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First patient registry launched for rare lung disease, primary ciliary dyskinesia

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The first international patient registry has been launched for primary ciliary dyskinesia (PCD) - a rare lung disease causing long-term and recurring respiratory infections, with no approved treatments and no cure.

Although progress has been made in understanding the disease in recent years, there is a lack of fundamental data on the number of cases of the disease, how the disease affects different people and the effects of different treatment options. Patient registries are effective tools to collect data from people with a rare disease to assess their condition, how it develops and how it responds to treatments.

PCD is a hereditary condition affecting children and occurs when a child inherits a "faulty" gene from their parent. It affects the cilia—tiny, microscopic moving structures that line the airways, ears and sinuses. Symptoms can include inability to remove mucus from the lungs, persistent blocked nose and sinusitis. It may lead to diffuse bronchiectasis (widening of the bronchi) and eventually to long-term respiratory failure.

In this new study, published online today (9 December, 2015) in the *European Respiratory Journal*, researchers describe how they established an international online platform to systematically collect data on cases of PCD diagnoses, symptoms displayed by patients, treatment and progression of the disease. The team worked closely with leading clinicians across the world and collaborated with several patient

organisations.

To date, 201 patients have been included in the registry. Patients are recruited by their healthcare professional once written consent has been obtained. The data will continue to be collected and used for benchmarking projects to help improve diagnosis and management of PCD. The registry will also serve as a platform to recruit patients for clinical trials to help develop new treatments for the disease.

Dr. Claudius Werner, lead author of the study from the University Hospital Münster, in Germany, said: "PCD is a rare, but debilitating, disease, which currently has numerous uncertainties related to its diagnosis and treatment. We believe this new online tool will advance our understanding of this rare disorder, help us recruit candidates for research studies and ultimately improve care for PCD patients.

"We will continue to build on the data in the registry by recruiting more referral centres to support this work, recruiting more patients and cleaning the data ready for analysis and a full comprehensive report of all items collected. We also plan to establish regional registry administrators to further disseminate knowledge about the registry and to facilitate access to the registry for patients from countries currently outside the current consortium."

More information: *European Respiratory Journal*, dx.doi.org/10.1183/13993003.00776-2015

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