Title: Clinical manifestations of primary ciliary dyskinesia: A review of the literature

Ms. Myrofora 6686 Goutaki mgoutaki@ispm.unibe.ch MD ¹, Mrs. Elisabeth 6696 Maurer emaurer@ispm.unibe/ch ¹, Dr. Philipp 6697 Latzin Philipp.Latzin@insel.ch MD ², Dr. Carmen 6698 Casaulta Carmen.Casaulta@insel.ch MD ² and Prof. Dr Claudia 6687 Kuehni kuehni@ispm.unibe.ch MD ¹. ¹ Institute of Social and Preventive Medicine, University of Bern, Bern, Switzerland and ² Dept of Paediatrics, University Children’s Hospital of Bern, Bern, Switzerland.

Body: Aim Data on clinical manifestations of Primary Ciliary Dyskinesia (PCD) are scarce. We aimed to review all publications that describe prevalence and severity of PCD related morbidity and to show changes in morbidity with age. Methods We searched PubMed to identify studies published since 1990, describing prevalence and severity of signs and symptoms in PCD patients. We identified 488 original papers. We excluded studies presenting only data on diagnostic evaluations or imaging, studies with <10 patients, written in non-European languages (N=1) or studies reporting on the same patients (N=3). Results 12 studies met our inclusion criteria. Together, they described 440 patients (mean per study 37, range 17-58). 7 came from pulmonary departments, mostly pediatric, 5 from ENT clinics. 4 included children, 5 children and young adults and 3 individuals of all ages. Only 1 study presented results stratified by age, allowing to investigate age-related changes. Clinical manifestations commonly described were: rhinitis, cough, otitis media, sinusitis, pulmonary infections and neonatal respiratory distress; none was assessed in all studies. There was a great heterogeneity in type of data (patient reported vs. chart reviews). Symptom prevalence varied between studies, many of which included highly selected populations (e.g. prevalence of sinusitis: 23-54% in pulmonary units, 59-100% in ENT clinics). Few studies described manifestations affecting other than the respiratory system (e.g. infertility). Conclusion This review underlines the necessity to study PCD in large unselected groups of patients, to allow to describe the natural history of the disease. Funding FP7 grant 305404, SNF 32003B-144068, Lungenliga Bern.