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## Genetic Diagnosis and Respiratory Management of Primary Ciliary Dyskinesia

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# LIST OF PUBLICATIONS





Paff T, Kooi LE, Moutaouakil Y, Riesebos E, Siermans EA, Daniels JMA, Weiss MM, Niessen HWM, Haarman EG, Pals G, Micha D. Diagnostic yield of a targeted gene panel in Dutch primary ciliary dyskinesia patients. *Submitted to Human Mutation*.

Paff T, Daniels JMA, Weersink EJ, Lutter R, Vonk Noordegraaf A, Haarman E. A randomised controlled trial on the effect of inhaled hypertonic saline on quality of life in primary ciliary dyskinesia. *Eur Respir J*. 2017; 49(2) pii:1601770.

Paff T, Loges NT, Aprea I, WU K, Bakey Z, Haarman EG, Daniels JMA, Siermans EA, Bogunovic N, Dougherty GW, Höben IM, Große-Onnebrink J, Matter A, Olbrich H, Werner C, Pals G, Schmidts M, Omran H, Micha D. Mutations in *PIH1D3* cause X-linked primary ciliary dyskinesia with outer and inner dynein arm defects. *Am J Hum Genet*. 2017; 100(1):162-168.

Lucas JS, Paff T, Goggin P, Haarman E. Diagnostic Methods in Primary Ciliary Dyskinesia. *Paediatr Respir Rev* 2016; 18:8-17.

Paff T, van der Schee M, Brinkman P, van Aalderen WMC, Haarman E, Sterk P. Breathomics in lung disease. *Chest* 2015; 147(1):224-231.

Joensen O, Paff T; Haarman EG, Skovgaard IM, Jensen ØP; Bjarnsholt T; Nielsen KG. Exhaled breath analysis using electronic nose in cystic fibrosis and primary ciliary dyskinesia patients with chronic pulmonary infections. *PLoS One* 2014; 9(12)e115584.

Paff T, Daniels JMA, Pals G, Haarman EG. Primary ciliary dyskinesia: from diagnosis to molecular mechanisms. *J Pediatr Genet* 2014; 3(2):115-127.

de Meij TG, Larbi IB, van der Schee MP, Lentferink YE, Paff T, Terhaar Sive Droste JS, Mulder CJ, van Bodegraven AA, de Boer NK, Electronic nose can discriminate colorectal carcinoma and advanced adenomas by fecal volatile biomarker analysis: proof of principle study. *Int J Cancer* 2014; 134 (5):1132-1138.

Paff T, van der Schee M, Daniels J, Pals G, Postmus P, Sterk P, et al. Exhaled molecular profiles in the assessment of cystic fibrosis and primary ciliary dyskinesia. *J Cyst Fibros* 2013;12(5):454-460.

Paff T, Onoufriadis AD, Shoemark A, Micha D, Kuyt B, et al. Splice site mutations in the axonemal outer dynein arm docking complex gene *CCDC114* cause primary ciliary dyskinesia. *Am J Hum Genet* 2013; 92(1):88-98.

Paff T, Oudesluys-Murphy AM, Wolterbeek R, Swart-van den Berg M, de Nie JM, Tijssen E, et al. Screening for refractive errors in children: the plusoptiX S08 and the Retinomax K-plus2 performed by a lay screener compared to cycloplegic retinoscopy. *J AAPOS* 2010; 14(6):478-83.

