LING337: Assignment 2 Option 1

Use the three texts below to describe how the language of science changes with changes in audience purpose and mode.
You should comment on:

- Genre(s)
- Technical language
- Lexical density
- Nominal groups and nominalisation
- Information organisation
- Relationship between writer and readers
- How writers establish their credibility

Text 1 is taken from *New Scientist*, Text 2 is from a popular science website and Text 3 was published in the journal *Nature*.

Length: 2000 words

Due date: April 15th 2016
(NB: This is the end of the first week of the mid-semester break. If you are going away, you will need to submit your assignment earlier than the due date, or make sure that you can submit on-line from your holiday destination.)

Weighting: 40%

Marking criteria

Your lecturer will use the following criteria to assess your assignment:

The student:

- identifies key features of each text with regard to each of the 7 nominated areas
- identifies and discusses changes in the texts from text 1 – text 3
- relates the analysis to the readings studied in weeks 1-6 of the unit. You must make explicit reference to at least five readings.
- structures the assignment as an information report (see handout on iLearn in folder named *Improving you academic writing*)
- gives adequate attention to cohesive argument, grammatical construction and technical conventions (grammar, spelling, punctuation)
- uses APA referencing procedures and includes a reference list

Assignment submission and return

- This assignment needs to be submitted to Turnitin using the link in the Assessments file on iLearn
- Add a footer to each page of the assignment, with page numbering and the unit code in the footer
- Type double-spaced

Marked assignments will be available on iLearn, and the mark registered in Grademark (on iLearn) usually 4 weeks after submission. An announcement will be posted on iLearn when marking is completed.

Late Submissions

Late submissions may attract a penalty of 5% of total marks per day. To avoid a penalty, please email the unit co-ordinator, Jean Brick (jean.brick@mq.edu.au) before the due date requesting an extension and explaining why you need one.

Ancient European hunter-gatherer was a blue-eyed boy

New Scientist, 26 January 2014 by Catherine de Lange

An ancient hunter-gatherer whose remains were found in a Spanish cave has a genome surprisingly similar to modern humans. The male, who lived 7000 years ago, had blue eyes and a host of immunity genes that were thought to have evolved later.

Blue-eyed boy *(Image: CSIC)*

In 2006, two exceptionally preserved human skeletons were found in a cave in Leon, Spain. Carles Lalueza-Fox of Pompeu Fabra University in Barcelona extracted DNA from one of the skeleton's teeth, and his team has now sequenced the whole genome.

"This is the first pre-agricultural European genome we have," says Lalueza-Fox. "It will help us to understand how the arrival of the Neolithic era – farming, new diet and new diseases related to animals – has shaped the genome of modern Europeans."

Farming came to Europe 7500 years ago, but didn't reach Spain until 1500 years later, says Lalueza-Fox. With it came livestock breeding and fixed settlements, replacing the hunter-gatherer lifestyle.

Farming genes

Genetic analyses have already hinted at some of the changes caused by the rise of farming, as people evolved to deal with shifts in diet and exposure to animal diseases. For instance, lactose tolerance – the ability to drink milk as an adult – probably evolved when farming spread.

The new genome backs this up: the Spanish hunter-gatherer seems to have been lactose-intolerant. But other aspects of his genome took Lalueza-Fox by surprise. "My predictions were completely wrong," he says.

For instance, the hunter-gatherer had the genes for darkly pigmented skin and hair like his African ancestors, but also blue eyes, which are a more European trait. "This suggests eye colour came first," says Lalueza-Fox, rather than lighter skin.

It makes sense that lighter skin evolved in cooler climates, perhaps to deal with a lack of vitamin D, says Mark Thomas of University College London. Light eye colour is harder to

explain. "What's the use?" he asks. It may be that blue eyes evolved by sexual selection, in which one or both sexes preferred partners with blue eyes for some cultural reason.

Healthy genome

The other surprise lay in the hunter-gatherer's immune system. His immune system genes, and genes that affect the risk of bacterial infection, were similar to those of modern humans. Previously, it was thought that many immunity genes evolved in farmers – partly to cope with diseases that spread due to close contact with animals, and partly because farmers lived in large, stationary populations through which disease could spread more easily.

Lalueza-Fox speculates that these hunter-gatherers may have been exposed to diseases like cholera, which are not spread by animals.

It's not the first time that changes thought to be brought about by farming have been shown to precede it. Earlier this year, it emerged that rotten teeth were common in some hunter-gatherers long before farming, and sugar-rich diets, became common.

Journal reference: *Nature*, DOI: 10.1038/nature12960

Text 2:

Science

http://www.news.com.au/technology/science/ancient-europeans-had-dark-skin-and-blue-eyes-researchers-say/story-fn5fsgyc-1226810985900

http://www.news.com.au/

Ancient Europeans had dark skin and blue eyes, researchers say

- 7 days ago January 27, 2014 5:52AM

Researchers say light-skinned Europeans, like actress Tilda Swinton, emerged "much later" than once believed - possibly only in the Neolithic era when erstwhile hunter-gatherers became farmers. Picture: AFP *Source:* AFP

THE DNA of a hunter-gatherer who lived in Spain some 7,000 years ago suggests that Europeans were dark-skinned until much more recently than previously thought, according to researchers.

Genetic material recovered from a tooth of La Brana 1, an ancient man whose skeleton was dug up in a deep cave system in Spain in 2006, revealed a strange combination of dark skin and blue eyes, according to a study in the journal *Nature*.

Europeans from the Mesolithic Period between 10,000 and 5,000 years ago, when La Brana lived, were thought to have already been fair-skinned due to low ultraviolet radiation levels at these high latitudes.

"Until now, it was assumed that light skin colour evolved quite early in Europe, (during) the Upper Palaeolithic... But this is clearly not the case," study co-author Carles Lalueza-Fox from Spain's Evolutionary Biology Institute, told AFP.

"This individual had the African variants for the pigmentation genes."

The Upper Palaeolithic or Late Stone Age stretched from 50,000-10,000 years ago, followed by the Mesolithic or Middle Stone Age that lasted until about 5,000 years ago, when it was followed in Europe by the Neolithic or New Stone Age.

Lalueza-Fox said light-skinned Europeans emerged "much later" than once believed - possibly only in the Neolithic era when erstwhile hunter-gatherers became farmers.

The cause, he said, may have been a change in diet and lower vitamin D intake associated with this lifestyle change.

In the absence of natural vitamin D, the human skin can produce its own in contact with the sun - but dark skins synthesise much less than fair ones - creating an evolutionary incentive for change.

In La Brana 1, Lalueza-Fox and his team also found the genetic signature for blue eyes and dark hair.

While the exact hue of skin cannot be determined, the scientists say its combination with blue eyes was not to be found in modern Europeans today.

It is widely accepted that Man's oldest common forefather was dark skinned, and that people became more pale as they moved further north out of Africa into colder climates with less sunlight.

Subsequent migrations and mixing created the wide range of hues we have today.

La Brana's genome is the first of a European hunter-gatherer to be fully sequenced.

When compared to today's humans, it was found to be most closely genetically related to northern Europeans like the Swedes or Fins.

The probe also found that La Brana 1 had not yet acquired the genetic mutation that allowed later humans to digest milk and starch more easily - an adaptation that probably coincided with the birth of agriculture in the Neolithic age.

The authors said more Mesolithic genomes will have to be analysed to determine how widespread La Brana's genomic traits really were.

Text 3:

Derived immune and ancestral pigmentation alleles in a 7,000-year-old Mesolithic European

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Nature (2014)
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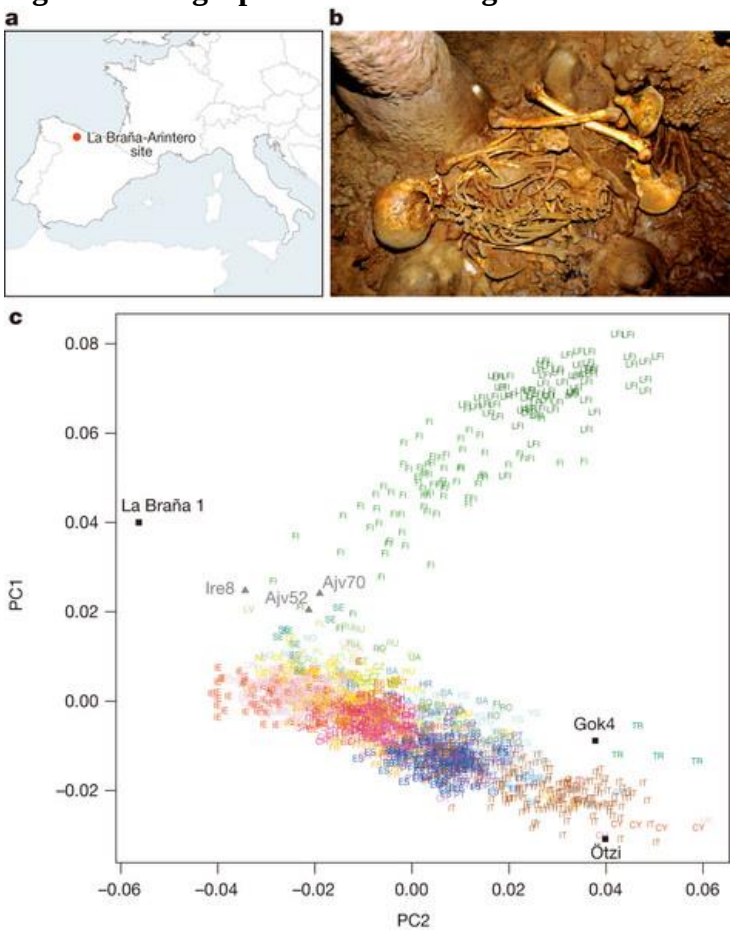
Ancient genomic sequences have started to reveal the origin and the demographic impact of farmers from the Neolithic period spreading into Europe[1, 2, 3]. The adoption of farming, stock breeding and sedentary societies during the Neolithic may have resulted in adaptive changes in genes associated with immunity and diet[4]. However, the limited data available from earlier hunter-gatherers preclude an understanding of the selective processes associated with this crucial transition to agriculture in recent human evolution. Here we sequence an approximately 7,000-year-old Mesolithic skeleton discovered at the La Braña-Arintero site in León, Spain, to retrieve a complete pre-agricultural European human genome. Analysis of this genome in the context of other ancient samples suggests the existence of a common ancient genomic signature across western and central Eurasia from the Upper Paleolithic to the Mesolithic. The La Braña individual carries ancestral alleles in several skin pigmentation genes, suggesting that the light skin of modern Europeans was not yet ubiquitous in Mesolithic times. Moreover, we provide evidence that a significant number of derived, putatively adaptive variants associated with pathogen resistance in modern Europeans were already present in this hunter-gatherer.

Main

Next-generation sequencing (NGS) technologies are revolutionizing the field of ancient DNA (aDNA), and have enabled the sequencing of complete ancient genomes[5, 6], such as that of Ötzi, a Neolithic human body found in the Alps[1]. However, very little is known of the genetic composition of earlier hunter-gatherer populations from the Mesolithic period (circa 10,000–5,000 years before present, bp; immediately preceding the Neolithic period).

The Iberian site called La Braña-Arintero was discovered in 2006 when two male skeletons (named La Braña 1 and 2) were found in a deep cave system, 1,500 m above sea level in the Cantabrian mountain range (León, Northwestern Spain) (Fig. 1a). The skeletons were dated to approximately 7,000 years bp (7,940–7,690 calibrated bp)[7]. Because of the cold environment and stable thermal conditions in the cave, the preservation of these specimens proved to be exceptional (Fig. 1b). We identified a tooth from La Braña 1 with high human DNA content (48.4%) and sequenced this specimen to a final effective genomic depth-of-coverage of 3.40× (Extended Data Fig. 1).

Figure 1: Geographic location and genetic affinities of the La Braña 1 individual.



a, Location of the La Braña-Arintero site (Spain). **b**, The La Braña 1 skeleton as discovered in 2006. **c**, PCA based on the average of the Procrustes transformations of individual PCAs with La Braña 1 and each of the five Neolithic samples[1, 3]. The reference populations are the Finnish HapMap, FINHM and POPRES. Population labels with labelling of ref. 12 with the addition of FI (Finns) or LFI (late-settlement Finns). Ajv70, Ajv52, Ire8 and Gok4 are Scandinavian Neolithic hunter-gatherers and a farmer, respectively[3]. Ötzi is the Tyrolean Ice Man[1].

We used several tests to assess the authenticity of the genome sequence and to determine the amount of potential modern human contamination. First, we observed that sequence reads from both the mitochondrial DNA (mtDNA) and the nuclear DNA of La Braña 1 showed the typical ancient DNA misincorporation patterns that arise from degradation of DNA over time[8] (Extended Data Fig. 2a, b). Second, we showed that the observed number of human DNA fragments was negatively correlated with the fragment length ($R^2 > 0.92$), as expected for ancient degraded DNA, and that the estimated rate of DNA decay was low and in agreement with predicted values[9] (Extended Data Fig. 2c, d). We then estimated the contamination rate in the mtDNA genome, assembled to a high depth-of-coverage (91×), by checking for positions differing from the mtDNA genome (haplogroup U5b2c1) that was previously retrieved with a capture method[2]. We obtained an upper contamination limit of 1.69% (0.75–2.6%, 95% confidence interval, CI) (Supplementary Information). Finally, to generate a direct estimate of nuclear DNA contamination, we screened for heterozygous positions (among reads with >4× coverage) in known polymorphic sites (Single Nucleotide Polymorphism Database (dbSNP) build 137) at uniquely mapped sections on the X chromosome[6] (Supplementary Information). We found that the proportion of false

heterozygous sites was 0.31%. Together these results suggest low levels of contamination in the La Braña 1 sequence data.

To investigate the relationship to extant European samples, we conducted a principal component analysis (PCA)[10] and found that the approximately 7,000-year-old Mesolithic sample was divergent from extant European populations (Extended Data Fig. 3a, b), but was placed in proximity to northern Europeans (for example, samples from Sweden and Finland)[11, 12, 13, 14]. Additional PCAs and allele-sharing analyses with ancient Scandinavian specimens[3] supported the genetic similarity of the La Braña 1 genome to Neolithic hunter-gatherers (Ajv70, Ajv52, Ire8) relative to Neolithic farmers (Gok4, Ötzi) (Fig. 1c, Extended Data Figs 3c and 4). Thus, this Mesolithic individual from southwestern Europe represents a formerly widespread gene pool that seems to be partially preserved in some modern-day northern European populations, as suggested previously with limited genetic data[2, 3]. We subsequently explored the La Braña affinities to an ancient Upper Palaeolithic genome from the Mal'ta site near Lake Baikal in Siberia[15]. Outgroup f_3 and D statistics[16, 17], using different modern reference populations, support that Mal'ta is significantly closer to La Braña 1 than to Asians or modern Europeans (Extended Data Fig. 5 and Supplementary Information). These results suggest that despite the vast geographical distance and temporal span, La Braña 1 and Mal'ta share common genetic ancestry, indicating a genetic continuity in ancient western and central Eurasia. This observation matches findings of similar cultural artefacts across time and space in Upper Paleolithic western Eurasia and Siberia, particularly the presence of anthropomorphic 'Venus' figurines that have been recovered from several sites in Europe and Russia, including the Mal'ta site[15]. We also compared the genome-wide heterozygosity of the La Braña 1 genome to a data set of modern humans with similar coverage (3–4×). The overall genomic heterozygosity was 0.042%, lower than the values observed in present day Asians (0.046–0.047%), Europeans (0.051–0.054%) and Africans (0.066–0.069%) (Extended Data Fig. 6a). The effective population size, estimated from heterozygosity patterns, suggests a global reduction in population size of approximately 20% relative to extant Europeans (Supplementary Information). Moreover, no evidence of tracts of autozygosity suggestive of inbreeding was observed (Extended Data Fig. 6b).

To investigate systematically the timing of selection events in the recent history of modern Europeans, we compared the La Braña genome to modern populations at loci that have been categorized as of interest for their role in recent adaptive evolution. With respect to two recent well-studied adaptations to changes in diet, we found the ancient genome to carry the ancestral allele for lactose intolerance[4] and approximately five copies of the salivary amylase (*AMY1*) gene (Extended Data Fig. 7 and Supplementary Information), a copy number compatible with a low-starch diet[18]. These results suggest the La Braña hunter-gatherer was poor at digesting milk and starch, supporting the hypotheses that these abilities were selected for during the later transition to agriculture.

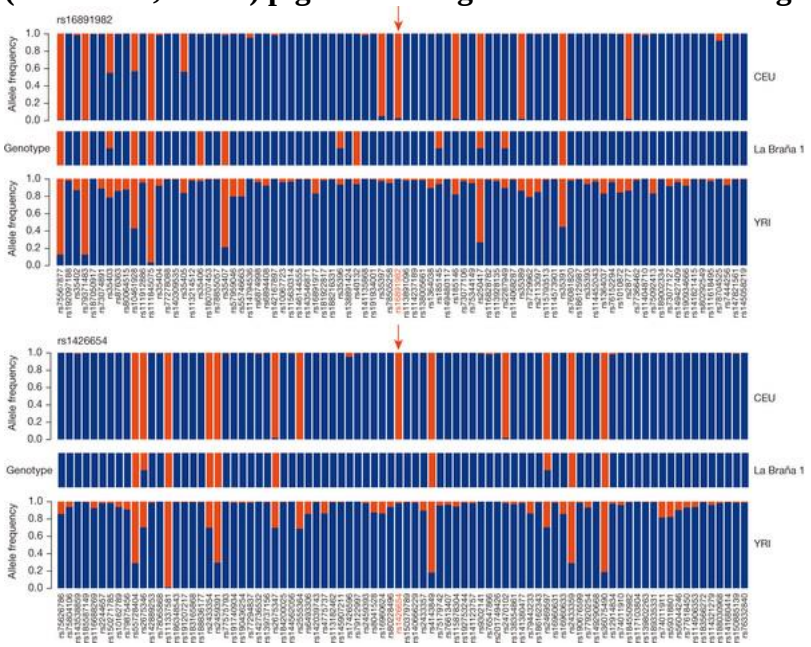
To expand the survey, we analysed a catalogue of candidate signals for recent positive selection based on whole-genome sequence variation from the 1000 Genomes Project[13], which included 35 candidate non-synonymous variants, ten of which were detected uniquely in the CEU (Utah residents with northern and western European ancestry) sample [19]. For each variant we assessed whether the Mesolithic genome carried the ancestral or derived (putatively adaptive) allele.

Of the ten variants, the Mesolithic genome carried the ancestral and non-selected allele as a homozygote in three regions: *C12orf29* (a gene with unknown function), *SLC45A2* (rs16891982) and *SLC24A5* (rs1426654) (Table 1). The latter two variants are the two

strongest known loci affecting light skin pigmentation in Europeans[20, 21, 22] and their ancestral alleles and associated haplotypes are either absent or segregate at very low frequencies in extant Europeans (3% and 0% for *SLC45A2* and *SLC24A5*, respectively) (Fig. 2). We subsequently examined all genes known to be associated with pigmentation in Europeans[22], and found ancestral alleles in *MC1R*, *TYR* and *KITLG*, and derived alleles in *TYRP1*, *ASIP* and *IRF4* (Supplementary Information). Although the precise phenotypic effects cannot currently be ascertained in a European genetic background, results from functional experiments[20] indicate that the allelic combination in this Mesolithic individual is likely to have resulted in dark skin pigmentation and dark or brown hair. Further examination revealed that this individual carried the *HERC2* rs12913832*C single nucleotide polymorphism (SNP) and the associated homozygous haplotype spanning the *HERC2–OCA2* locus that is strongly associated with blue eye colour[23]. Moreover, a prediction of eye colour based on genotypes at additional loci using HIrisPlex[24] produced a 0.823 maximal and 0.672 minimal probability for being non-brown-eyed (Supplementary Information). The genotypic combination leading to a predicted phenotype of dark skin and non-brown eyes is unique and no longer present in contemporary European populations. Our results indicate that the adaptive spread of light skin pigmentation alleles was not complete in some European populations by the Mesolithic, and that the spread of alleles associated with light/blue eye colour may have preceded changes in skin pigmentation.

Table 1: Mesolithic genome allelic state at 10 nonsynonymous variants recently selected in Europeans

Figure 2: Ancestral variants around the *SLC45A2* (rs16891982, above) and *SLC24A5* (rs1426654, below) pigmentation genes in the Mesolithic genome.



The SNPs around the two diagnostic variants (red arrows) in these two genes were analysed. The resulting haplotype comprises neighbouring SNPs that are also absent in modern Europeans (CEU) (n = 112) but present in Yorubans (YRI) (n = 113). This pattern confirms that the La Braña 1 sample is older than the positive-selection event in these regions. Blue, ancestral; red, derived.

For the remaining loci, La Braña 1 displayed the derived, putatively adaptive variants in five cases, including three genes, *PTX4*, *UHRF1BP1* and *GPATCH1 (*ref. 19), involved in the immune system (Table 1 and Extended Data Fig. 8). *GPATCH1* is associated with the risk of

bacterial infection. We subsequently determined the allelic states in 63 SNPs from 40 immunity genes with previous evidence for positive selection and for carrying polymorphisms shown to influence susceptibility to infections in modern Europeans (Supplementary Information). La Braña 1 carries derived alleles in 24 genes (60%) that have a wide range of functions in the immune system: pattern recognition receptors, intracellular adaptor molecules, intracellular modulators, cytokines and cytokine receptors, chemokines and chemokine receptors and effector molecules. Interestingly, four out of six SNPs from the first category are intracellular receptors of viral nucleic acids (*TLR3*, *TLR8*, *IFIH1* (also known as *MDA5*) and *LGP2*)[25].

Finally, to explore the functional regulation of the genome, we also assessed the La Braña 1 genotype at all expression quantitative trait loci (eQTL) regions associated to positive selection in Europeans (Supplementary Information). The most interesting finding is arguably the predicted overexpression of eight immunity genes (36% of those with described eQTLs), including three Toll-like receptor genes (*TLR1*, *TLR2* and *TLR4*) involved in pathogen recognition[26].

These observations suggest that the Neolithic transition did not drive all cases of adaptive innovation on immunity genes found in modern Europeans. Several of the derived haplotypes seen at high frequency today in extant Europeans were already present during the Mesolithic, as neutral standing variation or due to selection predating the Neolithic. *De novo* mutations that increased in frequency rapidly in response to zoonotic infections during the transition to farming should be identified among those genes where La Braña 1 carries ancestral alleles.

To confirm whether the genomic traits seen at La Braña 1 can be generalized to other Mesolithic populations, analyses of additional ancient genomes from central and northern Europe will be needed. Nevertheless, this genome sequence provides the first insight as to how these hunter-gatherers are related to contemporary Europeans and other ancient peoples in both Europe and Asia, and shows how ancient DNA can shed light on the timing and nature of recent positive selection.

Methods

DNA was extracted from the La Braña 1 tooth specimen with a previously published protocol[2]. Indexed libraries were built from the ancient extract and sequenced on the Illumina HiSeq platform. Reads generated were mapped with BWA[27] to the human reference genome (NCBI 37, hg19) after primer trimming. A metagenomic analysis and taxonomic identification was generated with the remaining reads using BLAST 2.2.27+ and MEGAN4 (ref. 28) (Extended Data Fig. 9). SNP calling was undertaken using a specific bioinformatic pipeline designed to account for ancient DNA errors. Specifically, the quality of misincorporations likely caused by ancient DNA damage was rescaled using the mapDamage2.0 software[29], and a set of variants with a minimum read depth of 4 was produced with GATK[30]. Analyses including PCA[10], Outgroup f_3[16] and D statistics[17] were performed to determine the population affinities of this Mesolithic individual (Supplementary Information).

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Contributions

C.L.-F. and E.W. conceived and lead the project. M.E.P. and J.M.V.E. provided anthropological and archaeological information. O.R. and M.E.A. performed the ancient extractions and library construction, respectively. I.O., M.E.A., F.S.-Q., J.P.-M., S.R., O.R., M.F.-C. and T.M.-B. performed mapping, SNP calling, mtDNA assembly, contamination estimates and different genomic analyses on the ancient genome. I.O., F.S.-Q., G.S., C.W.K.C., M.D., J.A.R., J.Q., O.R., U.M.M. and A.N. performed functional, ancestry and population genetic analyses. R.N. and J.N. coordinated the ancestry analyses. M.G.N., R.A.S. and P.S. coordinated the immunological, pigmentation and selection analyses, respectively. I.O., M.E.A., T.M.-B., E.W. and C.L.-F. wrote the majority of the manuscript with critical input from R.N., M.G.N., J.N., R.A.S., P.S. and A.N.

Competing financial interests

The authors declare no competing financial interests.

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NB: Extended data tables and figures are not included in this text. They are available on-line.