INTERPRETING GENETIC DISORDER TEST RESULTS

INFORMED BREEDING DECISIONS - Consider Genetic Disorders

WHAT ARE THE DISORDERS

Society tests may include the following disorders

❖ OLWS	Overo lethal white foal syndrome	
❖ HERDA	Hereditary equine regional dermal asthenia	
❖ PSSM1	Polysaccharide storage myopathy	
❖ HYPP	Hyperkalaemic periodic paralysis	
❖ GBED	Glycogen branching enzyme deficiency	
❖ MYHM	Myosin-Heavy Chain Myopathy	(from 1st July 2025)
❖ MH	Malignant Hyperthermia	(prior to 1st July 2025)

Horses that have been tested with the Society since 2016 will have their results published on the Online Stud Book.

For WELFARE REASONS, the Society strongly recommends carriers of Genetic Disorders are NOT mated to another carrier or affected horse. Owners of POSITIVE horses (carrier or affected) must disclose the disorder at all sales and breeding opportunities.

HOW TO READ GENETIC RESULTS

Search the horse on the Online Stud Book and review the horse's details, check the field titled - Genetic Disorder Results. This field lists the Genetic Disorders Tested and their results. We provide the following examples for members:

Two examples of NEGATIVE RESULTS (clear)

OLWS, HERDA, PSSM1, HYPP, GBED, MH - Clear n/n
Results show the horse is clear of the six disorders tested.

HERDA - Clear n/n.
Results show the horse is clear of Herda, no other disorders tested.

Four examples of POSITIVE RESULTS (carrier or affected)

GBED – Carrier n/Gb. OLWS, HERDA, PSSM1, HYPP, MH - Clear n/n.
Results show the horse is a carrier of GBED, but clear for the other disorders.

HERDA - Carrier n/Hr, GBED - Carrier n/Gb. OLWS, HERDA, PSSM1, HYPP, MH - Clear n/n.
Results show the horse is a carrier of HERDA and GBED, but clear for the other disorders.

HERDA - Carrier n/Hr.
Results show the horse has a single copy of Herda - CARRIER, no other disorders tested.

HERDA – Affected Hr/Hr.
Results show the horse has a double copy of Herda - AFFECTED, no other disorders tested.

WHAT HORSES ARE REQUIRED TO BE TESTED

Under Society Rules & Regulations, Genetic Disorder Testing is compulsory for new applications as follows:

- ❖ Base Registry - stallions, colts (6 panel)
- ❖ Base Registry - mares and fillies (Herda only)
- ❖ First Cross - mares and fillies (Herda only)
- ❖ Progeny of Genetic Disorder carriers (except geldings), and
- ❖ Any ASH stallion being Sire Registered (6 panel)

Sire Registered ASH Stallions have been required to be 6 panel Genetic Disorder Tested since 1st August 2016. The sire registration status for any previously Sire Registered ASH stallion not 6 panel Genetic Disorder Tested by 1st August 2019 has been suspended until such times as the Genetic Disorder Testing is complete.

BEFORE BREEDING

Consider Genetic Disorders

If breeding to a stallion or from a mare that has not been Genetic Disorder Tested, you may wish to consider testing the horses prior to breeding.

If the horse being used in your breeding programme is not an Australian Stock Horse, we recommend you check with other breed organisations to determine their Genetic Disorder status.

If your horse has been 6 panel Genetic Disorder Tested (CLEAR), does it mean your horse has no Genetic Disorders?

NO – There are other disorders which the Society does not require testing. The horse may be positive to alternate disorders. Other disorders could be determined in the future, or testing capability has not yet been developed.

HOW ARE THE RESULTS INTERPRETED

What do they mean for your breeding enterprise?

Genes occur in matching pairs on chromosomes, with one of each pair inherited from each parent. Genetic diseases occur when the DNA sequence in a gene is mutated.

The two types of genetic diseases that we test for in the 6-Panel GDT are **autosomal dominant** and **autosomal recessive**.

AUTOSOMAL RECESSIVE diseases require two copies of the mutation to cause disease. **Horses carrying one copy are unaffected and known as 'carriers'**. These carriers will still pass this mutation on to approximately half their offspring, so half their foals will also be carriers. **Mating to other carriers should be avoided because there is a 25% chance the resultant foal will be affected.**

AUTOSOMAL DOMINANT diseases require only one copy of the mutation to cause disease.

- **Horses carrying one copy of the mutation will be 'affected' to some degree and will pass this on to approximately half their offspring when mated to a non-carrier, so half their foals will be affected.**
- **Horses carrying two copies will always produce affected foals.**

GENETIC DISORDERS

Recessive v's Dominant – What is the Difference?

Genetic Information verified by Equine Genetic Research Centre (Racing Australia)

Genetic Disorder results for horses testing NEGATIVE n/n genotype, will be accepted for ASH Registration.

AUTOSOMAL RECESSIVE

HERDA

Hereditary Equine Regional Dermal Asthenia (HERDA) is a devastating disease that causes **severe skin blistering and lesions, leading to secondary infections and early death**. The condition renders horses unable to wear a saddle or harness, with age of onset ranging from birth to four years. **Affected horses are often euthanised**. Carriers of HERDA are relatively common, so **care should be taken to NOT mate two carriers together**.

- Horses with **n/n genotype** will not have HERDA and cannot transmit this HERDA variant to their offspring.
- Horses with **n/Hr or Hr/n genotype (single copy)** will not be affected by HERDA but **are CARRIERS**. They will transmit this HERDA variant to approx. 50% of their offspring. Matings between two carriers results in a 25% chance of producing an affected foal.
- Horses with **Hr/Hr genotype (double copy) will have HERDA, are AFFECTED**.
- **Horses with a single copy of HERDA will be ACCEPTED for ASH registration. Horses with a double copy will be REJECTED for ASH registration.**

GBED

Glycogen Branching Enzyme Deficiency is a metabolic genetic disease that is fatal in foetal and neonatal stages. **GBED prevents the muscle from properly storing glucose, and the affected foal will run out of energy, which will damage its organs**. Clinical signs include general weakness, low body temperature, seizures and difficulty rising. **GBED is always fatal, with most affected foals dying before the age of eight weeks. GBED can also cause abortions**.

- Horses with **n/n genotype** will not have GBED and cannot transmit this GBED variant to their offspring.
- Horses with **n/Gb or Gb/n genotype (single copy)** will not be affected by GBED but **are CARRIERS**. They will transmit this GBED variant to approx. 50% of their offspring.
- Horses with **Gb/Gb genotype (double copy) will have GBED, are AFFECTED and will die within days.**
- **Horses with a single copy of GBED will be ACCEPTED for ASH registration. Horses with a double copy will be REJECTED for ASH registration.**

OLWS

Overo Lethal White Syndrome is an inherited syndrome of foals born to parents of the overo coat-pattern lineage. Affected foals are born with blue eyes, pink skin and a white coat. OLWS results in **incomplete migration of nerve cells to the large intestine during embryonic development**; the caecum, colon and sometimes the rectum, undergo a large dilation and fill with faecal mass, and **affected foals may die within 72 hours of birth**.

- Horses with **n/n genotype** will not have the overo pattern and cannot transmit the OLWS variant to their offspring.
- Horses with **n/O or O/n genotype (single copy)** will have the overo pattern and **are CARRIERS**. They will transmit this OLWS variant to approx. 50% of their offspring.
- Horses with **O/O genotype (double copy) will have OLWS, are AFFECTED and will die within days.**
- **Horses with a single copy of OLWS will be ACCEPTED for ASH registration. Horses with a double copy will be REJECTED for ASH registration.**

Genetic Disorder Testing is available through the Society for Registered Australian Stock Horses for the following disorders:

OLWS, HERDA, PSSM1, HYPP, GBED, MYHM

AUTOSOMAL DOMINANT

PSSM1

Polysaccharide Storage Myopathy (PSSM) is a type of tying up caused by an abnormal build-up of sugars in muscle. Clinical signs include **painful cramps, muscle twitches, stiffness, sweating and reluctance to move**. Symptoms can vary considerably in severity. It can be controlled by **modifying the diet to reduce starch intake and with consistent exercise**.

- Horses with **n/n genotype** will not have PSSM1 and cannot transmit the PSSM1 variant to their offspring.
- Horses with **n/P1 or P1/n genotype (single copy) are AFFECTED** with PSSM1 and **may show signs of disease**. They will transmit the PSSM1 variant to approx. 50% of their offspring.
- Horses with **P1/P1 genotype (double copy)** are homozygous, **will have the PSSM1 variant** and **may be MORE SEVERELY AFFECTED** than n/P1 or P1/n horses. They will transmit this PSSM1 variant to 100% of their offspring.
- **Horses with a single or double copy of PSSM1 will be ACCEPTED for ASH registration.**

HYPP

Hyperkalemic Periodic Paralysis (HYPP) is a muscular disease caused by an inherited genetic mutation that leads to **uncontrolled muscle twitching or profound muscle weakness, unpredictable paralysis attacks, respiratory noises**, and **in severe cases, may lead to collapse and/or sudden death**. Symptoms can vary considerably in severity and can be managed to some extent by avoiding high potassium diets.

- Horses with **n/n genotype** will not have HYPP and cannot transmit this HYPP variant to their offspring.
- Horses with **n/H or H/n genotype (single copy) can display episodes of HYPP** and **are AFFECTED**. They will transmit this HYPP variant to approx. 50% of their offspring.
- Horses with **H/H genotype (double copy) can display episodes of HYPP** and **typically are MORE SEVERELY AFFECTED**. They will transmit this HYPP variant to 100% of their offspring.
- **Horses with a single or double copy of HYPP will be REJECTED for ASH registration.**

MYHM

Myosin-Heavy Chain Myopathy has two distinct clinical disease presentations, Immune-Mediated Myositis (characterized by episodes of **SEVERE MUSCLE ATROPHY** following an autoimmune event), or **SEVERE MUSCLE PAIN and DAMAGE** termed Non-Exertional Rhabdomyolysis or "tying-up" that is NOT associated with exercise and may (or may not) have muscle atrophy. The onset of disease is complex, and not all horses show clinical signs and onset may be related to an **IMMUNE CHALLENGE**, example: infection or vaccination.

- Horses with n/n genotype will not have MYHM and cannot transmit this MYHM variant to their offspring.
- Horses with **n/MYHM or MYHM/n genotype (single copy) are AFFECTED** by MYHM and **may show signs of disease**. They will transmit the MYHM variant to approx. 50% of their offspring.
- Horses with **MYHM/MYHM genotype (double copy)** are homozygous, **will have** the **MYHM variant** and is **more likely to be SEVERELY AFFECTED** than a horse with one copy. They will transmit this MYHM variant to 100% of their offspring.
- **Horses with a single or double copy of MYHM will be ACCEPTED for ASH registration.**

Genetic Disorder results for horses testing NEGATIVE n/n genotype, will be accepted for ASH Registration.