VETERINARY TECHNICAL DATASHEET

Primary Ciliary Dyskinesia, (PCD)

WISDOM HEALTH™

Mutation Found In :Old English Sheepdog

Disorder Type

- Lung

Disease Severity

- Moderate

Background

Primary ciliary dyskinesia (PCD) is a disorder found to affect ciliary formation in multiple breeds. Cilia of the body are found in the respiratory tract, genitourinary system, and middle ear. Cilia in the lungs work to keep the airways clear by propelling mucus and dirt onward in the respiratory tract. PCD is inherited in an autosomal recessive manner.

Key Signs

- Respiratory tract inflammations
- Infertility in males and females
- Hearing difficulties

Clinical Description

The clinical signs of PCD include respiratory tract inflammation such as pneumonia and also infertility caused by inadequate cilia function in the sperm and oviduct. Hearing difficulties can occur as well. If primary ciliary dyskinesia is associated with situs inversus (mirrored internal organs), the condition is called Kartagener syndrome. When situs inversus occurs, the heart for example, is located on the right side of the chest.

Mode of Inheritance

- autosomal recessive

Gene Name

- CCDC39

Next Steps

Therapy is limited to symptomatic treatments for resulting infections and general supportive care. There is no cure.

References

Merveille AC, Davis EE, Becker-Heck A, Legendre M, Amirav I, Bataille G, Belmont J, Beydon N, Billen F, Clément A, Clercx C, Coste A, Crosbie R, de Blic J, Deleuze S, Duquesnoy P, Escalier D, Escudier E, Fliegauf M, Horvath J, Hill K, Jorissen M, Just J, Kispert A, Lathrop M, Loges NT, Marthin JK, Momozawa Y, Montantin G, Nielsen KG, Olbrich H, Papon JF, Rayet I, Roger G, Schmidts M, Tenreiro H, Towbin JA, Zelenika D, Zentgraf H, Georges M, Lequarré AS, Katsanis N, Omran H, Amselem S. CCDC39 is required for assembly of inner dynein arms and the dynein regulatory complex and for normal ciliary motility in humans and dogs. Nat Genet 43(1):72-78, 2011.