SURVEILLANCE

Congenital paunch calf syndrome in Ireland

WE would like to report the occurrence of congenital paunch calf syndrome in Ireland. This is a homozygous recessive condition in the Romagnola breed that causes congenital deformities as described by Gentile and others (2004). Carrier animals appear normal.

A full-term stillborn purebred Romagnola calf examined at Kilkenny Regional Veterinary Laboratory had severe abdominal distension (hence the term paunch calf), subcutaneous oedema, ascites, a lobulated firm liver with two small (5 mm and 10 mm) blood-filled cysts, a small (1 cm) atrial septal defect, slight shortening of the face and bilateral exopthalmos. No abnormality of brain or palate was found. Apart from exopthalmos, these findings exactly match the gross description given by Gentile and others (2004).

Histological examination confirmed hepatic fibrosis. There was moderate to marked portal fibrosis and some portal-portal bridging fibrosis; mild to moderate periacinar fibrosis; perisinusoidal fibrous trabeculae of varying thickness and focally extensive areas of capsular fibrosis. These findings are consistent with paunch calf syndrome (Gentile and others 2004).

Genetic testing was carried out by Weatherbys DNA laboratory. Testing procedures used the customised international dairy and beef version 2 (IDBv2) SNP (single nucleotide polymorphism) chip (Mullen and others 2013, McClure and others 2014). IDBv2 SNP chip content includes probes specific for the KDM2B c.2503G>A mutation, which is associated with congenital paunch calf syndrome (Testoni and others 2012). The KDM2B c.2503G>A probes were validated using DNA supplied by Dr Cord Droegemueller from confirmed affected and carrier congenital paunch calf syndrome animals. Testing confirmed that the Irish animal was homozygous for the c.2503G>A mutation, hence confirming the congenital paunch calf syndrome diagnosis.

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