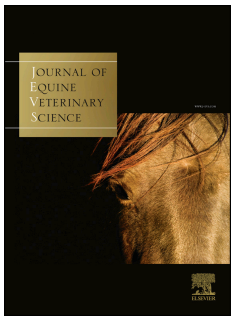
# Accepted Manuscript




Case Report: Hereditary Multiple Exostoses: A comprehensive examination of a 4 year old Dutch Warmblood male with maternal links to HME in 1st and 2nd generations

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**Case Report: Hereditary Multiple Exostoses: A comprehensive examination of a 4 year old Dutch Warmblood male with maternal links to HME in 1st and 2nd generations.**

**Abstract**

Hereditary Multiple Exostosis (HME) is a condition rarely diagnosed in horses that has not been previously reported in Dutch Warmblood horses. Its presentation resembles that diagnosed in humans and is predominantly active during periods of skeletal maturation. This case study reports a link between a Granddam with no obvious HME lesions and the 1st and 2nd generations presenting with HME, albeit with different sires. The severity in affected horses (n=5) varied from one foreleg to extensive multiple lesions of 123/205 bones in a half sibling in the 1st generation. In the 2nd generation, axial deviation was reported including similar lesions to those found in the 1st generation. Rib and limb lesions were the most commonly reported in the 1st and 2nd generations by age 4. However, the extensive examination of the 4 year Dutch Warmblood 1st generation male revealed lesions from the 1st cervical vertebra to the 1st caudal vertebra inclusive of rib and limb lesions. As all horses with this condition are untreatable, early detection and diagnosis would provide the owner / breeder with options to avoid financial loss and emotional stress.

**Key Words:** Hereditary multiple exostoses (HME), Osteochondroma, Skeleton, Cartilage, Axial Skeleton, Appendicular Skeleton.

## 1. Introduction

Hereditary Multiple Exostoses (HME) is a distinct skeletal disorder reported in humans, dogs, cats, lions, lizards, cattle and horses [1]. The synonyms in current literature include; Osteogenic disease; Chondral osteogenic dysplasia; Chondral osteoma; Dyschondroplasia; Deforming chondrodysplasia; Multiple hereditary osteochondromata; Multiple cartilaginous exostoses or Exostosis dysplasia [1]. Primarily, HME in horses affects endochondral bones during skeletal development and is often seen at birth. Gross skeletal presentation can predominantly be seen in the ribs, scapula and limbs (Figure 1), with lesions appearing as cartilage capped spongy chondro-osseous growths [2].

**Figure 1.** Four year old Dutch Warmblood male with HME of the ribs, scapula and carpus of the right foreleg (white arrows).

Familial relationships have previously been reported in the horse and human; one male line was identified in a Utah State University study in horses, and in humans, both male and female lineage was identified [2,3]. Diagnosis in horses is via radiographic evidence differentiating an Osteochondroma from HME, however in humans, radiographic evidence requires further diagnostic support [4]. Whereby in humans, HME is autosomal dominant with most displaying a mutation at the EXT-1 or EXT-2 genes; hence genetic diagnosis differentiates HME from other skeletal disorders such as metachondromatosis [5].