## **CORRESPONDENCE**

# Cutting edge genetic studies in primary ciliary dyskinesia

We would describe our genetic studies in primary ciliary dyskinesia as 'cutting edge', rather than 'beyond the fringe'. Indeed, we predict that in 5 years genetic testing will be more readily available and used worldwide for diagnostic studies in primary ciliary dyskinesia than high speed ciliary waveform analysis. Would Drs Bush and Hogg like to make a wager?

# Michael R Knowles, <sup>1</sup> Margaret W Leigh, <sup>2</sup> Maimoona A Zariwala <sup>3</sup>

<sup>1</sup>Cystic Fibrosis/Pulmonary Research and Treatment Center, University of North Carolina at Chapel Hill, Chapel Hill, North Carolina, USA; <sup>2</sup>Department of Pediatrics, University of North Carolina, Chapel Hill, North Carolina, USA; <sup>3</sup>Department of Pathology and Lab Medicine, University of North Carolina, Chapel Hill, North Carolina, USA

**Correspondence to** Dr Michael R Knowles, Cystic Fibrosis/Pulmonary Research and Treatment Center, University of North Carolina at Chapel Hill, 7019 Thurston

Bowles Building, CB# 7248, Chapel Hill 27599, North Carolina, USA; knowles@med.unc.edu

#### Competing interests None.

**Provenance and peer review** Commissioned; internally peer reviewed.

Accepted 11 January 2012 Published Online First 10 February 2012

*Thorax* 2012;**67**:464. doi:10.1136/thoraxjnl-2012-201609

#### REFERENCES

- Knowles MR, Leigh MW, Carson JL, et al. Mutations of DNAH11 in primary ciliary dyskinesia patients with normal ciliary ultrastructure. Thorax 2012;67:433—41.
- Hogg C, Bush A. Genotyping in primary ciliary dyskinesia: ready for prime time, or a fringe benefit? Thorax 2012;67:377—8.

# Authors' response

We thank Dr Knowles and colleagues for their interest in our editorial. The single gene locus responsible for cystic fibrosis was discovered more than 20 years ago, and the vast majority of patients with cystic fibrosis are still diagnosed on a functional measure, namely the sweat test. The issue for the diagnosis of Primary ciliary dyskinesia (PCD)

is surely not what is available, but what is accurate. And in response to the proposed wager, we are always happy to take candy from babies!

### Andrew Bush,1 Claire Hogg2

<sup>1</sup>National Heart and Lung Institute, Imperial College, London, UK; <sup>2</sup>Paediatric Respiratory, Royal Brompton Hospital, London, UK

**Correspondence to** Dr Andrew Bush, National Heart and Lung Institute, Imperial College, London, UK; a.bush@imperial.ac.uk

**Contributors** AB and CH contributed equally to the manuscript.

#### Competing interests None.

**Provenance and peer review** Commissioned; internally peer reviewed.

Accepted 13 January 2012 Published Online First 10 February 2012

*Thorax* 2012;**67**:464. doi:10.1136/thoraxjnl-2012-201620

#### **REFERENCE**

 Knowles MR, Leigh MW, Zariwala MA. Cutting edge genetic studies in primary ciliary dyskinesia. *Thorax* 2012;67:464.

464 Thorax May 2012 Vol 67 No 5