European Journal of Human Genetics (2014) 22, 957

www.nature.com/ejhg

LETTER

A more fitting term in the incidental findings debate: one term does not fit all situations

European Journal of Human Genetics (2014) **22,** 957; doi:10.1038/ejhg.2013.266; published online 20 November 2013

We applaud Christenhusz *et al*[1] for their search for a more fitting term to capture the nuances of the 'incidental findings' debate. Their review of the various terminologies currently being applied or proposed is informative but not necessarily dispositive.

The authors find that the term secondary variant 'best avoids the problems raised by other options as well as accurately describing the phenomenon', yet we believe that any successful term needs to capture situations where the so-called secondary variant is the only (clinically significant) variant found, that is, there is no primary variant. The use of the term 'secondary variant' may also suggest a temporal relationship where one finding is the first, the other the second. We therefore suggest that rather than clarifying the situation the use of primary and secondary variant terminology may serve only to exacerbate confusion in clinical practice.

The authors highlight that new genetic technologies are capable of returning far more information than focussed genetic testing ever could. Yet how this information is construed depends on the question originally asked. If a genome is simply sequenced, that is to say, there is no targeting of the test, then arguably there is no such thing as an incidental or a secondary finding. Yet, if particular genetic diagnoses are suspected or sought then a finding might be unexpected, incidental to the reason for the test, but equally pertinent to the patient and their healthcare. Labelling it as 'secondary' will not help.

Christenhusz *et al* also suggest that the term 'variant' is sufficiently neutral, avoiding the difficulties associated with words such as 'risk' or 'abnormality'. However, clinical genetic practice routinely describes possible outcomes from genetic testing, including polymorphisms as variants, not associated with known health risks. Using the term 'secondary variant' to describe a known predisposition to disease will be confusing.

We suggest there is no one term that adequately captures all perspectives, and different terms may need to be used in different settings. The two main arguments put forward to avoid the term incidental finding are that incidental suggests that the finding is not serious and that a finding cannot be incidental if it has been actively looked for. Both points, we believe, can be adequately addressed with patients during the consent process for testing, with examples to illustrate the types of incidental findings that can occur and that the nature of the test we are organising means that we may identify other things, incidental to the original reason for the test. After all, whatever terminology is adopted will need explanation: patients are no more likely to understand what secondary variants mean. We support Christenhusz *et al* that a consensus on terminology and definitions is important for clarity but suggest that more than one term will be required to cover the different meanings in different settings.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

Gillian Crawford*[,1,2], Angela Fenwick[1] and Anneke Lucassen[1,2]
[1]*Clinical Ethics and Law at Southampton (CELS), Faculty of Medicine, University of Southampton, Southampton, UK;*
[2]*Wessex Clinical Genetics Service, The Princess Anne Hospital, Southampton, UK*
E-mail: gc@soton.ac.uk

1 Christenhusz GM, Devriendt K, Dierickx K: Secondary variants – in defense of a more fitting term in the incidental findings debate. *Eur J Hum Genet* **21**: 1331–1334; advance online publication, May 22, 2013; doi:10.1038/ejhg.2013.89.