Primary ciliary dyskinesia in a Staffordshire bull terrier

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INTRODUCTION

Primary ciliary dyskinesia (PCD) is a diverse group of inherited structural and functional abnormalities of the respiratory and other cilia, which results in recurrent respiratory tract infections. Primary ciliary dyskinesia was diagnosed in a 14-week old Staffordshire bull terrier that had a history of respiratory disease from 7 weeks of age. Pneumonia was diagnosed on thoracic radiographs and transtracheal aspirate. Transmission electron microscopy of the bronchi and trachea indicated the presence of both primary and secondary ciliary dyskinesia. The most prominent primary defects consisted of absent inner dynein arms, absent radial spokes and absence of the central microtubules. These defects accounted for 62 % of the total number of cross-sections screened. Non-specific ciliary abnormalities encountered most often were compound cilia, swollen cilia, addition/deletion of peripheral doublets and disorganised axonemes (26 %). To the authors’ knowledge, this is the first case of PCD described in the Staffordshire bull terrier and the first report of PCD in South Africa.

CASE REPORT

Infectious tracheobronchitis was diagnosed in a 2-month-old male Staffordshire bull terrier. Pneumonia was diagnosed on thoracic radiographs and transtracheal aspirate. Transmission electron microscopy of the bronchi and trachea indicated the presence of both primary and secondary ciliary dyskinesia. The most prominent primary defects consisted of absent inner dynein arms, absent radial spokes and absence of the central microtubules. These defects accounted for 62 % of the total number of cross-sections screened. Non-specific ciliary abnormalities encountered most often were compound cilia, swollen cilia, addition/deletion of peripheral doublets and disorganised axonemes (26 %). To the authors’ knowledge, this is the first case of PCD described in the Staffordshire bull terrier and the first report of PCD in South Africa.

Key words: congenital, electron microscopy, immotile cilia, pneumonia.

bull terrier and treated with 50 mg Synulox® (Pfizer Laboratories, Rivonia Road, Sandton, South Africa) twice a day for 5 days. Despite treatment, the dog deteriorated in that the coughing had not resolved and the dog developed a mucoid-purulent ocular-nasal discharge. An inflammatory reaction was evident on peripheral blood smear evaluation and lung consolidation was present on survey thoracic radiographs. The dog was treated for bacterial pneumonia with fluid therapy and intravenous antibiotics. Although there was an improvement, the cough never resolved. At 11 weeks of age a culture taken from the nasal and pharyngeal areas yielded a non-resistant Chryseobacterium luteola, which is an opportunistic organism. As there was no resolution of the cough the patient was referred to Bryanston Veterinary Hospital for further evaluation at 13 weeks of age.

On clinical examination the dog was bright and alert, rectal temperature was normal, and had an irritable trachea, that evoked a cough response on palpation. Left shift neutrophilia and monocytosis was evident on haematology. Survey thoracic radiographs indicated a severe diffuse bronchopneumonia. On bronchoscopy the trachea and main-stem bronchi had a normal appearance but there was an accumulation of mucopurulent material in the distal trachea. Degenerative neutrophils, macrophages, and free and phagocytosed bacteria were present on transtracheal aspirate cytology, which is consistent for a bacterial infection. Transtracheal aspirate culture yielded resistant Bordetella bronchiseptica that was only sensitive to cefotaxime. At this point the owners opted for euthanasia as the long-term prognosis was guarded. The owners consented to removal of a portion of the distal trachea and main-stem bronchi for electron microscopy.

The main-stem bronchi and a portion of the distal trachea were fixed in 10% buffered formalin and processed for transmission electron microscopy by standard techniques using post-fixation in 1% osmium tetroxide, dehydration through a graded ethanol series, and embedding in an epoxy resin. Ultra-thin sections were contrasted with uranyl acetate and lead citrate and examined in a Philips CM10 transmission electron microscope (FEI, Eindhoven, The Netherlands) operated at 80 kV.

Specimen areas containing transversely sectioned cilia were recorded, with the bronchial sections yielding the most cross-sectioned ciliary profiles. Normal as well as deviations from the 9+2 microtubular arrangement were recorded into PCD or secondary ciliary dyskinesia according to the classification of Pizzi et al.7 (Table 1). The most prominent primary defects consisted of absent inner dynein arms, absent radial spokes, and absence of the central microtubules (Figs 2). These defects accounted for 62% of 401 cross-sections screened. The most frequently encountered non-specific ciliary abnormalities were compound cilia, swollen cilia (Figs 3, 4), addition/deletion of peripheral doublets, and disorganised axonemes (26%).

**DISCUSSION**

Primary ciliary dyskinesia occurs in dogs, cats, pigs, and people6, with the incidence in people being 1 per 16 000 births8. The disease usually manifests within the first few weeks of life; however, some animals have mild clinical signs that are not recognised until later in life (6 months to 10 years)2. Clinical signs include chronic muco-purulent nasal discharge, productive cough, exercise intolerance, and dyspnoea4. The dog described in this report showed early onset recurrent...
upper and lower respiratory tract disease that was poorly responsive to antibiotic therapy. On clinical examination nasal discharge and tracheal sensitivity were evident, both of which have been reported in the literature\textsuperscript{2,4,9}. As in the dogs in 1 report\textsuperscript{9}, this dog was bright and alert, despite the presence of severe respiratory disease and an inflammatory leukogram on haematology.

Reported radiographic findings in PCD include bronchitis, bronchiectasis, and consolidating pneumonia\textsuperscript{4}. The dog in this report showed severe bronchopneumonia, without any obvious signs of bronchitis or bronchiectasis, which may have developed had the dog lived longer. On bronchoscopy it showed the accumulation of purulent material within the trachea, which was most likely due to failure in the mucociliary elevator. Tracheal culture also yielded only normal flora.

Further evaluation of a patient with PCD requires either tracheal or nasal scintigraphy or EM\textsuperscript{2}. Although tracheal mucosal transit times measured by nuclear scintigraphy are suggestive of defective mucociliary clearance\textsuperscript{2}, a definitive diagnosis requires the electron microscopic evaluation of ciliary ultrastructure from nasal or respiratory biopsies, nasal cytobrush, or semen\textsuperscript{2,4,9}. Typical findings in PCD include shortening or loss of dynein arms, atypical microtubular orientation, and duplication or deletion of central or outer microtubule doublets\textsuperscript{2,4}. The dog in the report showed 62\% abnormal cilia, with defects being absent inner dynein arms, absent radial spokes, and absence of the central microtubules. Normal animals may show structural abnormalities in 2–5\% of cilia\textsuperscript{2}.

Therapy for PCD is aimed at controlling infections and facilitating respiratory secretions, with cough suppressants being contra-indicated\textsuperscript{4}. Reports of long-term survival of dogs with PCD are limited, with the majority of cases either dying or euthanased within 6 months of diagnosis\textsuperscript{2}. Response to therapy in this dog was poor and it was euthanased because of the poor prognosis.

It is likely that PCD is more common than reported, because affected dogs may be misdiagnosed as having fading puppy syndrome, aspiration pneumonia, or infectious causes of pneumonia. This report illustrates that PCD should be considered in a young dog with recurrent respiratory signs.

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**REFERENCES**

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