STATEMENT (CONCLUSIONS)

To the authors' knowledge, this is the first case series describing the clinical signs, MRI findings and outcome

of dogs diagnosed with this uncommon presentation of a common condition.

Suspected 3-hydroxy-3-methylglutaric aciduria (HMGA) in domestic shorthaired cats

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OBJECTIVES

3-hydroxy-3-methylglutaric aciduria (HMGA) is a metabolic disorder due to deficiency of 3-hydroxy-3-methylglutaryl-coenzyme A (HMG CoA) lyase, an enzyme involved in processing leucine and manufacturing ketones. HMGA is a rare (<1/100,000) autosomal recessive condition in humans, associated with metabolic crisis, stunted growth, psychomotor retardation, epileptic seizures and spasticity. We aim to describe the presentation of suspected HMGA in 4 cats.

METHODS

We describe the neurological and diagnostic findings in 4 stray kittens (2 consecutive litters) presented to a charity.

RESULTS

The only male kitten was presented at ~ 8 weeks old with a gait abnormality which progressed over a month until euthanasia. His litter sister had a slower course with initial signs at 12 weeks; ultimately euthanized at 5 months old. Two further female cats were presented at ~ 8 weeks old. All had an unusual gait with marked increased extensor muscle tone of the thoracic limbs giving a pronounced "goose stepping action" with spinocerebellar ataxia and intention tremor. They frequently fell, increased extensor tone and spasticity made rising difficult. Organic acid urinalysis from the first female revealed a HMGA of 45 mmol/mol creatinine (normal 0-2). Management with L-Carnitine at 100mg/kg/day did not result in clinical improvement. Post mortem samples were obtained from 3 of 4 cats; preliminary results showed axonal degeneration of olivopontocerebellar and spinocerebellar circuits. Genetic analysis is in process.

STATEMENT (CONCLUSIONS)

We describe a spontaneously occurring metabolic disorder characterised by marked increased extensor tone. Understanding the pathogenesis may inform on analogous conditions in all species