

REVOLUTIONARY DNA INSIGHTS INTO YOUR HORSE HEALTH

Report type A Thoroughbred, Arabian and similar

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

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	

Report summary

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

- 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.

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

Gene	TRPMI
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	PLODI
Allele	LHI
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	Μυτγή, τοεί
Allele	Μυτγή, τοεί
Mode of inheritance	Autosomal recessive

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 Mode of inhoritance	RAP Autonomal reconsive
Mode of inheritance	Autosomal recessive
Result Simplified interpretation	Homozygote N/N
Simplified Interpretation	Clinically normal, no possible transmission to offspring.

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	К1
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

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
Result Simplified interpretation	Clinically normal. The variant/mutation will be passed on to the
Simplified interpretation	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%.
Simplified interpretation	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy
Simplified interpretation	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy Polysaccharide storage myopathy (PSSM1) disorder is caused by
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy Polysaccharide storage myopathy (PSSM1) disorder is caused by abnormal storage of sugar (glycogen) in the muscles. It is common
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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
Simplified interpretation Category Phene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy Polysaccharide storage myopathy (PSSMI) 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
Simplified interpretation Category Phene Description	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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.
Simplified interpretation Category Phene Description Breed(s)	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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. All
Simplified interpretation Category Phene Description Breed(s) Chromosome	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy Polysaccharide storage myopathy (PSSMI) 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 PSSMI. All
Simplified interpretation Category Phene Description Breed(s) Chromosome Gene	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy Polysaccharide storage myopathy (PSSMI) 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 PSSMI. All 10 GYSI
Simplified interpretation Category Phene Description Breed(s) Chromosome Gene Allele	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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. All 10 GYS1 P1
Simplified interpretation Category Phene Description Breed(s) Chromosome Gene Allele Mode of inheritance	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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. All 10 GYS1 P1 Autosomal incomplete dominant
Simplified interpretation Category Phene Description Breed(s) Chromosome Gene Allele	Clinically normal. The variant/mutation will be passed on to the offspring with a probability of 50%. Disorder Polysaccharide storage myopathy 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. All 10 GYS1 P1

PheneGlanzmann's ThrombastheniaDescriptionGlanzmann's thrombasthenia (GT, coagulopathy) is a blood disorder in horses. The main symptom is repeated noseblee frequent other bleeding. The mutation affects the functions platelets (thrombocytes) and thus impairs blood clotting.	U U
disorder in horses. The main symptom is repeated noseblee frequent other bleeding. The mutation affects the functions	U U
frequent other bleeding. The mutation affects the functions	eds and
platelets (thrombocytes) and thus impairs blood clotting.	of blood
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.	

Disorder

Muscle integrity myopathy_P2

Phene Description

Category

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)allChromosome14GeneMYOTAlleleP2Mode of inheritanceAutosomal dominant - variable manifestationResultHomozygote N/NSimplified interpretationClinically normal, no possible transmission to offspring.

Category Phene Disorder

Muscle integrity myopathy_P4

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.

Disorder

Category

Muscle integrity myopathy_Px

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

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			
Phene Description	Androgen insensitivity syndrome Androgen insensitivity syndrome (AIS) causes an individual who is			
	Androgen insensitivity syndrome Androgen insensitivity syndrome (AIS) causes an individual who is genetically male to appear female. The affected male is sterile, small			
	Androgen insensitivity syndrome Androgen insensitivity syndrome (AIS) causes an individual who is			
Description	Androgen insensitivity syndrome 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.			
Description Breed(s)	Androgen insensitivity syndrome 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. Thoroughbred (Horse)			
Description Breed(s) Chromosome	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X			
Description Breed(s) Chromosome Gene	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X AR			
Description Breed(s) Chromosome Gene Allele	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X AR AIS2			
Description Breed(s) Chromosome Gene Allele Mode of inheritance	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X AR AIS2 X-linked recessive			
Description Breed(s) Chromosome Gene Allele Mode of inheritance Result	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X AR AIS2 X-linked recessive Homozygote N/N			
Description Breed(s) Chromosome Gene Allele Mode of inheritance	Androgen insensitivity syndrome 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. Thoroughbred (Horse) X AR AIS2 X-linked recessive			

Category Phene Description

Disorder

Androgen insensitivity syndrome

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	AIS3				
	Mode of inheritance	X-linked recessive				
	Mode of Infinentance					
	Result	Homozygote N/N				
		-				
	Result	Homozygote N/N				
	Result Simplified interpretation	Homozygote N/N Female: Clinically normal, no possible transmission to offspring.				
	Result	Homozygote N/N				

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.

Part 2 - All markers

Category	Phene	Chr	Gene	Allele	Result
Disorder	Dwarfism, ACAN-related	1	ACAN	Dl	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	ΜΥΟ5Α	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

+ 100 more markers in the full report

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.

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.

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.

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 with 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!

- 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 catalog 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 four 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, predisposition to diseases or identify genetic variants associated with certain traits.

Thinking of Whole Genome Sequencing, think of assembling a puzzle. The result is a comprehensive genetic map that reveals the secrets hidden within. It is not just science; it is a journey of discovery for every horse enthusiast.

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.

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.

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