

PCD Diagnostics Service

Dear Ladies and Gentlemen,

Primary ciliary dyskinesia (PCD) is a difficult-to-diagnose disease. In 2017, the European Respiratory Society (ERS) published new guidelines for the diagnosis of PCD. The interdisciplinary PCD Diagnostic Center at the University of Bern (PCD-UNIBE) offers a comprehensive PCD diagnostic service since January 2019, which complies with the new guidelines and the latest state of research.

Epithelial cell material obtained by means of a nasal brushing (performed with one small brush per nostril) is analyzed immediately after collection by high-speed video microscopy (HSVM) and examined for the presence of ciliary proteins by immunofluorescence (IF). It is also prepared to culture re-differentiated cell cultures (with basal, mucus-producing and ciliated cells). These cell cultures are re-evaluated by HSVM and can then be used for analysis by transmission electron microscopy (TEM) and IF. The use of cell cultures has three advantages: (1) More reliable evaluation with HSVM due to larger cell groups compared to primary samples. (2) Less influence due to secondary changes (e.g. mucus or infections during the sampling). (3) Avoidance of re-brushings.

In principle, all examinations (apart from TEM and genetics) are intended for an initial assessment in order to provide a most reliable diagnosis. However, depending on the findings, individual methods can be skipped. Those decisions are made within interdisciplinary meetings of the PCD Diagnostic Center. Normally, you will receive a provisional result (by email or telephone) after the primary HSVM (until approx. one week after brushing), as well as a definitive report after analysis of the cell cultures (approx. three months after brushing; if TEM is performed, after approx. four months).

Our offer costs between CHF 1350.- and max. CHF 3600.- (TARMED tariffs) and includes the following modules:

- **Execution and processing of nasal brushing** (CHF 85.-, CHF 56.- for external brushing [only after prior consultation with us])
- **Videomicroscopic analysis (HSVM)** of the beating pattern and frequency of the primary sample and cell culture (CHF 405.-; CHF 202.- if primary sample only)
- **Immunofluorescence:** staining of ciliary proteins and corresponding analysis (CHF 330-660.-, depending on number of proteins to be stained)
- **Cell culture** for HSVM, IF and TEM (CHF 560.-)
- **Transmission electron microscopy** (depending on effort, approx. CHF 1000.- for TEM of cell culture, approx. CHF 1850.- for TEM of fresh sample)
- **Diagnosis** as synthesis of all methods by the interdisciplinary committee (CHF 72.-)

Of course, only those methods required and performed for an assessment will be invoiced. Due to the relatively high costs, we recommend to consider obtaining a cost credit from the health insurance in advance.

Since 2019, we will also collaborate for PCD diagnostics with the University Children's Hospital Zurich (Dr. Andreas Hector and Dr. Andreas Jung, Senior Physicians Pediatric Pneumology and specialists in PCD diagnostics) and the University Children's Hospital Lausanne (Dr. Sylvain Blanchon, Head Pediatric Pneumology, PCD Specialist) to discuss difficult cases together. If other colleagues are interested to join, we are, of course, happy to extend the group.

We look forward to (further) cooperate with you and will be happy to answer any questions you may have (pcd@insel.ch).

For the interdisciplinary PCD Diagnostic Centre University of Bern



Loretta Müller, PhD

Pediatric Pneumology, Inselspital
Coordinator PCD Diagnostics



Stefan Tschanz, PD Dr. med.

Institute of Anatomy, University of Bern
Specialist HSVM and TEM



Carmen Casaulta, Dr.

Pediatric Pneumology, Inselspital
Head Scope "Rare Lung Diseases"



Philipp Latzin, Prof. Dr.

Pediatric Pneumology, Inselspital
Head of department Ped. Pneumology

Information about the registration process:

1. Contact the "Interdisciplinary PCD Diagnostic Centre University of Bern" via pcd@insel.ch
2. Possible dates: Nasal brushings can be performed and processed **Monday-Thursday**.
3. Principally, we prefer the patients to be sent to our PCD consultation service for diagnostics, so that we potentially can perform further diagnostic methods (e.g. measurement of nasal NO) depending on the patient's medical history and clinical state.
However, we still accept - in justified cases - external samples, preferably taken by experienced persons. The procedure is then as follows:
 - a. Approximately one week before the date of the planned brushing, we will send you the registration form as well as a tube with culture medium and two brushes.
 - b. You organize a courier service to bring the sample to our laboratory within 2-3 hours.
Address: DBMR - LPM, Pediatric Pneumology (Lab 377), Loretta Müller/Andrea Stokes, Murtenstrasse 28, 3008 Bern.
 - c. We kindly ask you to give us the medical history as detailed as possible (e.g. on the referring form) so that we can consider this for the diagnostic syntheses within the interdisciplinary meetings.