PAINT HORSE ASSOCIATION OF AUSTRALIA

TESTING / GENETIC SCREENING REQUEST FORM

Horses Registration Name: _____

Registration Number: _____ **DOB** ___/___/_____ **Sex** _____

Sire Name _____ **Reg #** _____

Dam Name _____ **Reg #** _____

Members Name _____ **Memb No** _____

Address _____

City/Town _____ **State** _____ **Postcode** _____

Email _____

☐ **Please send the kit via email** ☐ **Please Send kit via Post**

Please indicate the requested tests (DNA parent Validation is compulsory for all horses)

☐ **DNA Parent Validation $100**

☐ **DNA Parent Validation + 1 Gen Test $115**

☐ **DNA Parent Validation + 2 Gen Test $120**

☐ **DNA Parent Validation + 3 Gen test $ 125**

☐ **DNA Parent Validation + 5 Gen Test $160**

☐ **5 Panel Test (please indicate the test required) $105**

☐ **3 Panel Test (please indicate the test required) $85**

☐ **2 Panel Test (please indicate the test required) $80**

☐ **1 Panel test (please indicate the test required) $60**

Genetic Conditions Tests

☐ **HYPP** ☐ **HERDA** ☐ **MH** ☐ **GBED** ☐ **PSSM1** ☐ **MYHM** ☐ **OWLS (Overo)**

Coat Colour Tests

☐ **CCC Red Factor (red/black)** ☐ **AG – Agouti (Bay)** ☐ **CD – Cream Dilution**

☐ **PRL – Pearl** ☐ **CHAMPAGNE** ☐ **SILVER** ☐ **DUN**

Coat Pattern Test

☐ **TOBIANO** ☐ **SW1** ☐ **SW2** ☐ **SW3** ☐ **W5** ☐ **W10** ☐ **W20**

☐ **SABINO 1**

PAYMENT DETAILS

VISA/MASTERCARD – Will attract a 2.00 % Merchant Fee

Card Number _ _ _ _/_ _ _ _/_ _ _ _/_ _ _ _ Expiry_ _/_ _ CCV _ _ _

Card Holder Name _____ Signature _____

Direct deposit details: BSB: 062 534 Acc: 1025 3938 Date of deposit _ _/_ _/ _____ Ref # _____

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PAINT HORSE ASSOCIATION OF AUSTRALIA

Genetic & Colour Screening Tests

Symbol	Name	Nature of Characteristic or Disease
Genetic Disease Testing		
OLWFS/OVERO	Overo Lethal White Foal Syndrome	Overo Lethal White Foal Syndrome occurs when a horse is homozygous (OWLFS/OWLFS) for the OLWFS mutation. These foals are born almost or completely white, but do not have properly formed intestinal nerves and cannot pass faeces. They only survive a few days if not euthanised for compassionate reasons. These foals are born to parents who both carry (OWLFS/n) OLWFS. Carrier horses (OLWFS/n) have no documented health issues. Because OLWFS can be minimally expressed, it is important to test any horse that might be a carrier even if it has little to no white on it, to prevent the birth of an affected foal. A single O allele causes the "frame" or "frame overo" spotting pattern. Expression of white in frame horses is variable, ranging from lots of white to minimal or just a few white hairs. It is possible, although unusual, for a horse with no white to carry Frame.
GBED	Glycogen Branching Enzyme Deficiency	A metabolic genetic disease that is fatal in foetal and neonatal stages, with most affected foals dying before 8 weeks of age. It is inherited as a recessive trait, so only homozygous (GBED/GBED) horses are affected. If a horse is a carrier (n/GBED), it will not show any clinical signs of GBED but has a 50% chance of passing GBED to its offspring. GBED is caused by a mutation in the GBE1 gene that prevents the proper storage of glucose.
HERDA	Hereditary Equine Regional Dermal Asthenia	Characterised by severe skin lesions, often on the horses back. The disorder affects the collagen that holds the skin in place, making it much easier to tear off than normal. The lesions are painful and prone to infection. HERDA is caused by a mutation in the PPIB gene and is recessive, so the horse must be homozygous (HERDA/HERDA) to be affected. If a horse is a carrier (n/HERDA), it will not show any clinical signs of HERDA. However, there is a 50% chance it will pass the variant to its offspring, so mating to other carriers should be avoided to prevent the birth of an affected foal.
HYPP	Hyperkalemic Periodic Paralysis	Causes delayed muscle relaxation. HYPP is a dominant trait, meaning a horse only needs 1 copy of the mutation (HYPP/n) to be affected. There is some evidence that homozygous horses (HYPP/HYPP) are more severely affected than heterozygotes. The severity and onset of symptoms can be managed with diet, particularly by avoiding high potassium feeds. HYPP is caused by a mutation in the SCN4A gene.
MH	Malignant Hyperthermia	A muscle disorder that may only become apparent if the horse is subjected to extreme stress or exposed to a halogenated anaesthetic. The mutation triggers the release of excess calcium in skeletal muscle cells causing a high temperature (hence the name), increased heart rate and blood pressure, sweating and muscle rigidity. MH is frequently fatal but is rare and only found in some

		Quarter Horse and paint families; however, because it is potentially fatal it is recommended all possible carriers be tested before undergoing anaesthesia. MH is associated with a mutation in the RyR1 gene and is a dominant trait, meaning a horse only needs 1 copy of the mutation (MH/n) to be affected.
PSSM1	Polysaccharide Storage Myopathy	Disorder of skeletal muscle, causing a build-up of abnormal sugars in muscles, leading to tying up. Symptoms can vary widely in severity and age of onset. It is a complex disorder that can often be controlled with changes to diet and exercise management. PSSM1 is associated with a mutation in the GYS1 gene and is inherited in a dominant fashion, so a horse only needs to carry one copy (PSSM1/n) to show symptoms. There is some evidence that homozygous horses (PSSM1/PSSM1) are more severely affected than heterozygotes.
MYHM (IMM)	Myosin-Heavy Chain Myopathy	MYHM can cause two different types of disease. One is muscle weakness and stiffness, followed by a rapid, significant loss of muscle, particularly from the topline of the horse. This is often associated with an infection or vaccination, particularly with Strangles or other respiratory viruses. This syndrome was originally named "immune mediated myositis" or IMM. It was later found that horses carrying this mutation were also susceptible to non-exertional rhabdomyolysis (The second type). This manifests as stiffness, firm muscles, short stride and often is accompanied by dark coloured urine (indicator of muscle damage). Unlike many other forms of tying up, it is not associated with exercise. The identification of this second set of clinical signs associated with the mutation caused IMM to be renamed Myosin-heavy chain myopathy. MYHM is co-dominant, meaning that the action of the variant is independent of the second variant. If a horse has one copy of the MYHM mutation it can be affected with MYHM. MYHM has 'incomplete penetrance' – so not every horse carrying this mutation will show the same severity of symptoms. A horse that has two copies (MYHM/MYHM) will pass MYHM to all offspring and is likely to be more severely affected than a horse with one copy (MYHM/n)
Equine Colour Pattern Testing		
A/a	Agouti	Controls distribution of black pigment in a horse's coat. The gene determines whether the non-chestnut horse is black or bay. The dominant allele (variant) 'A' limits black pigment to the points, and the horse will be bay. If the horse is homozygous for the recessive allele (a/a), the horse will be black. If the horse is heterozygous for the recessive allele (A/a), the horse will be bay. If the horse has no copies of the recessive allele (A/A) the horse will be bay
E/e	Chestnut/Red Factor	Inheritance of the base colours of chestnut, bay and black is dependent on two genes. The first is MC1R which will determine whether black pigment is expressed. The dominant allele (variant) is indicated with a capital E and allows black pigment on the coat. The recessive allele (e/e) prevents black pigment, so the horse is chestnut. When mated, two chestnuts can only ever produce

		chestnut foals. If the horse is heterozygous for the recessive allele (E/e), the horse is bay or black. When the horse has no copies (E/E), the horse is bay or black.
Cr	Cream Dilution	Cream dilution is responsible for palomino, buckskin, smoky black, cremello, perlino and smoky cream coat colours. Cream dilution is caused by a mutation in the MATP gene. If one copy of cream is detected (Cr/n), then only red pigment is diluted and the horse is palomino (Chestnut base), buckskin (Bay base) or smoky black (black base). If two copies are detected (Cr/Cr), the horse is diluted to cremello (chestnut base), perlino (bay base) or smoky cream (black base). Double dilute horses also have blue eyes and pink skin. It is usually not possible to accurately determine a double dilutes base colour without genetic testing.
Ch	Champagne Dilution	Champagne dilutes chestnut to a gold colour, like palomino. Classic champagne on black will dilute the horse to a dark tan colour, while champagne on bay will dilute the body to Amber with brown points. The eyes are blue green at birth and darken to amber as the horse grows. A horse heterozygous (Ch/n) will have a diluted coat and amber eyes. A horse homozygous (Ch/Ch) will look the same as a horse with one copy but will pass on the champagne allele to offspring.
D/nd1/nd2	Dun	Dun lightens the body colour, leaving the head, lower legs, mane and tail undiluted. It also can cause a darker dorsal stripe, shoulder stripes, and sometimes leg barring and concentric marks on the forehead (known as 'primitive markings'). There are 3 important variations of the Dun gene. D causes dun dilution and primitive markings, nd1 causes no dilution but there may be some primitive markings present (particularly a dorsal stripe and leg barring), and nd2 causes no dilution and no primitive markings. A horse carrying two copies of dun (D/D) will have a diluted coat and show primitive markings, as well as pass the D allele to all offspring. A horse carrying one copy of dun dilution and non-dun1 (D/nd1) or non-dun2 (D/nd2) will have a diluted coat and show primitive markings. Horses carrying two copies of non-dun1 (nd1/nd1), or a copy of both non-dun (nd1/nd2) might show primitive markings. Nd2/nd2 do not carry either of the dun dilution alleles.
OLWFS/OVERO	Overo Lethal White Foal Syndrome	See page 1.
To	Tobiano	Tobiano is a distinct white spotting pattern involving large defined markings. The head is often not affected by To, although it can still carry white markings caused by other genetic markers. Horses carrying one copy (To/n) likely have clean edged white patches on its body, extending over the topline. Horses homozygous (To/To) likely have clean edged white patches over the body and will pass To onto all offspring.

Prl	Pearl	A rare dilution usually only observed in Quarter horses and Spanish horse breeds. This mutation is recessive so the horse must have two copies (Prl/Prl) to express the effect and will have a lightened coat, mane, and tail, in addition to bright eye colours and will pass the gene onto all offspring. Those heterozygous (Prl/n) will not have a dilute coat but have a 50% chance of passing pearl to offspring. Pearl also interacts with Cream dilution to produce pseudo-double cream dilute phenotypes. This is because the Pearl mutation is found in the same gene as Cream dilution.
Sab1	Sabino-1	Sabino is a term used to describe several spotting patterns. Sabino-1 is a specific mutation that causes white on the legs (often high) and belly, and a blaze. Extensive roaning is often evident, particularly at the edges of the white patterns. Homozygous horses (Sab1/Sab1) are almost completely, if not completely, white. Horses heterozygous (Sab1/n) likely have large white stockings, a blaze and roaning or white on the belly.
Z	Silver	Silver dilution only affects black pigment. A black horse will show a flaxen or silver-grey mane and tail and a chocolate-coloured coat, often with dapples, accounting for the 'Silver Dapple' name. A bay horse will have a solid red coat with lighter legs and a flaxen/silver mane and tail. Silver is also associated with a range of ocular defects known as Multiple Congenital Ocular Abnormalities (MCOA) syndrome. If a horse has one copy of silver (Z/n) they will have fewer and less severe eye abnormalities than a homozygous silver (Z/Z) horse. A horse heterozygous for silver (Z/n) will pass silver onto 50% of offspring. A horse homozygous (Z/Z) will pass silver onto all offspring and will likely have more severe MCOA.
SW1, SW2, SW3	Splashed White	Splashed White is characterised by a large blaze, sometimes extending into a hood covering nearly the entire head, extensive white on the legs, and variable white on the body. The horse can look like it was dipped in white paint feet first. The eyes can be wholly or partially blue and the horse may be deaf. Deafness cannot be diagnosed just by looking at the distribution of white on the horse. The mutations that cause all the known splashed white patterns have been identified in one of two genes. SW1, SW3 mutations are found on the MITF gene, while the mutations for SW2 occur on the PAX3 gene. Two copies of SW1 are often associated with a larger amount of white on the body than just one copy. Horses heterozygous (SW1/n, SW2/n, SW3/n) likely have a blaze and white stockings. Horses homozygous (SW1/SW1 & SW2/SW2) likely have more extensive white than heterozygous horses. These horses will transmit splashed white to all offspring.