

Primary Ciliary Dyskinesia

Primary Ciliary dyskinesia, in dogs, is a rare genetic (inherited) disorder with defective structure and/or function of cilia that can lead to chronic respiratory or otic disease, reversed organs, fertility problems, and even hydrocephalus. Electron microscopy can be used to examine the cilia for defects in the ultrastructure of the cilia associated with ciliary dyskinesia.

Proper fixation and shipment of the sampled tissues are important for accurate diagnosis. Please follow the guidelines below:

Sample tissues:

Nasal, bronchial, or tracheal biopsies may be submitted. The patient should be free of respiratory infections prior to obtaining the biopsy, if possible, as secondary changes associated with inflammation can confound diagnosis.

All specimens should be placed in a fixative immediately after obtaining said specimens. **DO NOT SEND FRESH SPECIMENS OR SPECIMENS IN ANY LIQUID OTHER THAN AN APPROPRIATE EM FIXATIVE OR NEUTRAL-BUFFERED FORMALIN!**

Oviduct and sperm (infection, or testes) * are optimal samples because they are typically without associated infection; however, we realize this is not possible in patients who have been spayed/neutered. If submitting reproductive tissue, it would be advantageous to send a biopsy from the respiratory area as a second sample, and in male dogs, at least 1 ml of semen added to 4 ml of fixative before neutering.

Male dogs occasionally have aspermia, and female dogs at certain stages of the cycle have few ciliated cells in the oviduct. Therefore, there is always a possibility of limited ciliated cells in the reproductive tissues.

*If sending reproductive organs (ovaries [include the Fallopian tubes/oviducts] or testicles [epididymis attached please]), we recommend placing them in Neutral-Buffered formalin and sending them as you would biological samples for histology.

Fixation of tissues:

The best fixative for any of the tissues sampled is a buffered glutaraldehyde fixative (2% - 3% glutaraldehyde in a buffer pH 7.2 – 7.4). Biopsies may be bisected or even trisected to allow proper penetration of the fixative during shipping. A maximum of 2-3mm in any one direction is a good rule of thumb for proper fixation with glutaraldehyde. The amount of fixative should more than double the tissue sample in the container used for shipping. Respiratory brushings and sperm ejaculate (up to 1mL in volume) are added to a 4-5mL container of fixative.

If glutaraldehyde is not readily available, the next best fixative to use is Neutral-Buffered Formalin (NBF), which is used in most histology laboratories. If NBF is on hand and has not

expired, it will be acceptable to send the sample tissues. Make sure to indicate which fixative is used on the submission form (type and percentage in buffer).

Here at GEM, we will no longer send EM fixative to your facility. We recommend purchasing glutaraldehyde from Electron Microscopy Sciences. Here are a few suggested items for ordering: <https://www.emsdiasum.com/glutaraldehyde-2>

Glutaraldehyde, Pre-Made, Ready-to-Use, in Buffer:

Storage	Catalog #	Description	Pack
2-8°C	16539-45	Glutaraldehyde 3% in Sodium Phosphate Buffer, pH 7.2	10 x 10 mL vials
2-8°C	16536-06	Glutaraldehyde 2% Paraformaldehyde 2% in Phosphate Buffer, pH 7.4	10 x 10 mL vials
2-8°C	16538-06	Glutaraldehyde 3% Paraformaldehyde 3% in Phosphate Buffer, pH 7.4	10 x 10 mL vials

Submission forms and past history:

Include as much history as possible about the patient. Copies of previous reports or other data relevant to the patient's case may be added or attached to our submission form. Use our EM Submission Form when submitting tissue samples. The form is fillable; fill in what is applicable including: (1) choose the right fixative; (2) place "Ciliary Dyskinesia" under the Clinical Diagnosis; (3) the service requested is Transmission Electron Microscopy; and (4) give us the person to receive the invoices under Accountant Name & Email.

Shipment of specimens:

Send all samples double contained overnight with any accompanying forms. There are instructions on our EM Submission Form. Glutaraldehyde-fixed samples are shipped under refrigeration (cold pack) overnight. Formalin-fixed samples are shipped overnight; cold pack recommended during summer months. Days of the week to ship are Monday through Thursday. Ship as you would any Biological Specimen to the following address:

Georgia Electron Microscopy
University of Georgia
ISTEM Research Bldg 1, Suite 1000
302 East Campus Road
Athens, GA 30602
(706) 542-5537

It is important to pass on the tracking information to our facility as soon as your sample ships. Do not hesitate to notify us a sample is on its way. Timing is essential when receiving samples like this for a more accurate diagnosis.

Turnaround time:

The current turnaround time for the heavy caseload of diagnostic specimens is 4-6 months. We know all submissions are considered rush samples, and we are doing the best we can under staffing limitations. We are hoping 2025 will be a better year to “catch up” on cases taking longer than normal. We are running into cases where the tissues submitted do not have enough cilia. Please make sure the proper area is excised of the respiratory tract which anatomically contains cilia. Testicles have been the most problematic in locating cilia in the efferent ductules. Feel free to contact our facility if you would like an update of your case; we will give you a status where it is in the processing queue. 2024 proved to be the busiest we have ever been with ciliary dyskinesia submissions; and these are in addition to the routine workload GEM receives. We ask for your patience as we work through each case, giving the utmost attention to proper protocols in processing and sectioning of your submitted specimens.

Estimated fees and invoicing:

Beginning January 1, 2025:

- (a) Electron microscopy for ciliary dyskinesia specimens: \$462.00 – if a second sample is submitted, the extra fee may be waived in some cases. Normally, the fee is \$40 more (\$502.00).
- (b) Pathologist’s fee for evaluation and reporting has increased: could be one of three charges: \$75.00, \$150.00, or \$225.00 depending on how much time spent analyzing the images and reporting results.
- (c) Total cost, one sample: \$537.00, \$612.00, or \$687.00.
- (d) Your clinic will receive one invoice from us at the completion and reporting of your submission. We would appreciate payment within 30 days of receiving the invoice.
- (e) There are no discounts for running this procedure. We are a cost-recovery core facility, and we charge based on the supplies, equipment, and technology time used to achieve the imaging required for optimal results.

If there are any other questions, either Mary Ard or Dr. Elizabeth Howerth should be able to answer them.

Updated January 2025