Re: Pitfalls of automated comparative sequence analysis as a single platform for routine clinical testing for NF1 (Messiaen and Wimmer)

We are grateful to the authors for pointing out the errors in assignment of the predicted effect of some of the sequence variants that we reported. The same sense mutations Q282Q, C680C, K1724K, and R1808R are incorrectly recorded as protein truncating in table 2. The Y489C and G629R mutations are previously reported splice mutations. The E91X sequence change is 271G→T. The D176E and R873C sequence variants should have been recorded only in table 3. We apologise for these errors.

However, this does not detract from the main message of our paper that all of these sequence variants were ascertained using a single method. The reported method is an accurate and efficient way of testing for sequence changes.

J L Whittaker, C Mattocks, D Baralle, P Tarpey, C ffrench-Constant, M Bobrow
Department of Medical Genetics, Box 134, Addenbrooke’s Hospital, Cambridge, UK

Correspondence to: J L Whittaker, Department of Medical Genetics, Box 134, Addenbrooke’s Hospital, Cambridge, UK; joanne.whittaker@addenbrookes.nhs.uk

Competing interests: none declared

Reference