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PEROSOMUS ELUMBIS IN A STILLBORN RHESUS MACAQUE (*Macaca mulatta*): A CASE REPORT

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Abstract

Perosomus Elumbis (PE) is a rare congenital disorder characterized by absence of caudal spine (lumbar, sacral, and coccygeal vertebrae). Here, we present the first reported case of PE in a rhesus macaque (*Macaca mulatta*), and relate our findings to those described in other species.

Keywords

Congenital; non-human primate; stillborn; *Perosomus Elumbis*

1. INTRODUCTION:

Perosomus Elumbis (PE) is a rare congenital disorder characterized by absence of caudal spine (lumbar, sacral, and coccygeal vertebrae) [1–4]. Significant gross findings often include hypoplasia of the pelvic bones and hind limbs, with contracture of the hind limb muscles [1–4]. PE frequently presents in conjunction with one or more anomalies of the urogenital or gastrointestinal systems, including, renal agenesis [1, 4], cryptorchidism [2], and atresia ani [2–4]. PE is well described in domestic animals including cattle [1, 4], buffalo [5], sheep [2], and horses [3]. PE has never been reported in non-human primates (NHPs). Here, we describe the first case of PE in a rhesus macaque (*Macaca mulatta*), and relate our findings to those described in other species.

2. CASE REPORT

A 128-day stillborn female fetus was presented for necropsy. The fetus was 16.5 cm long and weighed 0.298 kg. The pelvis and hind limbs were disproportionately small and there was marked contracture of the hind limbs (arthrogryposis). Both feet were medially rotated (*talipes varus*) (Figure 1A–D), and there was a flap of skin which extended across the caudal aspect of the knee joint (popliteal webs) (Figure 1C). The lumbar, sacral, and coccygeal vertebrae were absent and the pelvis did not articulate with the remaining thoracic spine

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(spinopelvic disassociation) (Figure 1D). The thoracic and cervical vertebrae were grossly normal. Other abnormalities included a horseshoe shaped kidney (renal fusion) positioned on the midline at the level of the iliac crest (Figure 1B), absent left adrenal gland, hypoplastic mandible and maxilla (Figure 1A), and excess visceral adipose tissue. Radiographs performed post-fixation, demonstrated a complete absence of lumbar, sacral, and coccygeal vertebrae (Figure 1D). Longitudinal and cross sections of the soft tissue in the area of the lumbar spine showed areas of haphazardly oriented nerve fibers, neurons, and glial cells, but lacked the organization of normal spinal cord; there was a complete absence of bone. Other tissues were histologically unremarkable.

The dam of the fetus was acquired from another facility in 2013. Since acquisition, she had 5 offspring, of which 1 was aborted (no necropsy performed), one was stillborn (grossly autolyzed, no histopathology performed), and one was found dead (18 day old, severely autolyzed, no food in stomach, no histopathology was performed), one living offspring, and one stillborn with severe malformations (present case). The maternal blood sugar level at the time of birth of this fetus was 247 mg/dL (range 39-83mg/dL). At the time of manuscript submission, the dam was euthanized for unrelated reasons. Histopathological examination of dam showed presence of amyloid in the islets of Langerhans.

3. DISCUSSION

Perosomus Elumbis (PE) is a rare congenital malformation featuring agenesis of the caudal spine, well described in domestic animals **including Danish Holstein cattle** [1], **a sheep** [2], **a thoroughbred foal** [3], **a Holstein calf** [4], **and a buffalo** [5], and experimentally induced in mice [6]. The etiology of PE is not known and likely multifactorial; however, in Holstein cattle there is thought to be a strong heritable component [1, 2, 4]. In this case, the lumbar, sacral, and coccygeal vertebrae were absent and additional abnormalities included horseshoe kidney, unilateral adrenal gland agenesis, mandibular-maxillary hypoplasia, arthrogryposis, and excess visceral adipose tissue. The gross and radiographic appearance of the spine and hind limbs, particularly arthrogryposis, and features like club foot (*talipes varus*), popliteal webs, and horseshoe kidney were consistent with lesions reported in other species [1–5, 7–10]. Lesions such as unilateral adrenal gland agenesis [1], excess visceral adipose tissue, and craniofacial malformations such as lateral-lateral narrowing of the skull [3], brachygnathia [5], and synophthalmia [2] have also been reported with PE.

The term PE is commonly used in veterinary literature, while broadly similar lesions in humans have been categorized as caudal regression syndrome (CRS) and lumbo-sacral agenesis based upon the location of the lesion and the stage of embryological neurulation affected [8]. Both PE and CRS are characterized by segmental agenesis of caudal spine and share the aforementioned external and visceral abnormalities [1, 2, 4, 11, 12]. PE and CRS appear to refer to the same complex of congenital malformations rooted in embryological developmental anomalies. Most cases of PE (animals) are lethal, while in CRS, human patients with surgical interventions have lived well into adulthood [9, 13] [9] [14]. Spinopelvic dissociation is common in animals with PE [1, 2, 4, 11], while in most reported cases of CRS the caudal spine is fused to some portion of pelvis. In humans, spinopelvic disassociation is more often reported in cases of stillbirth and infant mortality [15–17].

Histologic findings common to CRS and PE include myelodysplasia of the caudal spinal cord, hydromyelia, and replacement of musculature with adipose tissue [1, 9, 15]. Some other congenital malformations associated with CRS include imperforate anus, hemivertebrae, kyphosis, pulmonary hypoplasia, hydrocephalus [8], costal dysplasia, diaphragmatic hernia, and cardiovascular defects (dextrocardia, ventricular septal defects, atrial septal defects, and patent ductus arteriosus) [8, 10]. While the degree of disability and dysfunction depend heavily on the level of the defect and extra-axial malformations, even severely affected patients frequently retain some sensory ability in their lower limbs [9]. The extensive overlap in the malformations of CRS and PE is suggestive of a shared pathogenesis probably rooted in a disturbance of early embryogenesis [8, 10, 17]. Maternal diabetes has been suggested to be a risk factor for CRS [8, 13, 17, 18]. The dam's hyperglycemic status on the date of delivery of this fetus (247mg/dL; range 39-83mg/dL) and diagnosis of amyloidosis in the dam's pancreas are strong circumstantial evidence, however, the lack of a regular workup of blood glucose levels precludes us to confirm gestational diabetes as a predisposing cause in this case.

4. CONCLUSION

This is the first reported case of PE in a non-human primate; documentation of additional cases are needed to further characterize the condition in NHPs and perhaps develop it as an animal model for CRS in humans. Development of such an animal model would lead to better characterization of risk factors and development of therapeutic modalities for this life crippling condition in humans.

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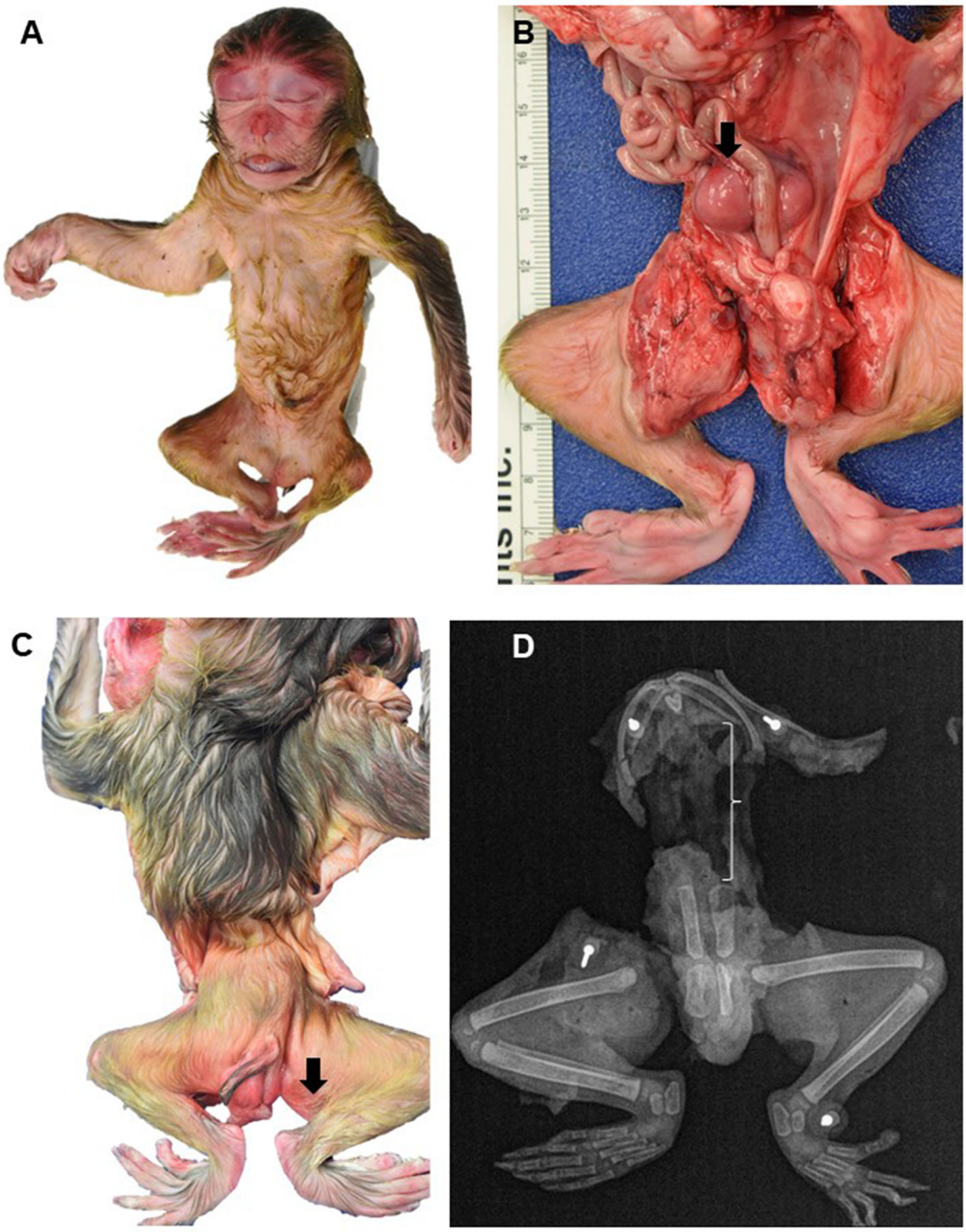

Figure1.

Gross presentation of rhesus macaque fetus with *Perosomus Elumbis*. A) Ventro-dorsal view of macaque fetus showing disproportionately small hind limbs with arthrogryposis and inwardly rotated feet. B) Opened abdomen showing fused kidney at the midline (arrow). C) Dorso-ventral view of the macaque fetus showing popliteal webs on caudal thigh skin (arrow). D) Dorso-ventral radiograph showing complete loss of lumbar, sacral, and coccygeal vertebrae. The white parenthesis shows the loss of lumbar vertebrae with only soft tissues occupying the space from the end of the thoracic vertebrae at the level of the xiphoid

cartilage to the iliac crest. NOTE: Bright white objects are pins used to hold the cadaver steady during radiographic procedure.