REVOLUTIONARY DNA INSIGHTS INTO YOUR HORSE HEALTH

Report type A Thoroughbred, Arabian and similar

EquineTest: Horse Genetic Sequencing

Customer and Order Details

Order ID	
Customer	
Customer's Veterinarian	
Date of sample collection	
Date of sample receivement	
Raw data sent to customer	Delivered on 12.3.2024
Report	Report Type A - Thoroughbred, Arabian and similar Version 0.1 Generated on 13.3.2024

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Horse Details

Name	
Date of birth	
Registration number	
Microchip	
Breed	
Sex	
Phenotype notes	
Sire details Breed Registration number Phenotype notes	
Dam details Breed Registration number Phenotype notes	

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Report summary

The examined horse deviates from the norm in 2 observed traits.

1. In the **Px trait, the horse is homozygous (Px/Px)**. Px is associated with the disease MIM (Muscle integrity myopathy previously called PSSM2)(, but its significance remains unclear and a statistically significant correlation with MIM_Px has not yet been sufficiently proven. Therefore, Px is only considered a risk factor.
2. In the **PRKDC trait, the horse is heterozygous (n/SCID)**. Since this trait is associated with the autosomal recessive disease SCID (Severe Combined Immunodeficiency), the horse is healthy but is a carrier of the trait. The variant/mutation has a 50% chance of being passed on to the offspring. If the foal inherits the mutation in both variants of the PRKDC gene, the disease will manifest, resulting in the foal's death within 6 months of age.

Breeding

Breeding is possible. However, due to the found mutations, it is necessary to select stallions that are not carriers of the mutated PRKDC-SCID and Px traits, and the related traits for MIMP2, P3, P4, P8, K1, even in the heterozygous form. In such a case, there would be a 25-50% risk of giving birth to an affected foal.

Management

Given the presence of the Px trait, if the horse exhibits signs of muscle problems (stiffness, cramps), we recommend adhering to a diet low in starch and sugars and regular movement without overexertion at once.

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Part 1 - Main markers with interpretation

Category	Disorder
Phene	**Lavender foal syndrome/ Coat Color Dilution Lethal**
Description	Lavender foal syndrome (LFS), also known as Coat Color Dilution Lethal (CCDL), causes neurological dysfunction in newborn foals (an inability to stand, unusually light coat color and seizures). LFS is thought to be created by an autosomal recessive gene at MYO5A. This genetic disease is untreatable and lethal. Affected individuals are mostly dying within a few hours or days. A fatal condition of Arabian horses and breeds with Arabian blood, it is often associated with those that have Egyptian bloodlines.
Breed(s)	Arabian Horse
Chromosome	1
Gene	MYO5A
Allele	MYO5A
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Congenital stationary night blindness, TRPM1-related**
Description	Congenital stationary night blindness (CSNB) is characterized by a nonprogressive scotopic visual deficit. This disorder is associated with mutation in TRMP1 gene. Horses with CSNB likely have normal vision during daylight but they may exhibit apprehension in dimly lit conditions and may show problematic behavior in light and dark condition. Affected individuals occasionally manifest an improper eye alignment and involuntary eye movement. Many affected horses can be successfully managed. This genetic disorder is also linked to coat color in horses (white spotting).
Breed(s)	Thoroughbred (Horse), American Miniature Horse (Horse) Appaloosa (Horse) British Spotted Pony, Spotted Pony, United Kingdom of Great Britain and Northern Ireland (Horse) English Spotted Pony, Australia (Horse) Knabstrupper (Horse) Noric (Horse) Pony Of the Americas, Germany (Horse)
Chromosome	1

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Gene	TRPM1
Allele	TRP2
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Warmblood Fragile Foal Syndrome Type 1/ kyphoscoliotic Ehlers-Danlos syndrome, PLOD1-related**
Description	Warmblood Fragile Foal Syndrome Type 1 (WFFS) is considered in cases of abortion, stillbirth, skin lesions and malformations of the skin in neonatal foals. Affected horses show the first signs of the disease immediately or a few days after birth. The skin is extremely sensitive and fragile. Wounds in the skin are often affected by secondary infections, which heal very poorly and are often fatal for the foal. Affected individuals usually do not live more than a few weeks. The suffering of affected animals is often ended by euthanasia.
Breed(s)	Thoroughbred (Horse) Warmblood breeds and related breeds
Chromosome	2
Gene	PLOD1
Allele	LH1
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Cerebellar abiotrophy**
Description	Equine Cerebellar Abiotrophy (CA) is a neurological disease. CA is characterized by post-natal degeneration of the neuron cells in cerebellum. Symptoms of CA are intention head tremors, ataxia, exaggerated or paddling action of the forelegs, a wide-based stance, and a lack of menace response. Affected horses may startle easily and fall and rise from a reclining position could be difficult. CA usually appear between six weeks and four months of age.
Breed(s)	Arab (Horse)Bashkir Curly (Horse)Trakehner (Horse)Welsh Pony (Horse)
Chromosome	2
Gene	MUTYH, TOE1
Allele	MUTYH, TOE1
Mode of inheritance	Autosomal recessive

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Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Muscle integrity myopathy_P3**
Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_P3 is caused by affecting the filamin C protein in muscles. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	all
Chromosome	4
Gene	FLNC
Allele	P3
Mode of inheritance	Autosomal dominant - variable manifestation
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Hypoparathyroidism, RAPGEF5-related**
Description	Horses with Hypoparathyroidism are presented with tachycardia, hyperhidrosis, diarrhea, and muscle rigidity or stiff gait. Absence of parathyroid tissue was recorded in all of the cases. Foals with this disorder lived from a few days to several weeks.
Breed(s)	Thoroughbred (Horse)
Chromosome	4
Gene	RAPGEF5
Allele	RAP
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

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Category	Disorder
Phene	**Muscle integrity myopathy_K1**
Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_K1 is caused by affecting the Kollagen VI in muscles. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	all
Chromosome	6
Gene	COL6A3
Allele	K1
Mode of inheritance	Autosomal dominant - variable manifestation
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Muscle integrity myopathy_P8**
Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_P8 is caused by affecting the thiolreductase enzyme in muscles. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	all
Chromosome	6
Gene	PYROXD1
Allele	P8

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Mode of inheritance	Autosomal dominant - variable manifestation
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Severe combined immunodeficiency disease**
Description	Severe combined immunodeficiency disease (SCID) results in the absence of immune cells (B and T lymphocytes). Affected foals suffer from a variety of infections that are unresponsive to veterinary therapy. These infections are often caused by agents that are rarely fatal in immunocompetent animals but foals carrying the disease do not have a developed immune system. Because of this, the infection of the affected individual is usually fatal. Disease arises usually at 1 month of age and foal dies by 5 months of age.
Breed(s)	Arab (Horse)
Chromosome	9
Gene	PRKDC
Allele	PRKDC
Mode of inheritance	Autosomal recessive
Result	**Heterozygote N/MUT**
Simplified interpretation	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%.

Category	Disorder
Phene	**Polysaccharide storage myopathy**
Description	Polysaccharide storage myopathy (PSSM1) disorder is caused by abnormal storage of sugar (glycogen) in the muscles. It is common that horses refuse working or seem lazy, but they are suffering from painful muscle cramping. Disease manifestation is individual, from a few episodes per year to constant escalating muscle pain. Muscle atrophy, exertional rhabdomyolysis, wrong posture, problematic walk or progressive weakness are some of the symptoms of PSSM1.
Breed(s)	All
Chromosome	10
Gene	GYS1
Allele	P1
Mode of inheritance	Autosomal incomplete dominant
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

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Category	Disorder
Phene	**Glanzmann's Thrombasthenia**
Description	Glanzmann's thrombasthenia (GT, coagulopathy) is a blood clotting disorder in horses. The main symptom is repeated nosebleeds and frequent other bleeding. The mutation affects the functions of blood platelets (thrombocytes) and thus impairs blood clotting.
Breed(s)	Thoroughbred (Horse), Quarter Horse (Horse)
Chromosome	11
Gene	ITGA2B
Allele	IA1
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Muscle integrity myopathy_P2**
Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_P2 is caused by affecting the myotilin protein in muscles. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	all
Chromosome	14
Gene	MYOT
Allele	P2
Mode of inheritance	Autosomal dominant - variable manifestation
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Muscle integrity myopathy_P4**

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Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_P4 is caused by affecting myosin 3 protein in muscles. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	all
Chromosome	14
Gene	MYOZ3
Allele	P4
Mode of inheritance	Autosomal dominant - variable manifestation
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Muscle integrity myopathy_Px**
Description	Muscle integrity myopathy (MIM) previously called PSSM2 is a term used for two different muscle diseases. The former is known as MFM (myofibrillar myopathy) and the latter as RER (recurrent exercise rhabdomyolysis). Unlike the long-known PSSM1, MIM (PSSM2) is not associated with the problem of glycogen accumulation.MIM is a problem in the horse's muscle structure that results in damage, tearing and breakdown of muscle fibers. MIM_Px is caused by affecting the Ca2+ ion channel in muscles resulting in RER manifestations of varying intensity. The main symptoms of MIM are reluctance to move, abnormal gait, pain, stiffness, muscle tremors, excessive sweating, inability to stand, muscle atrophy, pigmenturia (brown coloured urine), elevated serum creatine kinase (CK) and aspartate aminotransferase (AST).
Breed(s)	Thoroughbred and Arabian horse
Chromosome	16
Gene	CACNA2D3
Allele	Px
Mode of inheritance	Autosomal dominant - variable manifestation

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Result	**Homozygote MUT/MUT**
Simplified interpretation	Horse affected by MIM_Px of varying intensity. The causality is not yet well understood. Variant/mutation will be passed to the offspring.

Category	Disorder
Phene	**Occipitoatlantoaxial malformation**
Description	Occipitoatlantoaxial malformation (OAAM) is a rare developmental disease. It causes defects in the first cervical vertebra (atlas) resembles the base of the skull (occiput) and the second cervical vertebra (axis) resembles the atlas. Affected individuals has problems with posture, standing and head rotation. Ataxia in varying degrees is common.
Breed(s)	Arab (Horse)
Chromosome	18
Gene	HOXD3
Allele	hD3
Mode of inheritance	Autosomal recessive
Result	**Homozygote N/N**
Simplified interpretation	Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	Androgen insensitivity syndrome
Description	Androgen insensitivity syndrome (AIS) causes an individual who is genetically male to appear female. The affected male is sterile, small malformations of the external genitalia may appear. Females carrying the mutation are only carriers.
Breed(s)	Thoroughbred (Horse)
Chromosome	X
Gene	AR
Allele	AIS2
Mode of inheritance	X-linked recessive
Result	**Homozygote N/N**
Simplified interpretation	Female: Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Androgen insensitivity syndrome**
Description	Androgen insensitivity syndrome (AIS) causes an individual who is genetically male to appear female. The affected individual is sterile,

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	small malformations of the external genitalia may appear. Females carrying the mutation are only carriers.
Breed(s)	Thoroughbred (Horse)
Chromosome	X
Gene	AR
Allele	AIS3
Mode of inheritance	X-linked recessive
Result	**Homozygote N/N**
Simplified interpretation	Female: Clinically normal, no possible transmission to offspring.

Category	Disorder
Phene	**Androgen insensitivity syndrome**
Description	Androgen insensitivity syndrome (AIS) causes an individual who is genetically male to appear female. The affected individual is sterile, small malformations of the external genitalia may appear. Females carrying the mutation are only carriers.
Breed(s)	Thoroughbred (Horse)
Chromosome	X
Gene	AR
Allele	AIS5
Mode of inheritance	X-linked recessive
Result	**Homozygote N/N**
Simplified interpretation	Female: Clinically normal, no possible transmission to offspring.

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Part 2 - All markers

Category	Phene	Chr	Gene	Allele	Result
Disorder	Dwarfism, ACAN-related	1	ACAN	D1	N/N
Disorder	Dwarfism, ACAN-related	1	ACAN	D2	N/N
Disorder	Dwarfism, ACAN-related	1	ACAN	D3*	N/N
Disorder	Dwarfism, ACAN-related	1	ACAN	D4	N/N
Disorder	Lavender foal syndrome/ Coat Color Dilution Lethal	1	MYO5A	MYO5A	N/N
Disorder	Congenital stationary night blindness, TRPM1-related	1	TRPM1	TRP2	N/N
Disorder	Hydrocephalus	1	B3GALNT2	Hr	N/N
Disorder	Hereditary equine regional dermal asthenia (HERDA)	1	PPIB	PPIB	N/N
Disorder	Coat colour, albinism, oculocutaneous type VI	1	SLC24A5	Tiger-eye 1	N/N
Disorder	Coat colour, albinism, oculocutaneous type VI	1	SLC24A5	Tiger-eye 2	N/N
Colour	Coat colour, Leopard Complex Spotting	1	TRPM1	LP	N/N
Disorder	Warmblood Fragile Foal Syndrome Type 1/ kyphoscoliotic Ehlers-Danlos syndrome, PLOD1-related	2	PLOD1	LH1	N/N
Disorder	Cerebellar abiotrophy	2	MUTYH, TOE1	MUTYH, TOE1	N/N
Colour	Coat colour, dominant white	3	KIT	W2	N/N
Colour	Coat colour, dominant white	3	KIT	W4	N/N
Colour	Coat colour, dominant white	3	KIT	W6	N/N
Colour	Coat colour, dominant white	3	KIT	W9	N/N
Colour	Coat colour, dominant white	3	KIT	W15	N/N
Colour	Coat colour, dominant white	3	KIT	W16	N/N

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Colour	Coat colour, dominant white	3	KIT	w17a	N/N
Colour	Coat colour, dominant white	3	KIT	W17b	N/N
Colour	Coat colour, dominant white	3	KIT	W19	N/N
Colour	Coat colour, dominant white	3	KIT	W20	N/N
Colour	Coat colour, dominant white	3	KIT	W1	N/N
Colour	Coat colour, dominant white	3	KIT	W3	N/N
Colour	Coat colour, dominant white	3	KIT	W7	N/N
Colour	Coat colour, dominant white	3	KIT	W8	N/N
Colour	Coat colour, dominant white	3	KIT	W11	N/N
Colour	Coat colour, dominant white	3	KIT	W13	N/N
Colour	Coat colour, dominant white	3	KIT	W18	N/N
Colour	Coat colour, dominant white	3	KIT	SB1 (sabino 1)	N/N
Colour	Coat colour, dominant white	3	KIT	W5	N/N
Colour	Coat colour, dominant white	3	KIT	W10	N/N
Colour	Coat colour, dominant white	3	KIT	W12	N/N
Colour	Coat colour, dominant white	3	KIT	W14	N/N
Colour	Coat colour, dominant white	3	KIT	W22	N/N
Colour	Coat colour, dominant white	3	KIT	W21	N/N
Colour	Coat colour, dominant white	3	KIT	W23	N/N
Colour	Coat colour, dominant white	3	KIT	W24	N/N
Colour	Coat colour, dominant white	3	KIT	W25	N/N
Colour	Coat colour, dominant white	3	KIT	W26	N/N
Colour	Coat colour, dominant white	3	KIT	W27	N/N

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Colour	Coat colour, dominant white	3	KIT	W28	N/N
Colour	Coat colour, dominant white	3	KIT	W30	N/N
Colour	Coat colour, dominant white	3	KIT	W31	N/N
Colour	Coat colour, dominant white	3	KIT	W32	N/N
Colour	Coat colour, dominant white	3	KIT	W33	N/N
Colour	Coat colour, dominant white	3	KIT	W34	N/N
Colour	Coat colour, extension	3	MC1R	e	N/N
Colour	Coat colour, extension	3	MC1R	e^a	N/N
Disorder	Myotonia	4	CLCN1	M	N/N
Disorder	Muscle integrity myopathy_P3	4	FLNC	P3	N/N
Disorder	Hypoparathyroidism, RAPGEF5-related	4	RAPGEF5	RAP	N/N
Disorder	JEB - Junctional Epidermolysis bullosa, LAMC2-related	5	LAMC2	LAMC2	N/N
Disorder	Muscle integrity myopathy_K1	6	COL6A3	K1	N/N
Disorder	Muscle integrity myopathy_P8	6	PYROXD1	P8	N/N
Colour	Coat colour, white spotting, PAX3-related	6	PAX3	SW2	N/N
Colour	Coat colour, white spotting, PAX3-related	6	PAX3	SW4	N/N
Colour	Coat colour, white spotting, PAX3-related	6	PAX3	SW10	N/N
Disorder/Colour	Coat colour silver (Multiple ocular defects)	6	PMEL	Z1	N/N
Disorder	Multiple ocular defects	6	PMEL	Z2	N/N
Colour	Coat colour, phaeomelanin dilution, MFSD12-related	7	MFSD12_	Mu	N/N
Disorder	Naked foal syndrome	7	ST14	FS	N/N

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Disorder	Epidermolysis bullosa, junctionalis, LAMA3-related	8	LAMA3	J	N/N
Disorder	Hoof wall separation syndrome	8	SERPINB11	HWSD	N/N
Colour	Coat colour, dun	8	TBX3	nd1	N/N
Colour	Coat colour, dun	8	TBX3	nd2=d ?	N/N
Disorder	Severe combined immunodeficiency disease	9	PRKDC	PRKDC	N/MUT
Disorder	Polysaccharide storage myopathy	10	GYS1	P1	N/N
Disorder	MH - Malignant hyperthermia	10	RYR1	RYR1	N/N
Disorder	Thrombasthenia	11	ITGA2B	IA2	N/N
Disorder	Curly coat	11	KRT25	Crd	N/N
Disorder	Myositis, immune-mediated	11	MYH1	IMM	N/N
Disorder	Glanzmann's Thrombasthenia	11	ITGA2B	IA1	N/N
Disorder	Hyperkalemic Periodic Paralysis, HYPP	11	SCN4A	H	N/N
Disorder	Curly coat with hypotrichosis	11	SP6	SP6	N/N
Disorder	Ocular squamous cell carcinoma	12	DDB2	SCC	N/N
Disorder	Dwarfism, Friesian	14	B4GALT7	D	N/N
Disorder	Night blindness, congenital stationary, GRM6-related	14	GRM6	GRM	N/N
Disorder	Muscle integrity myopathy_P2	14	MYOT	P2	N/N
Disorder	Muscle integrity myopathy_P4	14	MYOZ3	P4	N/N
Colour	Coat colour, champagne	14	SLC36A1	Ch ?	N/N
Colour	Coat colour, white spotting	16	MITF	SW5	N/N
Colour	Coat colour, white spotting	16	MITF	SW6	N/N
Colour	Coat colour, white spotting	16	MITF	SW8, MITF^244Glu	N/N
Colour	Coat colour, white spotting	16	MITF	SW7	N/N

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Colour	Coat colour, white spotting	16	MITF	SWM	N/N
Colour	Coat colour, white spotting	16	MITF	SW3	N/N
Colour	Coat colour, white spotting	16	MITF	SW1	N/N
Colour	Coat colour, white spotting	16	MITF	SW2	N/N
Colour	Coat colour, white spotting	16	MITF	SW9	N/N
Disorder	Muscle integrity myopathy_Px	16	CACNA2D3	Px	MUT/MUT
Disorder	Megacolon	17	EDNRB	O/LWO	N/N
Disorder	Occipitoatlantoaxial malformation	18	HOXD3	hD3	N/N
Colour	Coat colour, cream dilution	21	SLC45A2	C^Cr	N/N
Colour	Coat colour, cream dilution	21	SLC45A2	C^prl	N/N
Colour	Coat colour, cream dilution	21	SLC45A2	C^sun	N/N
Colour	Coat colour, cream dilution	21	SLC45A2	C^sno	N/N
Colour	Coat colour, agouti	22	ASIP	A	N/N
Disorder	Gaitedness	23	LOC100147177	Ga	N/N
Colour	Coat colour, grey/gray	25	STX17	G	N/N
Disorder	Glycogen storage disease IV	26	GBE1	gbed	N/N
Disorder	Immunodeficiency syndrome, SLC5A3-related	26	SLC5A3	IS	N/N
Disorder	Skeletal atavism	PAR	SHOX	Del-1	N/N
Disorder	Skeletal atavism	PAR	SHOX	Del-2	N/N
Disorder	Androgen insensitivity syndrome (AIS)	X	AR	AIS1	N/N
Disorder	Androgen insensitivity syndrome (AIS)	X	AR	AIS4	N/N
Disorder	Incontinentia pigmenti	X	IKBKG	IKB	N/N
0	Brindle 1	X	MBTPS2	B1	N/N
Disorder	Androgen insensitivity syndrome	X	AR	AIS2	N/N
Disorder	Androgen insensitivity syndrome	X	AR	AIS3	N/N

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Disorder	Androgen insensitivity syndrome	X	AR	AIS5	N/N
Disorder	Ovotesticular DSD (Disorder of Sexual Development)	Y	SRY	DSD	N/N
Disorder	Dwarfism, ACAN-related	1	ACAN	D1	N/N

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Dictionary

Allele

An allele is one of two or more versions of DNA sequence (a single base or a segment of bases) at a given genomic location. An individual inherits two alleles, one from each parent, for any given genomic location where such variation exists. If the two alleles are the same, the individual is **homozygous** for that allele. If the alleles are different, the individual is **heterozygous**.

Autosomal Dominant Disorder

Autosomal dominant is a pattern of inheritance characteristic of some genetic disorders. "Autosomal" means that the gene in question is located on one of the numbered, or non-sex, chromosomes. "Dominant" means that a single copy of the mutated gene (from one parent) is enough to cause the disorder. A child of a person affected by an autosomal dominant condition has a 50% chance of being affected by that condition via inheritance of a dominant allele.

- **N/N –Clinically normal, no possible transmission to offspring.**
- **N/MUT – Disease likely, possible transmission to offspring.**
- **MUT/MUT – Disease likely, transmission to offspring.**
    - **N – normal allele**
    - **MUT – mutant allele**

Autosomal Recessive Disorder

Autosomal recessive is a pattern of inheritance characteristic of some genetic disorders. "Autosomal" means that the gene in question is located on one of the numbered, or non-sex, chromosomes. "Recessive" means that two copies of the mutated gene (one from each parent) are required to cause the disorder.

- **N/N – Clinically normal, no possible transmission to offspring.**
- **N/MUT – Clinically normal, possible transmission to offspring.**
- **MUT/MUT – Disease likely, possible transmission to offspring.**
    - **N – normal allele**
    - **MUT – mutant allele**

Chromosome

The genome is organized into chromosomes that contain most of the DNA of a living organism. Horses have 31 pairs of autosomes (non-sex chromosomes) and one pair of sex chromosomes (X and Y). One copy of each chromosome comes from the sire, and one copy comes from the dam.

Dominant and recessive

Dominant refers to the relationship between two versions of a gene. Individuals receive two versions of each gene, known as alleles, from each parent. If the alleles of a gene are different, one allele will be expressed; it is the dominant gene. The effect of the other allele, called recessive, is masked.

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Gene

The gene is considered the basic unit of inheritance. Genes contain the information needed to specify physical and biological traits. Most genes code for specific proteins, or segments of proteins, which have differing functions within the body.

Genome

The genome is the entire set of DNA instructions found in a cell. A genome contains all the information needed for an individual to develop and function.

Genotype

The unique combination of alleles within an individual at a particular locus. The genotypes at a single locus, or more often, multiple loci, underlie traits or phenotypes. It can be represented by symbols. For example, BB, Bb, bb could be used to represent a given variant in a gene. Genotypes can also be represented by the actual DNA sequence at a specific location, such as CC, CT, TT. DNA sequencing and other methods can be used to determine the genotypes at millions of locations in a genome in a single experiment. Some genotypes contribute to an individual's observable traits, called the phenotype.

Homozygous and Heterozygous

Homozygous, as related to genetics, refers to having inherited the same versions (alleles) of a genomic marker from each biological parent. Thus, an individual who is **homozygous** for a genomic marker has two identical versions of that marker. By contrast, an individual who is **heterozygous** for a marker has two different versions of that marker.

Locus

A locus, as related to genomics, is a physical site or location within a genome (such as a gene or another DNA segment of interest), somewhat like a street address. The plural of locus is loci.

Mode of inheritance

- Autosomal recessive: Recessive mutations require two mutated copies for disease to develop.
- Autosomal dominant: Dominant mutations are expressed when only one copy of that mutation is present.
- Autosomal incomplete dominant: n incomplete dominance, the variants (alleles) are not expressed as dominant or recessive; rather, the dominant allele is expressed in a reduced ratio.
- X-linked recessive: X-linked recessive disorder often have affected males, but rarely affected females, in each generation.
- X-linked dominant: For X-linked dominant diseases, however, a mutation in one copy of an X-linked gene will result in disease for both males and females.

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Mutation

- A mutation is a change in the DNA sequence of an organism. Mutations can result from errors in DNA replication during cell division, exposure to mutagens or a viral infection.

Phenotype

Phenotype refers to an individual's observable traits, such as height, eye colour and blood type. A person's phenotype is determined by both their genomic makeup (genotype) and environmental factors.

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Technology & Methodology

A Clear Advantage: Introducing our cutting-edge Whole Genome Sequencing service!

As a devoted horse owner, you strive to provide the best care for your equine companion. Now, imagine having the power to delve deep into your horse's unique genetic code to understand its health, potential, and heritage like never before.

Present market-dominating methods of horse DNA analysis you may be aware of study specific informative markers one by one. This typically requires a three-step procedure:

- Sampling, e.g. blood or hair collection to isolate DNA
- Wet-lab processing of the sample having in mind what marker is in question
- Data analysis and reporting

But what if you start thinking of another DNA marker? Your horse may and its sample will for sure undergo another round of it (sampling and/or analysis), with a new marker under focus. Of course, you can combine analysis of several markers at a time but always you need to specify them first and always there will be questions like: *What other test should I consider? Why have I not included this marker too?*

Therefore, as you can see yourself, although these traditional and widely used approaches offer valuable insights into specific traits, they suffer from obvious drawbacks, the inability to provide a full picture being the most important one.

This is not the case of Whole Genome Sequencing services! Our technological advantages are, namely:

- **We cover the broad picture encoded in the entirety of your horse's genome and capture the full spectrum of genetic information**. In fact, the broadest you can think of. With Whole Genome Sequencing, you gain a comprehensive understanding of your horse's genetic makeup.
- **Whole Genome Sequencing is assumption-free.** Every specific genetic marker you have ever thought or heard of will be covered and you do not need to tell us upfront what you are looking for in your horse’s DNA. In fact, even markers you or scientists are not aware of yet will be covered too! It is like having the entire storybook, and not just a few words or chapters at best when using traditional methods!

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- **We require a single sampling of your horse only.** Once you send us your horse's sample and we isolate the DNA, we will sequence it and the data can be stored forever. If you later decide for another look into your horse genomic makeup or if there are new traits discovered or associations found by scientists in the future, there is no need of additional sampling. The book of your horse's DNA will be available at hand.
- **The more horse genomes are sequenced, the more data is available to compare them to each other.** By analyzing tens or hundreds of genomes, people will be able to uncover previously unknown genetic predispositions for specific diseases or key performance characteristics of sport horses. If you have the whole genome of your horse analyzed, we can draw conclusions that are basically related to the individual in question. But if you have the genomes of your entire stable analyzed, we can start comparing them to each other, getting a new and higher level of information. We can create a large catalogue of known and new traits to help the community understand the horse DNA landscape. And you can be a part of it!

For the Curious Minds: **How Whole Genome Sequencing Works?**

The horse's DNA or genome is made up of 2.7 billion bases, simplified to the letters A, G, C, T. The combination of these 4 letters in the DNA strand encodes approximately 20,000 genes that are responsible for the appearance, characteristics and health of the individual. Until recently, however, no technology was available to read the 2.7 billion letters of DNA quickly and cost-effectively. But the latest sequencing technologies now allow us to do just that.

The very first step, after isolating your horse's DNA, is to break that DNA into small fragments and create what is called a sequencing library. We then read the fragments in this library one by one using a device called a sequencer to identify the order of the bases (A, T, C, G) in the fragment. In a third step, we assemble these short fragments using computer programs to get a complete picture of the genome of the test horse. Finally, we compare the information obtained with a reference genome and, on the basis of this comparison, we can then diagnose genetic diseases, predispose to diseases or identify genetic variants associated with certain traits.

EquineTest: Horse Genetic Sequencing

Thinking of Whole Genome Sequencing, think of assembling a puzzle. The result is a comprehensive genetic map that reveals the secrets hidden within. With a single test, we can bring invaluable knowledge to horse owners, breeders, trainers and enthusiasts now and in the future.

Embark on a genetic journey with us, and empower yourself as a horse owner. Our Whole Genome Sequencing service is not just about data – it is about forming a stronger bond with your horse, understanding its uniqueness, and making informed decisions for its health and future.

EquineTest: Horse Genetic Sequencing

Disclaimers

This Is Not a Diagnosis

These results are intended for informational purposes only and do not constitute medical care. They indicate genetic predispositions, not certainties. An increased risk doesn't guarantee the development of a condition or trait. Conversely, it's possible for a condition to manifest even if you don't have the genetic predisposition for it.

Consult with a Veterinarian

Our test is not a substitute for a professional veterinary examination. Therefore, do not make significant decisions about your horse's health or management without consulting with your veterinarian

Evolution of Genetic Insights

Our equine genetic test outcomes rely on the most recent scholarly work in the field. But science is an ever-advancing frontier; thus, fresh research could pave the way for enhanced understandings and updated interpretations of these genetic data as time goes on

Next steps

Opt for our cutting-edge online storage to access your horse's genetic data and reports conveniently through our secure online platform: €10 per horse and month for online data storage + future discovery analyses.

www.equinetest.com

SEQme s.r.o.
Dlouha 176, 26301 Dobris
Czech Republic

hello@equinetest.com