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Exertional Rhabdomyolysis in a Swiss Warmblood horse family

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The objectives of this study were to determine if the glycogen synthase (*GYS1*) mutation, which causes polysaccharide storage myopathy (PSSM) in Quarter Horses, is associated with ER in a high-prevalence family of Warmblood horses and to describe signalment, clinical signs, and potential risk factors for development of ER in Warmblood horses. Three groups were evaluated: a family consisting of a sire with ER and 71 of his descendants, 17 unrelated Warmblood horses with ER and 66 unaffected unrelated control Warmblood horses. History on husbandry, feeding, use and performance was assessed by interviewing the horse owners using a standardized questionnaire. All horses were genotyped for *GYS1*. In 10 ER-affected family horses, muscle histopathology was evaluated. Signs of ER were reported in 39% of horses within the family. Fifty-one % of horses in the family, 12% of the unrelated Warmblood horses with ER and no controls possessed the *GYS1* mutation. Horses possessing the *GYS1* mutation in the high-prevalence family had a 7.1-times increased risk for developing ER compared to those with the normal genotype (*P*=0.0005). All muscle samples from horses in the family with ER showed polysaccharide accumulation typical for PSSM. Overall, mares were more frequently affected than males (OR 2.1, *P*=0.04). Environmental factors that affect clinical signs of ER were not identified. Compared to unaffected horses, ER-affected horses had a higher risk for poor rather than excellent willingness to perform (OR 11.0, *P*=0.003). Concluding, PSSM associated with the *GYS1* mutation is an identifiable cause of ER in Warmbloods. Due to its dominant mode of inheritance, breeding animals with the *GYS1* mutation results in a high prevalence of ER.