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Pediatric Respiratory Medicine
Imperial College, London, UK

Prof. Dr. Kris De Boeck
Kinderlongziekten en mucoviscidosereferentiecentrum
Katholieke Universiteit Leuven
Universitair Ziekenhuis Leuven.

Prof. Dr. Christiane Knoop
Service de Pneumologie et Centre de Mucoviscidose
Hôpital Erasme, Université Libre de Bruxelles

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Kindergeneeskunde, Universitair Ziekenhuis Brussel
Vrije Universiteit Brussel.

Prof. Dr. Inge Gies
Kindergeneeskunde, Universitair Ziekenhuis Brussel
Vrije Universiteit Brussel.

Prof. Dr. Eef Vanderhelst
Pneumologie en mucoviscidosereferentiecentrum
Universitair Ziekenhuis Brussel
Vrije Universiteit Brussel

Prof. Dr. Yvan Vandenplas, Chair
Kindergeneeskunde, Universitair Ziekenhuis Brussel
Vrije Universiteit Brussel

Prof. Dr. Anne Malfroot, Promotor
Kinderlongziekten en mucoviscidosereferentiecentrum
Universitair Ziekenhuis Brussel
Vrije Universiteit Brussel

VRIJE UNIVERSITEIT BRUSSEL

PhD in Medical Sciences
2016-2017

INVITATION to the Public defence of

Elke DE WACHTER

To obtain the academic degree of **'DOCTOR IN MEDICAL SCIENCES'**

Challenges in making the diagnosis of Cystic Fibrosis in the 21st century.

Monday 26 June 2017
Auditorium **Vanden Driessche**, 18:00
Faculty of Medicine and Pharmacy, Laarbeeklaan 103, 1090 Brussel

How to reach the campus Jette:
http://www.vub.ac.be/english/infoabout/campuses

Summary of the dissertation

Cystic fibrosis (CF) is mostly diagnosed with a sweat test. Otherwise, a genetic confirmation can be made if two CF-causing mutations are found on each CFTR-allele (Cystic Fibrosis Transmembrane Conductance Regulator). Insights in the basic defect of CF have resulted in the recognition of a wide spectrum of disease severity related to CFTR-protein dysfunction. Apart from the 'classic' CF patients, having pancreatic insufficiency and respiratory disease resulting in a reduced life expectancy, other conditions with CFTR-dysfunction exist. Demarcation lines between these different entities are blurred. This thesis aims to give insights in the barriers a clinician can face in making a CF diagnosis.

Nasal potential difference (NPD) measurement can be helpful in diagnostic inconclusiveness. In a semi-blinded study we showed that two different NPD methods (needle vs abrasion) could equally discriminate CF from non-CF individuals. The two methods were equally accepted by the study subjects, concluding that the operator's preferred NPD-method may be used.

Little is known about rare CFTR-mutations (RM), present in only few CF-patients. To investigate RMs in Belgium, a CF-registry study was conducted. We found out that 6.5% of the Belgian CF patients carry at least one RM. Sixty-four RMs were found, of which 21 had not been previously reported in the global mutation database. As a group, these patients had milder disease compared to controls with classic CF; however a wide spectrum in disease severity was seen. To understand the disease liability of these RMs, a prospective collection of electrophysiological and clinical data is needed. This effort will enable to support or withdraw a CF diagnosis in patients with rare mutations in the future

Curriculum Vitae

Elke De Wachter was born on January 23rd 1973 in Willebroek, Belgium. After studying mathematics-science in the Koninklijk Technisch Atheneum Willebroek she started medical school at the Vrije Universiteit Brussel (VUB) in 1991. She obtained her medical degree with great distinction in June 1998. She gained experience in the centre of revalidation for CF in De Haan and the CF-centre of Groningen and UZ Brussel (formerly AZ-VUB). After obtaining her degree as paediatrician at the VUB in 2003, she continued working in the CF-clinic of the UZ Brussel and the department of paediatric pulmonology. From 2008 on, her interest in nasal potential difference (NPD) measurement resulted in the education of this technique in the Universitätsklinikum from Giessen (Germany) and the Royal Brompton Hospital in London (UK). Since 2009 she is an active member of the European CF Society (ECFS) - Diagnostic Network Working Group. She obtained the ECFS-certification for NPD-measurements in 2013 and participated in international studies applying the NPD-technique. She is a member of the scientific committee of the ECFS (diagnosis and screening; from 2014-2017). At national level she is a member of the board of the Belgian CF-registry. Since 2016 she is the president of the medical committee of the Belgian CF-centres.

In her hospital she is in charge of diagnostic difficulties for CF, the paediatric allergy clinic and runs the paediatric pulmonology clinic under supervision of Prof Dr Anne Malfroot. She is a clinical tutor within the paediatric course at the Faculty of Medicine at the VUB and (co-) authored over 20 publications in the field of cystic fibrosis and paediatric pulmonology.

Elke is married to Frank Pauwels and mother of two wonderful children Sam (2006) and Aline (2010).