ETALON DIAGNOSTICS

REPORT DATE: APRIL 5, 2023

HORSE: ECHO ETERNAL

OWNER: AMY ASHBY

Genetic profile test results

HORSE ID: 042021_019

PACKAGES: APHA, SINGLE TEST - IMMUNE-MEDIATED MYOSITIS (IMM)

Horse and owner information

Horse Echo Eternal	Date of birth 06-06-2014
Breed Paint (Tovero)	Age 8 y.o.
Color Black	Sex Stallion
Discipline Breeding	Height 16 Hands
Registry American Paint Horse Association	Reg number 1050037
Sire BBF DR DIXON	Dam PRISSY PUDDIN
Sire Reg & No. American Paint Horse Association 386443	Dam Reg & No. American Paint Horse Association 521784
Owner Amy Ashby	Address 2118 PINE ROAD
Phone 7135031180	City, State CLEVELAND, TX
Email ashbyakgm@gmail.com	Postal code 77328

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Results Summary

Variant summaries:

Genetic note: **Immune-mediated Myositis (MY): No Immune-mediated Myositis variants detected.**

Color: **a/a, E/E, nd1/nd2, LWO/n, TO/n**

Speed: **Sprint Type**

Temperament: **Curious**

Gait: **Neg for DMRT3**

Performance and Abilities:

Curious
Two Curiosity variants; horse may be more curious than vigilant.

Non-"Gaited" DMRT3
No DMRT3 variants; likely non-gaited (*variants for novel "gait" abilities are currently in research).

Sprint
Two Sprint type variants; horse may accel at short distance, quick bursts of speed over endurance type activity.

Health Variants:

Lethal White Overo (LWO) - LWO/n
LWO/n - One Frame/Lethal White Overo (LWO) variant detected, resulting in "Carrier" status and may exhibit White markings. Caution is recommended when breeding to avoid another carrier and thus a 25% chance of foal fatality.

Coat color:

Black Homozygous (base)
Black (E) is the base coat color for this horse and is a relatively uncommon coat color on its own (usually it is found in combination with other colors or modifiers such as in Bay horses). A visible difference between a true black (Ee or EE), a dark chestnut (ee) or a bay (Aa or AA + E) can sometimes be seen in the fine hairs around the eyes and muzzle. For a horse to be "homozygous black", it must have TWO copies of the Black variant (EE) and horse has 100% chance of passing Black to any offspring.

Tobiano (TO) - TO/n
TO/n - One Tobiano (TO) variant detected; may result in large white markings on body, face and legs (occasionally produces 'minimal' or 'slipped' Tobiano horse with very little white only on face and lower legs (ongoing heritability study for minimal Tobiano). Horse has 50% chance of passing to any offspring.

Overo (LWO) - LWO/n
LWO/n - One Frame/Lethal White Overo (LWO) variant detected; may result in White markings and possibly blue eye(s). Horse has 50% chance of passing to any offspring.

non-Dun Primitive Markings (nd) - nd1/nd2
nd1/nd2 - Non-Dun (nd) Primitive Markings may result in dorsal striping, leg barring and "shadows" on the face and shoulder. Still in research, it is suspected in some cases that nd1 may also cause mild coat color dilution, though not as extensive as the Dun (D) variant. Horse with one copy of nd1 variant will pass nd1 to 50% of any offspring.

Blue Eyes Possible
Horses with the Tobiano variant (TO/n or TO/TO) in addition to the Lethal White Overo variant (LWO/n) have an increased likelihood of having one or two blues eyes.

Tovero (TO/TO, LWO/n or TO/n, LWO/n)
Horses with at least one Tobiano variant (TO/n or TO/TO) and one Lethal White Overo variant (LWO/n) are often referred to as "Tovero"

Coat color

Base

Agouti (A) - a/a

ASIP

Negative

Gene or region: **ASIP**

a/a - No Dominant Agouti variants detected. Agouti (A) restricts black pigment to the outer regions of the body, the legs, mane & tail, nose, ear tips causing the otherwise black horse to appear Bay. Agouti is invisible on the red based coat.

Black (E) - E/E

MC1R

Black Based

Gene or region: **MC1R**

E/E - Two Black variants detected and no Red.

Modifiers

Grey (G) - n/n

STX17A

Negative

Gene or region: **STX17A**

No Grey (G) variants detected.

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non-Dun Primitive Markings (nd) - nd1/nd2

TBX3

Possibly Affected

Gene or region: **TBX3**

nd1/nd2 - One non-Dun Primitive Markings variant detected. Non-Dun Primitive Markings can appear as a dorsal stripe, leg barring, shadows on the face and shoulders even in the absence of the Dun variant.

Dilutes

Sunshine (SUN) - n/n

SLC45ASUN

Negative

Gene or region: **SLC45ASUN**

No Sunshine variants detected.

Champagne (CH) - n/n

SLC36A1

Negative

Gene or region: **SLC36A1**

No Champagne (CH) variants detected.

Silver (Z) - n/n

PMEL17

Negative

Gene or region: **PMEL17**

No Silver (Z) variants detected. Silver affects only black base (E) coat colors and is invisible on a red-based coat. On black or bay, It has a disproportionate diluting effect on the mane and tail, and is sometimes called, "Chocolate Flaxen".

Cream (CR) - n/n

SLC45A2

Negative

Gene or region: **SLC45A2**

No Cream (CR) variants detected.

Pearl (PRL) - n/n

SLC45A2

Negative

Gene or region: **SLC45A2**

No Pearl (prl) variants detected.

Dun (D) - n/n

TBX3

Negative

Gene or region: **TBX3**

No Dun (D) variants detected. Dun is a modifier that dilutes the base coat color often revealing Primitive Markings such as a dorsal stripe, leg barring, shadows on the face and shoulders.

Whites

Frame/Lethal White Overo (LWO) - LWO/n

EDNRB

Likely Affected

Gene or region: **EDNRB**

LWO/n - One Frame/Lethal White Overo variant detected. May result result in White markings and possibly blue eye(s).

Tobiano (TO) - TO/n

ECA3

Likely Affected

Gene or region: **ECA3**

TO/n - One Tobiano variant detected; likely white markings. This horse may pass Tobiano to 50% of any offspring.

Leopard Complex Spotting (LP) - n/n

TRPM1

Negative

Gene or region: **TRPM1**

No Leopard Complex Spotting variants detected.

Pattern 1 (PATN1) - n/n

RFWD3

Negative

Gene or region: **RFWD3**

No Pattern (PATN1/n) 1 variants detected.

Sabino1 (SB1) - n/n

KIT

Negative

Gene or region: **KIT**

No Sabino (SB1) variants detected.

Splashed White (SW1) - n/n

MITF

Negative

Gene or region: **MITF**

No Splashed White 1 (SW1) variants detected.

Splashed White (SW2) - n/n

PAX3

Negative

Gene or region: **PAX3**

No Splashed White 2 (SW2/n) variants detected.

Splashed White (SW3) - n/n

MITF

Negative

Gene or region: **MITF**

No Splashed White 3 (SW3) variants detected.

Splashed White (SW4) - n/n

PAX3

Negative

Gene or region: **PAX3**

No Splashed White 4 (SW4) variants detected.

Dominant White (W) - n/n

KIT

Negative

Gene or region: **KIT**

No Dominant White (1-21) variants detected.

Health Variants

Color Related Risk

Lethal White Overo (LWO) - LWO/n	EDNRB	Carrier

Gene or region: **EDNRB**

LWO/n - One Frame/Lethal White Overo (LWO) variant detected, resulting in "Carrier" status and may exhibit White markings. Caution is recommended when breeding to avoid another carrier and thus a 25% chance of foal fatality.

Congenital Stationary Night Blindness (CSNB) - n/n	TRPM1	Negative

Gene or region: **TRPM1**

n/n - No Leopard Complex Spotting (LP) variants detected, which is related to the presence of Congenital Stationary Night Blindness (CSNB) if horse is LP/LP. Horses with one copy of the Leopard Complex Spotting (LP) variant are not currently known to suffer any ill effects as a result. Horses with Congenital Stationary Night Blindness (CSNB) which may experience the inability to see in low to no-light conditions.

Multiple Congenital Ocular Anomalies (MCOA) - n/n	PMEL17	Negative

Gene or region: **PMEL17**

n/n - No Silver variants detected which is related to the presence of Multiple Congenital Ocular Anomalies (MCOA).

Immune System

Foal Immunodeficiency Syndrome (FIS) - n/n	SLC5A3	Negative

Gene or region: **SLC5A3**

No Foal Immunodeficiency Syndrome (FIS) variants detected.

Severe Combined Immunodeficiency (SCID) - n/n	DNAPK	Negative

Gene or region: **DNAPK**

No Severe Combined Immunodeficiency (SCID) variants detected.

West Nile Virus Symptom Susceptibility Risk (WNVR) - n/n	OAS1	Negative

Gene or region: **OAS1**

No West Nile Virus Symptom Susceptibility Risk (WNVR) variants detected. Normal susceptibility to West Nile Virus symptoms. (*NOT a test for West Nile Virus).

Immune-mediated Myositis (MY) - n/n	MYH1	Negative

Gene or region: **MYH1**

No Immune-mediated Myositis variants detected. Normal susceptibility for IMM or nonexertional rhabdomyolysis.

Equine Herpes Myeloencephalopathy Risk (EHMR) - after contracting Equine Herpes Virus type 1 (EHV1)	TSPAN9	Not ordered

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Muscle Disorders

Glycogen Branching Enzyme Deficiency (GBED) - n/n	GBE1	Negative

Gene or region: **GBE1**

No Glycogen Branching Enzyme Deficiency (GBED) variants detected.

Hyperkalemic Partial Paralysis (HYPP) - n/n	SCN4A	Negative

Gene or region: **SCN4A**

No Hyperkalemic Partial Paralysis (HYPP) variants detected.

Malignant Hyperthermia (MH) - n/n	RYR1	Negative

Gene or region: **RYR1**

No Malignant Hyperthermia (MH) variants detected.

Myotonia (MYT) - n/n	CLCN4	Negative

Gene or region: **CLCN4**

No Myotonia (MYT) variants detected.

Polysaccharide Storage Myopathy type 1 (PSSM1) - n/n	GYS1	Negative

Gene or region: **GYS1**

No Polysaccharide Storage Myopathy type 1 (PSSM1) variants detected.

Neurologic Disorders

Cerebellar Abiotrophy (CA) - n/n	MUTYH	Negative

Gene or region: **MUTYH**

No Cerebellar Abiotrophy (CA) variants detected.

Lavender Foal Syndrome (LFS) - n/n	MYO5A	Negative

Gene or region: **MYO5A**

No Lavender Foal Syndrome (LFS) variants detected.

Hydrocephalus (HDC)	B3GALNT2	Not ordered
Recurrent Laryngeal Neuropathy (RLN)	ECA3	Not ordered

Reproductive Disorders

Androgen Insensitivity Syndrome (AIS) - n/n	AR	Negative

Gene or region: **AR**

No pattern of Androgen Insensitivity Syndrome (AIS) variants detected.

Impaired Acrosomal Reaction - Subfertility Risk (IAR) - iar/iar, iar/n	FKBP6IAR1...		Not Affected

Gene or region: **FKBP6IAR1, FKBP6IAR2**

No pattern for Impaired Acrosomal Reaction (IAR) - Subfertility Risk variants detected.

Skin, Hoof and Connective Tissue Disorders

Hereditary Equine Regional Dermal Asthenia (HERDA) - n/n	PPIB		Negative

Gene or region: **PPIB**

No Hereditary Equine Regional Dermal Asthenia (HERDA) variants detected.

Junctional Epidermolysa Bullosis type 1 (JEB1) - n/n	LAMC2		Negative

Gene or region: **LAMC2**

No Junctional Epidermolysa Bullosis type 1 (JEB1) variants detected.

Junctional Epidermolysa Bullosis type 2 (JEB2) - n/n	LAMA3		Negative

Gene or region: **LAMA3**

No Junctional Epidermolysa Bullosis type 2 (JEB2) variants detected.

"Warmblood" Fragile Foal Syndrome (FFS)	PLOD1	Not ordered
Hoof Wall Separation Disease (HWSD)	SERPINB11	Not ordered
Naked Foal Syndrome (NFS)	st14nfs	Not ordered

Chronic Idiopathic Anhidrosis Risk (CIAR)	KCNE4	Not ordered

Occular Disorders

Equine Recurrent Uveitis Susceptibility Risk (ERUR)	BIEC2536712WB	Not ordered
Equine Recurrent Uveitis Symptom Severity (ERUS)	BIEC2421990WB	Not ordered
Squamous Cell Carcinoma Susceptibility Risk (SCC)	DDB2	Not ordered

Skeletal Disorders

Dwarfism (D)	ACAND1...	Not ordered
Friesian Dwarfism (FD)	B4GALT7Dfriesian	Not ordered
Kissing Spines Susceptibility (KSS)	ECA25	Not ordered
Lordosis	ECA20...	Not detected

Gene or region: **ECA20, ECA20, ECA20, ECA20**

Horses with one copy in each of the four Lordosis regions are not currently known to suffer any ill effects as a result. Horses with two copies in each of the four Lordosis regions exhibit signs of swayback. Currently studies are only proven in the N. Am. Saddlebred breed.

Endocrine Disorders

Equine Metabolic Syndrome Susceptibility Risk (EMS)	BIEC2263524EMS	Not ordered
Laminitis Susceptibility Risk - Equine Metabolic Syndrome related (LAM)	BIEC2263524_LAM	Not ordered

Blood and Vascular Disorders

Glanzmann Thrombasthenia (GT)	ITGA2BG...	Not ordered

Health

Glanzmann Thrombasthenia (GT)	ITGA2BG...	Not ordered

Height

Height (H1)	LCORL	Not ordered
Height (H2)	HMGA2	Not ordered

Performance and Abilities

Gait Type

Non-"Gaited" DMRT3 — DMRT3 — Detected

Gene or region: **DMRT3**

No DMRT3 variants; likely non-gaited (*variants for novel "gait" abilities are currently in research).

Performance

Sprint — MSTN — Likely Affected

Gene or region: **MSTN**

Two Sprint type variants; horse may accel at short distance, quick bursts of speed over endurance type activity.

Temperament

Curious — DRD4 — Detected

Gene or region: **DRD4**

Two Curiosity variants; horse may be more curious than vigilant.