ETALON DIAGNOSTICS

Genetic profile test results

REPORT DATE: APRIL 25, 2023

HORSE: BCH MAINE'S CASSIOPEIA WISH SE 19-18-0379

OWNER: [redacted] ICHO Charlotte Wittrup Bengaard

HORSE ID: 041823_004

PACKAGES: ICHO MINIPANEL

Horse and owner information

Horse
BCH Maine's Cassiopeia Wish SE 19-18-0379

Date of birth
07-30-2018

Breed
Curly Horse

Age
4 y.o.

Color
Bay Tobiano

Sex
Mare

Discipline
-

Height
-

Registry

Reg number

Sire
Maine Dream SE 19-15-0368

Dam
Missouri's Juana SE 19-05-0170

Sire Reg & No.

Dam Reg & No.

Owner
[redacted] ICHO Charlotte Wittrup Bengaard

Address
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Phone
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Tularosa, NM

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88352

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HORSE: BCH MAINE'S CASSIOPEIA WISH SE 19-18-0379

HORSE ID: 041823_004

Results Summary

Variant summaries:

Genetic note: **Curly Coat (CU): Two Curly Coat 2 (SP6) variants detected.**

Color: **A/a, E/e, nd1/nd2, TO/n**

Modifier: **CU2/CU2** SP6 +/+ homozygous

Speed: **Endurance Type**

Temperament: **Curious/Vigilant** DRD4 +/-

Gait: **Neg for DMRT3**

Performance and Abilities:

Curious & Vigilant
One Curiosity and one Vigilance variant; horse is likely both curious and vigilant (*ongoing study).

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

Endurance
Endurance type myostatin; horse may accel at longer distance travel versus short distance sprint type activity.

Health Variants:

Horse has not tested positive for any known disease variants on this panel.

Coat color:

Bay
Black (E) + Agouti (A): Bay (A, E) is a very common coat color in horses. Bay requires at least one black variant (E) and at least one Agouti/Bay variant (A). The Agouti restricts the black pigment to the outer points including the mane, tail, lower legs, and sometimes the tips of the ears. Bay horses have black skin under their coat, except beneath white markings. Horse with two Black (EE) and two Agouti (AA) have a 100% chance of passing Black and Agouti to any offspring. Horse with one Black (Ee) and one Agouti (Aa) have a 50% chance of passing Black or Red and a 50% chance of passing Agouti 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.

Curly Coat (CU) - n/n, CU2/CU2
n/n, CU2/CU2 - No Curly Coat 1 (KRT25) variants and two Curly Coat (SP6) variants detected. Horse may exhibit curly hair in varying degrees. Horse has a 100% chance of passing CU2 to any offspring.

Coat color

Base

Agouti (A) - A/a	ASIP	Likely Affected

Gene or region: **ASIP**

A/a - One Dominant Agouti variant detected. Agouti (which causes "Bay" on black) 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 - One Black variant and one Red variant detected.

Modifiers

Tiger Eye (TE1) - Not Ordered	SLC24A5TE1	Not ordered

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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.

Tiger Eye (TE2) - Not Ordered	SLC24A5TE2MUT	Not ordered

Grey (G) - n/n	STX17A	Negative

Gene or region: **STX17A**

No Grey (G) variants detected.

Brindle (BR1) - n/n	MBTPS2BR1		Negative

Gene or region: **MBTPS2BR1**

n/n - No Brindle (BR1) variants detected. Horse with Brindle (BR1) may display overall haircoat showing streaks of darker and lighter hair, similar to the brindle coat color in other species.

Dilutes

Sunshine (SUN) - not tested	SLC45ASUN		Not ordered

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

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.

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

Gene or region: **EDNRB**

No Frame/Lethal White Overo (LWO) variants detected.

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.

Coat Type

Curly Coat (CU2) - CU2/CU2

SP6

Likely Affected

Gene or region: **SP6**

CU/CU - Two Curly Coat (CU) variants detected. Horse may have curly coat, mane and tail and pass this trait to offspring 100% of the time.

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HORSE ID: 041823_004

Curly Coat (CU1) - n/n	KRT25	Negative

Gene or region: **KRT25**

No known Curly Coat (CU-KRT25) variants detected.

Health Variants

Color Related Risk

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

Gene or region: **EDNRB**

n/n - No Lethal White Overo (LWO) variants detected.

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 **I**mmunodeficiency Syndrome (**FIS**) variants detected.

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

Gene or region: **DNAPK**

No Severe Combined **I**mmunodeficiency (**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 (IMM)	MYH1		Not ordered
Equine Herpes Myeloencephalopathy Risk (EHMR) - after contracting Equine Herpes Virus type 1 (EHV1)	TSPAN9		Not ordered

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) - n/n, 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

Endurance	MSTN		Likely Affected

Gene or region: **MSTN**

Endurance type myostatin; horse may accel at longer distance travel versus short distance sprint type activity.

Temperament

Curious & Vigilant	DRD4		Detected

Gene or region: **DRD4**

One Curiosity and one Vigilance variant; horse is likely both curious and vigilant (*ongoing study).
