis}	13	2	GI	D	S
48	Glau	Glaucoma - primary	1	/	N/S	D	S
49	GPT	Guttural pouch tympany	2	2	Mult	D	S
50	GT	Glanzmann thrombasthenia	2	3	CV	D	S
51	Hep	Hepatic fibrosis	1	1	GI	D	S
52	HERDA	Hereditary equine regional dermal asthenia (HERDA) {hyperelastosis cutis,	11	2	Int	D	S

#	Abbr	Condition	Total papers	No breeds affected	Organ system	C or D	S or B
		cutaneous asthenia, dermatosparaxis, Ehlers-Danlos-like syndrome}					
53	Hydr	Hydrops conditions	1	/	UG	D	B
54	HYPP	Hyperkalaemic periodic paralysis (HYPP)	14	4	N/S	D	S
55	IAD	Inflammatory airway disease	1	/	RS	D	B
56	IBH	Insect bite hypersensitivity (IBH) {Recurrent seasonal (summer) pruritis (RSP), sweet itch}	4	4	Int	D	S
57	ILH	Idiopathic left laryngeal hemiplegia	1	3	N/S	D	S
58	IntH	Internal hydrocephalus	1	/	N/S	D	S
59	Ihypo	Iridal hypoplasia	2	2	N/S	C	S
60	JEB	(Herlitz) junctional epidermolysis bullosa (JEB)	10	4	Int	D	S
61	JAL	Juvenile Arabian leukoderma {leukoderma and leukotrichia}	1	1	Int	D	S
62	JIE	Juvenile idiopathic epilepsy	1	1	N/S	D	S
63	Lam	Laminitis	4	1	MS	D	S
64	Leuk	Leukoderma	1	/	Int	D	S
65	Leuktr	Leuktrichia	1	/	Int	D	S
66	StaFert	Stallion subfertility	2	1	UG	D	B
67	LWFS	(Overo) lethal white foal syndrome (LWFS) {Ileocolonic aganglionosis}	14	4	GI	C	S
68	MADD	Multiple acyl-CoA dehydrogenase deficiency (MADD)	1	2	MS	D	S
69	FertDys	Male fertility dysfunction	2	/	UG	D	B
70	MCOA	Equine multiple congenital ocular anomalies syndrome (MCOA)	3	1	N/S	C	B
71	BicBra	Medial luxation of the biceps brachii	1	/	MS	D	S
72	Mega	Megalocornea	1	1	N/S	C	S
73	Mela	Melanoma	7	2	Int	C	S
74	MH	Malignant hyperthermia - susceptibility	5	1	MS	D	S
75	Narc	Narcolepsy and cataplexy	1	4	N/S	D	B
76	NCL	Neuronal / ceroid lipofuscinosis (NCL)	2	2	N/S	D	S
77	ND	Navicular disease	5	1	MS	D	S
78	Oacmc	Osteoarthritis - carpometacarpal joint	1	1	MS	D	S
79	Oash	Osteoarthritis - scapulohumeral	2	3	MS	D	S
80	OAAM	Occipitoatlantoaxial malformation (OAAM)	4	4	MS	D	S
81	OC	Osteochondrosis (OC)	3	3	MS	D	B
82	Ocf	Osteochondrosis (OC) - fetlock joints	2	1	MS	D	S
83	Och	Osteochondrosis (OC) - hock joints	2	1	MS	D	S

#	Abbr	Condition	Total papers	No breeds affected	Organ system	C or D	S or B
84	OC/ND	Osteochondrosis (OC) and navicular disease	1	/	MS	D	B
85	OC/OCD	Osteochondrosis (OC) and Osteochondritis dissecans (OCD) - fetlock and hock joints	2	2	MS	D	B
86	OCD	Osteochondritis dissecans (OCD)	4	7	MS	D	B
87	OP	Organophosphate susceptibility	1	1	MS	D	S
88	Oss	Ossification of the cartilages of the front feet	3	5	MS	D	S
89	Lip	Pedunculated lipomas	1	2	GI	D	S
90	PHA	Pelger-Huët anomaly (PHA)	2	1	CV	D	S
91	Plate	Platelet function defect	2	2	CV	D	B
92	POFf	Palmar osseous fragments (POF) - fetlock joints	8	1	MS	D	S
93	POFh	Plantar osseous fragments (POF) - hock joint	2	1	MS	D	S
94	POF	Palmar/plantar osseous fragments (POF) ?	1	1	MS	D	B
95	POI	Post-operative ileus	1	1	GI	D	S
96	Poly	Polydactyly	1	1	MS	C	S
97	RAO	Recurrent airway obstruction (RAO)	5	/	RS	D	S
98	RER	Recurrent exertional rhabdomyolysis (RER) {rhabdomyolysis, tying-up syndrome}	8	2	MS	D	S
99	Ret	Reticulated leuktrichia	1	1	Int	D	S
100	Rho	Rhodococcus equi - susceptibility	2	1	RS	D	S
101	RLN	Recurrent laryngeal neuropathy {idiopathic laryngeal hemiplegia}	1	/	N/S	D	S
102	RP	Retained placenta	1	1	UG	D	S
103	Sarc	Sarcoid	1	/	Int	D	S
104	SCID	Severe combined immunodeficiency (SCID)	14	1	Imm	D	S
105	SCID/FP	Severe combined immunodeficiency (SCID) /Fell pony immunodeficiency syndrome (FPIS)	1	3	Imm	D	B
106	SCL	Subchondral cystic lesions (SCL)	1	4	MS	D	S
107	SDFT	Superficial digital flexor tendon (SDFT) susceptibility	1	1	MS	D	B
108	IgM	Selective IgM deficiency	2	2	Imm	D	S
109	Shiv	Shivers	2	1	MS	D	B
110	Atav	Skeletal atavism - complete ulnas and fibulas	1	1	MS	C	S
111	Fert	Stallion fertility disorders	3	2	UG	D	B
112	Stereo	Stereotypic behaviour	2	1	Beh	D	B
113	StereoW	Stereotypic behaviour - weaving	2	1	Beh	D	S

#	Abbr	Condition	Total papers	No breeds affected	Organ system	C or D	S or B
114	SteroBW	Stereotypic behaviour - box walking	1	1	Beh	D	S
115	SteroCB	Stereotypic behaviour - crib biting	1	1	Beh	D	S
116	SuscResp	Susceptibility to clinical equine respiratory disease of bacterial origin	1	1	RS	D	B
117	SuscStre	Susceptibility to stress	1	1	Beh	D	B
118	Temp	Temperament	2	1	Beh	D	B
119	Trach	Tracheal collapse	1	1	RS	C	S
120	Twin	Twinning	3	1	UG	D	S
121	Umb	Umbilical hernia	1	/	MS	D	S
122	Feet	Uneven feet	1	/	MS	C	B
123	vWD	vWD	2	2	CV	D	S
124	Wry	Wry nose	2	/	MS	C	S
125	XX	XX sex reversal	4	8	UG	D	S
126	XY	XY sex reversal	4	4	UG	D	S

Appendix 3. Inherited disorders. Disorders are arranged alphabetically by condition. The text below gives details on the conventions used in the table.

Condition: The name of the disorder with common abbreviations in brackets and common synonyms in curly brackets

Organ system: The primary organ system affected by the disorder

C or D: The disorder category (C - Conformation or phenotypic related i.e. disorder reported to result directly from selection for a conformational or phenotypic trait; D - disorder where no evidence of a direct link with conformation in the literature reviewed)

S or B: Disorder description classification (S – Specific i.e. referring to a defined condition with a unifying aetiology; B – Broad i.e. potentially referring to multiple related, or even unrelated, specific conditions)

Breed: The breeds affected by the disorder as denoted by an individual code (And = Andalusian, Ap = Appaloosa, A = Arabian / Arab, B = Belgian, Bre = Breton, Cam = Camargue, Cly = Clydesdale, CN = Cob Normand, Com = Comtois, CoP = Connemara Pony, C = Criollo, DalP = Dales Pony, Fa = Falabella, FP = Fell Pony, Fin = Finnhorse, Fb = Freiberger, F = Friesian, Got = Gotland, Gro = Groningen, Haf = Haflinger, Han = Hanoverian, HP = Highland Pony, Hol = Holsteiner, Ice = Icelandic, L = Lipizzan, Mare = Maremmano, M = Miniature Horse, Morg = Morgan, Murg = Murgese, Must = Mustang, NF = Norwegian Fjord, Old = Oldenburg, P = Paint, Pa = Palomino, PF = Paso Fino, Perc = Percheron, PP = Peruvian Paso, QH = Quarter Horse, RMH = Rocky Mountain Horse, Sb = Saddlebred, SP = Shetland Pony, SH = Shire HorseSGC = South German Coldblood, Stb = Standardbred, Su = Suffolk, TWH = Tennessee Walking Horse, TB = Thoroughbred, Trak = Trakehner, WP = Welsh Pony and Cob).

Type: A two letter code denotes a type of horse that this disorder affects (Co = Cob, DH = Draft (or Draught) Horse, RP = Riding Pony, WBl = Warmblood).

Age of onset: The onset of the disorder and/or clinical signs.

Prevalence: Denoted using the following convention: percentage prevalence of the disorder observed in that breed (country of study using conventional international abbreviations, number of horses from which the prevalence was calculated).

Prognosis: Scored between 0-4 with 0 representing a disease with "a short isolated bout, and a complete return to normal" and 4 representing "imminent death as a direct result of the condition, or related euthanasia.

Mode of inheritance: The way in which the condition is inherited – autosomal recessive, autosomal dominant, x-linked, polygenic, co-dominant or mixed.

References: For each disorder the reference sources that were used in addition to those outlined in the methods section are numbered with brief citation at the end the table (full citations follow the appendices).

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Agammaglobulinemia	Immune	D	S	QH, Stb, TB		<1 year			X-linked	2, 3
Albinism	Integument	D	S			Foetal/birth			Autosomal dominant	4
Angular and flexural limb deformity - dwarfism	Musculoskeletal	C	S	M		Foetal/birth				5
Anterior segment dysgenesis (ASD)	Nervous/sensory	D	S	RMH					Co-dominant	6, 7
Anterior segment dysgenesis (ASD) /Multiple congenital ocular anomalies (MCOA)	Nervous/sensory	C	B	RMH					Co-dominant	8
Bilateral aniridia	Nervous/sensory	D	S	B,					Autosomal dominant	9
Cataracts	Nervous/sensory	D	S	And, A, B, Morg, QH, RMH, TB						10, 11
Central core disease (CCD)	Musculoskeletal	D	S							12
Coat colour dilution lethal (CCDL) {lavender foal syndrome}	Multiple	D	S	A		Foetal/birth		4	Autosomal recessive	13, 14
Cerebellar abiotrophy	Nervous/sensory	D	B	A, Got, Old		<1 year		3 to 4	Autosomal recessive	6, 15-17
Cervix abnormalities	Urogenital	D	S	Cly, SH		Foetal/birth				18
Chronic LAD	Respiratory	D	B							19, 20

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Cleft palate	Musculoskeletal	D	S				All: 0.268% (US, 8954)	3 to 4		21
Colic	Gastrointestinal	D	B	A, M, TB						22, 23
Complex heart anomalies	Cardiovascular	D	B							24
Conformation defects	Musculoskeletal	C	B	TB	RP	Foetal/birth	TB: Back at the knee - 4.16% Base narrow 13.39% Offset knees - 12.94% Upright pasterns - 18.74% Tied below the knee - 1.48% Turned out feet - 30.10% Turned in feet - 19.39% Weak hocks - 5.29% Weak pasterns - 6.35% (UK, 3916)			25, 26
Conformation defects - back at the knee	Musculoskeletal	C	S							27
Congenital atresia or stenosis of the cerebral aqueduct	Nervous/sensory	D	S						X-linked	28
Congenital miosis	Nervous/sensory	C	S	RMH						29
Congenital nuclear cataracts	Nervous/sensory	D	S	Morg						10
Chronic progressive lymphedema (CPL)	Integument	D	S	B, Cly, SH				2 to 4		30
Cryptorchidism	Urogenital	D	S	TB			All: 2-8% (?, ?)		Polygenic	31-33
Congenital stationary night blindness (CSNB) {equine night blindness}	Nervous/sensory	C	S	Ap		Foetal/birth	Ap: 33% (CA, 30)	1 to 3	Autosomal recessive	9, 34-37

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Cervical vertebral compressive myelopathy (CVCM) {Cervical vertebral malformation (CVM), wobbler syndrome}	Musculoskeletal	D	S	QH, TWH, TB	WBl			2-4		17, 38-40
Deafness associated with pigmentation - white coat or blue eyes	Nervous/sensory	C	B	P				Life long deafness		41
Degeneration of the iridocorneal angle	Nervous/sensory	D	S							42
Dental disorders - overjet lesion	Musculoskeletal	C	S	QH		Foetal/birth				43
Dental disorders - congenital	Musculoskeletal	C	B							44
Degenerative joint disease (DJD) - hock joint {bone spavin}	Musculoskeletal	C	S	Han	WBl		Han:12.0%; 14.6% - multiply affected horses (DE, 5231)			45, 46
Degenerative joint disease (DJD) - distal and proximal inter-phalangeal, fetlock and hock joints {bone spavin}	Musculoskeletal	C	B	Han, Ice	WBl		Ice: 30.3% (IS, 614); Han: 17.7% (DE, 3748)			47-51
Developmental orthopaedic disease (DOD)	Musculoskeletal	D	B	TB						52
Dominant white coat colour	Integument	C	S			Foetal/birth		4	Autosomal dominant	53

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Degenerative suspensory ligament desmitis (DSLD) {equine systemic proteoglycan accumulation (ESPA)}	Musculoskeletal	D	S	PP						6, 54
Dwarfism	Musculoskeletal	C	S	F, M		Foetal/birth			Autosomal recessive	55, 56
Equine degenerative myeloencephalopathy (EDM) - predisposition	Nervous/sensory	D	S	Morg		1 to 5 years		3		16, 17
Equine epitheliogenesis imperfecta (EI)	Integument	D	S							6
Exercise induced pulmonary haemorrhage (EIPH) {epistaxis}	Respiratory	D	S	TB			TB: 2.1% (ZA, ?)		Polygenic	1, 57
Enterolithiasis	Gastrointestinal	D	S	A, M, Morg, Sb						58, 59

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Equine polysaccharide storage myopathy (EPSSM)	Musculoskeletal	D	S	And, Ap, A, B, Cly, CN, Haf, Hol, M, Morg, Must, NF, P, Perc, QH, RMH, Sb, SH, Stb,TWH, TB, WP	DH, WBl	5 to 10 years	B: 36% (US, 103); QH: 6% (US, 164); QH: 6-12% (US, 164); QH: 11.3% (US, 200), P: 4.5% (US, 180); DH: 86% (US, 7), Morg: 64% (US, 11), A: 62% (US, 34), RP 62% (US, 8), Ap: 56% (US, 16), TWH: 50% (US, 6), QH: 41% (US, 68), P: 33% (US, 27), WBl: 28% (US, 14), TB: 27% (US, 22).	0-3	Autosomal dominant	6, 17, 60-88
Equine melanotic disease	Integument	C	S							89
Equine recurrent uveitis (ERU)	Nervous/sensory	D	S	Ap	WBl		10% (US, ?)			42, 90
Erythrocyte FAD Deficiency	Cardiovascular	D	S	Must						91
Flexural limb deformity	Musculoskeletal	C	B						Autosomal dominant	92
Foal rejection	Behavioural	D	S	A			A: 5.13% (US, 702)			93
Fell pony immunodeficiency syndrome (FPIS) {Fell pony foal syndrome}	Immune	D	S	FP		<1 year	FP: 40-60% (NL, UK, ?)	4	Autosomal recessive	2, 94-101
Glucose-6-phosphate dehydrogenase deficiency	Cardiovascular	D	S	Sb						91

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Glycogen branching enzyme deficiency (GBED) {Glycogen storage disease IV, Transglucosidase deficiency, Andersen disease, amylopectinosis}	Gastrointestinal	D	S	P, QH		Foetal/birth	QH: 8.3% (US, 338), P: 7.1% (US, 197); QH: 11.0% (US, 200); P: 3.9% (US, 180)	4	Autosomal recessive	6, 60, 64, 65, 71, 78, 91, 102-108
Glaucoma - primary	Nervous/sensory	D	S							42
Guttural pouch tympany	Multiple	D	S	A, P		<1 year			Polygenic or mixed monogenic-polygenic	109, 110
Glanzmann thrombasthenia	Cardiovascular	D	S	Old, QH, TB				3		111, 112
Hepatic fibrosis	Gastrointestinal	D	S	Fb	DH				Autosomal recessive	113
Hereditary equine regional dermal asthenia (HERDA) {hyperelastosis cutis, cutaneous asthenia, dermatosparaxis, Ehlers-Danlos-like syndrome}	Integument	D	S	P, QH		1 to 5 years	QH: 3.5% (US, 200), P: 1.7% (US, 180); QH: 3.5% (US, 1079)	2 to 4	Autosomal recessive	6, 60, 64, 65, 103, 114-119
Hydrops conditions	Urogenital	D	B		DH	Foetal/birth				120

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Hyperkalaemic periodic paralysis (HYPP)	Nervous/sensory	D	S	Ap, C, P, QH		1 to 5 years	QH: 4.4% (US, 978); QH: 1.5% (US, 200). P: 4.5% (US, 180)	0 to 4	Autosomal dominant	6, 64, 65, 71, 78, 103, 121-127
Inflammatory airway disease	Respiratory	D	B				TB: 13.0% (UK, 148)			128
Insect bite hypersensitivity (IBH) {Recurrent seasonal (summer) pruritis (RSP), sweet itch}	Integument	D	S	Ice, SP, SH, WP	DH	1 to 5 years	SP: 8.8% (NL, 6073); SH: 11.6% (UK, 1088), SH: 37.7% (DE, 77)			6, 129-131
Idiopathic left laryngeal hemiplegia	Nervous/sensory	D	S	B, Cly, Perc	DH		B, Perc and Cly: 35% (US, 183), B: 42% (US, 97), Perc: 31% (US, 58), Cly: 17% (US, 28)			132
Internal hydrocephalus	Nervous/sensory	D	S							9
Iridal hypoplasia	Nervous/sensory	C	S	QH, RMH			RMH: 14% (US, 514)		Autosomal dominant	28, 29
(Herlitz) junctional epidermolysis bullosa (JEB)	Integument	D	S	B, Bre, Com, Sb	DH	Foetal/birth	B: 32% (US, 176), B: 17.1% (CA, 328), Bre: 15.9% (FR, 63), Com: 7.8% (FR, 51); Sb: ~4% (?, ?); Sb: 5.1% (US, 175)	2-4 (euthanased)	Autosomal recessive	6, 64, 65, 124, 133-137

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Juvenile Arabian leukoderma {leukoderma and leukotrichia}	Integument	D	S	A		1 to 5 years				4
Juvenile idiopathic epilepsy	Nervous/sensory	D	S	A		<1 year		2 to 3		138
Laminitis	Musculoskeletal	D	S	QH		Foetal/birth				6, 139-141
Leukoderma	Integument	D	S							4
Leuktrichia	Integument	D	S							4
Stallion sub-fertility	Urogenital	D	B	Han						142, 143
(Overo) lethal white foal syndrome (LWFS) {Ileocolonic aganglionosis}	Gastrointestinal	C	S	M, P, QH, TB		Foetal/birth	P: 21.3% (US, 180)	4	Autosomal recessive	4, 6, 21, 60, 64, 65, 103, 124, 144-149
Multiple acyl-CoA dehydrogenase deficiency (MADD)	Musculoskeletal	D	S	Gro, Trak		> 10 years		4		150
Male fertility dysfunction	Urogenital	D	B							151, 152
Equine multiple congenital ocular anomalies syndrome (MCOA)	Nervous/sensory	C	B	RMH		Foetal/birth	RMH: 50% (CA, 97); RMH: 14% (US, 514)		Autosomal dominant	29, 153, 154

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Medial luxation of the biceps brachii	Musculoskeletal	D	S					1-4		155
Megalocornea	Nervous/sensory	C	S	RMH			RMH: 8% (US, 514)			29
Melanoma	Integument	C	S	Cam, L		5 to 10 years	Cam: 31.4% (FR, 264) - (67% at ages >15 years); L: 50% (AT, 296)	0-1		156-162
Malignant hyperthermia - susceptibility	Musculoskeletal	D	S	QH			QH: 1.3% (US, 225)	Potentially fatal (est mortality 34%)	Autosomal dominant	64, 71, 163-165
Narcolepsy and cataplexy	Nervous/sensory	D	B	M, Morg, SP, Su		Foetal/birth		1		16
Neuronal / ceroid lipofuscinosis (NCL)	Nervous/sensory	D	S	Ice, PP		<1 year		4	Autosomal recessive	166, 167
Navicular disease	Musculoskeletal	D	S	Han,			Han: ~20% (DE, 3748); Han: 25.8% (DE, 5231) - 14.6% - multiply affected horses			45, 47, 168-170
Osteoarthritis - carpometacarpal joint	Musculoskeletal	D	S	A,						171
Osteoarthritis - scapulohumeral	Musculoskeletal	D	S	Fa, M, SP,				3-4		172, 173
Occipitoatlantoaxial malformation (OAAM)	Musculoskeletal	D	S	Ap, A, Morg, TB		Foetal/birth		0-4		16, 40, 174, 175
Osteochondrosis (OC)	Musculoskeletal	D	B	QH, Sb, TB	DH					1, 176, 177
Osteochondrosis (OC) - fetlock joints	Musculoskeletal	D	S	SGC						178, 179

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Osteochondrosis (OC) - hock joints	Musculoskeletal	D	S	SGC						178, 179
Osteochondrosis (OC) and navicular disease	Musculoskeletal	D	B							6
Osteochondrosis (OC) and Osteochondritis dessicans (OCD) - fetlock and hock joints	Musculoskeletal	D	B	Han, SGC	DH					180, 181
Osteochondritis dessicans (OCD) – incl fetlock joint	Musculoskeletal	D	B	B, Cly, Han, Mare, Perc, SGC, SH	DH					182, 183, 184, 185
Organophosphate susceptibility	Musculoskeletal	D	S	M						186
Ossification of the cartilages of the front feet	Musculoskeletal	D	S	CoP, DalP, FP, Fin, HP,	Co	1 to 5 years	Norwegian coldblood: 11.5% (NO, 392)			187-189
Pedunculated lipomas	Gastrointestinal	D	S	A, Sb,						190
Pelger-Huët anomaly (PHA)	Cardiovascular	D	B	And, A,						191, 192
Platelet function defect	Cardiovascular	D	S	TB		1 to 5 years				193, 194
Palmar osseous fragments (POF) - fetlock joints	Musculoskeletal	D	S	Han, SGC			Han: 27.91% (DE, 5102); Han: 23.5% (DE, 5231) - 14.6% - multiply affected horses			45, 170, 178, 179, 195-198

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Plantar osseous fragments (POF) - hock joint	Musculoskeletal	D	B	Han,			Han: 9.25% (DE, 5102); Han: 9.2% (DE, 5231) - 14.6% - multiply affected horses			170, 195
Palmar/plantar osseous fragments (POF)	Musculoskeletal	D	S	Stb						199
Post-operative ileus	Gastrointestinal	C	S	A,						200
Polydactyly	Musculoskeletal	D	S	Murg					Autosomal dominant	201
Recurrent airway obstruction (RAO)	Respiratory	D	S		RP				Polygenic	1, 6, 202-204
Recurrent exertional rhabdomyolysis (RER) {rhabdomyolysis, tying-up syndrome}	Musculoskeletal	D	S	Stb, TB			TB: 7.7% (JP, 6538); TB: 5% (?, ?)	1 to 3	Autosomal dominant	6, 62, 70, 71, 78, 205-207
Reticulated leuktrichia	Integument	D	S	QH,		1 to 5 years				4
Rhodococcus equi - susceptibility	Respiratory	D	S	TB		Foetal/birth		3 to 4		208, 209
Recurrent laryngeal neuropathy {idiopathic laryngeal hemiplegia}	Nervous/sensory	D	S							210
Retained placenta	Urogenital	D	S	F			F: 54% (NL, 436)			211
Sarcoid	Integument	D	S			5 to 10 years				212

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Severe combined immunodeficiency (SCID)	Immune	D	B	A		<1 year	A: 0.18% (US, 250); A: 2.8.% (UK, 106)	4	Autosomal recessive	2, 6, 64, 65, 99, 103, 124, 213-219
Severe combined immunodeficiency (SCID) /Fell pony immunodeficiency syndrome (FPIS)	Immune	D	S	FP, PF, TB		Foetal/birth		4		220
Subchondral cystic lesions (SCL)	Musculoskeletal	D	B	B, Cly, Perc, SH	DH					183
Superficial digital flexor tendon (SDFT) injury	Musculoskeletal	D	S	TB			TB: 11.31% (JP, 8198)			221
Selective IgM deficiency	Immune	D	B	A, QH,		1 to 5 years		4		2, 99
Shivers	Musculoskeletal	C	S	B	DH	> 10 years	B: 18% (US, 103)	4		80, 222
Skeletal atavism - complete ulnas and fibulas	Musculoskeletal	D	B	M,		Foetal/birth		3 to 4		223
Stallion fertility	Urogenital	D	B	SP, TWH						6, 224, 225
Stereotypic behaviour	Behavioural	D	S	TB			All: 3.5% (CH, 2536)			226, 227
Stereotypic behaviour – weaving	Behavioural	D	S	TB			TB: 4.17% (CA, 263)			228, 229

Condition	Organ system	C or D	S or B	Breed	or type	Age at onset	Prevalence	Prognosis	Mode of inheritance	References
Stereotypic behaviour - box walking	Behavioural	D	S	A			A: 7.32% (CA, 81)			229
Stereotypic behaviour - crib biting	Behavioural	D	B	TB			TB: 6.82% (CA, 263)			229
Susceptibility to clinical equine respiratory disease of bacterial origin	Respiratory	D	B	WP						230
Susceptibility to stress	Behavioural	D	B	TB						227
Temperament	Behavioural	C	S	TB						231, 232
Tracheal collapse	Respiratory	D	S	M						233
Twinning	Urogenital	D	S	TB		Foetal/birth	TB: 3.5% (PL, 1910); TB: 3.5% (PL, 2033)		Polygenic	234-236
Umbilical hernia	Musculoskeletal	C	B			Foetal/birth	WBl: 29.5% (NL, 44)	1		237
Uneven feet	Musculoskeletal	D	S				WBl: 5.3% (NL, 44840)			238
vWD	Cardiovascular	C	S	QH, TB				4	Autosomal recessive	239, 240
Wry nose	Musculoskeletal	D	S			Foetal/birth				44, 174
XX sex reversal	Urogenital	D	S	And, Ap, A, P, Pa, PF, QH, Sb, D30		Foetal/birth		Unable to reproduce	Autosomal recessive	6, 241-243
XY sex reversal	Urogenital	D	S	A, Han, QH, TB+D30		Foetal/birth			X-linked	6, 32, 241, 244

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