SEVERE COMBINED IMMUNODEFICIENCY (SCID)

By Beth Minnich © 2011

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History

Severe Combined Immunodeficiency (SCID) is perhaps the most well recognized genetic disorder associated with the Arabian breed. Broad awareness of SCID within the Arabian horse community extends back to the 1970’s and the test for SCID was one of the first DNA tests developed for an equine genetic disorder (along with Hyperkalemic Periodic Paralysis/ HYPP and Overo Lethal White Syndrome/OLWS). Even with this well established history, during the last few years with increased interest and attention on other genetic disorders, such as Cerebellar Abiotrophy (CA) and Lavender Foal Syndrome (LFS), it is important (especially for newcomers to the breed) not to think that SCID is a thing of the past and ignore it in light of more “en vogue” disorders. SCID is lethal disorder involving the immune system. Although caused by different mutations, immunodeficiency disorders are also found in other species including humans, mice and dogs. SCID is an autosomal recessive trait, caused by a mutation on chromosome 9, which results in a foal being born with an improperly functioning immune system.

During the 1960’s, clinical reports from Australia described a condition which would become known as SCID and in 1973, SCID was first reported in the veterinary literature by researchers at Washington State University (WSU)/USA. In 1977, a group of researchers at WSU proposed an autosomal recessive mode of inheritance, which was confirmed in 1980 based on breeding experiments conducted at WSU by a second research group. However, it wasn’t until 1997 that a direct DNA test for SCID was ultimately developed at the University of Texas/USA.

Clinical Signs

SCID is caused by a mutation involving the enzyme DNA-PKcs (DNA-protein kinase catalytic subunit) which is a key component of the immune defense system. Affected foals are born normal, however because the foal’s immune system is not functioning properly, as the antibodies the foal receives from its dam’s colostrum wear off, the foal is unable to fight off infection from bacteria and viruses found in the normal living environment. As such, the foal will eventually die from an opportunistic infection (most commonly from respiratory diseases, such as pneumonia), usually by about 5 months of age.

A preliminary diagnosis can be made using a blood sample from the foal. A lymphocyte count of <1,000/mm³ (a normal count is in the range of 2,500 – 3,000/mm³) and a lack of immunoglobin M (IgM) are indicative of SCID. Confirmation is available through DNA testing or through a necropsy showing a lack of development of the thymus and lymph nodes.

Supporting Research

Those who are newer to the breed may not fully appreciate how significant the SCID research effort was. However, those who have been involved with Arabians since the 1970’s/1980’s will remember well how much of a concern SCID was and how devastating the disorder was to some breeding programs. Through efforts of the international Arabian horse community, lead by the International Arabian Horse Association’s FOAL Commission in conjunction with the Morris Animal Foundation, Arabian horse owners and breeders helped raised over $200,000 in funding, along with providing valuable samples for advancing SCID research. In addition, SCID information provided by FOAL and the Arabian horse magazines was invaluable for helping to raise awareness of SCID and educating owners and breeders. Development of a test for SCID was such a significant accomplishment that “SCID Testing” was listed as #10 in the Arabian Horse Association’s (US) Centennial 100 voting. The Arabian horse community