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A genomic history of Aboriginal Australia

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1 **The genomic history of Australia**

2 **The human population history of Australia remains contentious, not least because of a lack of**
3 **large extensive genomic data. We generated high-coverage genomes for 83 geographically diverse**
4 **Aboriginal Australians (all speakers of Pama-Nyungan languages) and 25 Papuans from the New**
5 **Guinea Highlands. We find that Papuan and Aboriginal Australian ancestors diversified from**
6 **each other 25-40 thousand years ago (kya), suggesting early population structure in the ancient**
7 **continent of Sahul (Australia, New Guinea and Tasmania). However, all contemporary**
8 **Aboriginal Australian studied descend from a single founding population that differentiated**
9 **around 10-32 kya. We find evidence for a population expansion in northeast Australia during the**
10 **Holocene (past c.10 kya) associated with limited gene flow from this region to the rest of**
11 **Australia. This is broadly consistent with the spread of the Pama-Nyungan languages and**
12 **cultural changes taking place across the continent in the mid-Holocene. We find evidence for a**
13 **single out of Africa dispersal for all contemporary humans and estimate that Aboriginal**
14 **Australians and Papuans shared a common ancestor with other Eurasians 60-100 kya, with**
15 **subsequent admixture with different archaic populations. Finally, we report evidence of selection**
16 **in Aboriginal Australians potentially associated with living in the desert.**

17 During most of the last 100 ky, Australia, Tasmania and New Guinea formed a single continent, Sahul,
18 which was separated from Sunda (the continental landmass including mainland and western island
19 Southeast Asia) by a series of deep oceanic troughs never exposed by changes in sea level (the
20 Wallacean region as defined by biogeographers). Colonisation of Sahul is thought to have required at
21 least 8-10 separate sea crossings between islands¹, potentially constraining the occupation of Australia
22 and New Guinea by earlier hominins². The age of the first occupation of Australia has been disputed.
23 There are several archaeological sites in Australia dating to 40-45 kya (Figure 1), long argued to
24 represent the age of first occupation³ despite a few sites dating to ≥ 50 kya. However, recent studies
25 support the earlier dates, suggesting that Sahul was first settled by 47.5-55 kya⁴⁻⁶. This is consistent
26 with the earliest evidence for modern humans in Sunda at a similar time⁷ (Figure 1). Moreover skeletal
27 remains that share morphological similarities with the ancestors of Aboriginal Australians and Papuans
28 are found in South East Asia up until about 3,5 kya⁸, suggesting that the ancestors of Aboriginal
29 Australians and Papuans extended from Sahul to Sunda.

30 Historically, the morphological diversity among Aboriginal Australians was interpreted by some as
31 indicating multiple ancestral migrations⁹⁻¹¹, or descent from Javanese *Homo erectus*, with varying levels
32 of gene flow from contemporaneous populations¹². However, statistical analyses indicate that
33 Australian crania show no evidence of *H. erectus* admixture¹³. Still, the distinctiveness of the
34 Australian archaeological record has led to the suggestion that the ancestors of Aboriginal Australians
35 and Papuans (hereafter referred to as Australo-Papuans), as well as a small number of other
36 populations, left the African continent earlier than the ancestors of present-day Eurasians¹⁴. Although
37 such multiple dispersals from Africa are supported by some genetic studies^{15,16}, others have found
38 support for only one out of Africa (OoA) event, with one¹⁷ or two¹⁸ independent founding waves into
39 Asia, of which the earlier contributed to Australo-Papuan ancestry^{19,20}. Recent genomic results have
40 also shown that both Aboriginal Australian²⁰ and Papuan²¹ ancestors likely admixed with Neanderthal
41 and Denisovan archaic hominins after leaving Africa.

42 Once in Sahul, contact among groups would have been affected by rising sea-levels that separated the
43 Australian continent from New Guinea and Tasmania 7-14.5 kya through the formation of the Arafura
44 Sea and Bass Strait^{22,23} (Figure 1). These events still appear to be part of the oral tradition of several
45 Aboriginal Australian communities²⁴. Similarly, environmental variation accentuated during the last
46 glacial maximum (LGM) 19-26.5 kya, leading to greater desertification of Australia²⁵ and more
47 challenging temperature gradients, appears to have had an impact on the number and density of human
48 populations^{26,27}. In the same context, morphological and physiological studies find that Aboriginal
49 Australians living in the desert areas today have unique adaptations²⁸⁻³⁰, such as the absence of the
50 increased metabolic rates observed in Europeans when exposed to the freezing night temperatures
51 common in the desert^{31,32}.

52 At the time of European contact, Aboriginal Australians spoke over 250 distinct languages³³, two-thirds
53 of which belong to the Pama-Nyungan family. The place of origin of this language family, which
54 covers 90% of the Australian mainland, has been debated³⁴, as has the effect of its extensive diffusion
55 on its internal phylogenetic structure³³. The pronounced similarity among Pama-Nyungan languages,
56 together with shared socio-cultural patterns, have been interpreted as the result of a recent, mid-
57 Holocene, expansion³⁵. Other changes in the mid-late Holocene (~4 kya) include the efflorescence of
58 backed blades (microliths³⁶) and the introduction of the dingo³⁷. The spatial distribution of microliths

59 roughly correlates with the Pama-Nyungan languages. It has even been suggested that Pama-Nyungan
60 languages, dingoes and backed blades all reflect a recent migration into Australia³⁸. Although an
61 external origin for backed blades has been rejected³⁶, dingoes were certainly introduced, most likely via
62 island south-east Asia³⁷. Rock art traditions also suggest contact between Sulawesi (Indonesia) and
63 Australia³⁸. Intriguingly, a recent genetic study found evidence of Indian gene flow into Australia at the
64 approximate time of these Holocene changes³⁹. Finally, substantial contact with Asians and Europeans
65 is well documented in historical times⁴⁰⁻⁴³, suggesting potentially complex admixture among present-
66 day Aboriginal Australians.

67 After a century of research, the origins and evolutionary history of Aboriginal Australians continue to
68 be debated. To date, only three whole genome sequences have been described - one deriving from a
69 historical tuft of hair from Western Desert Australia²⁰ and two others from cell lines with limited
70 provenance information⁴⁴. In this study we report the first extensive investigation of Aboriginal
71 Australian genomic diversity by reporting and analysing the high-coverage genomes of 83 Pama-
72 Nyungan-speaking Aboriginal Australians and 25 Highland Papuans.

73 **Dataset**

74 We collected saliva samples for DNA sequencing in collaboration with Aboriginal Australian
75 communities and individuals in Australia (S01). We sequenced high-depth genomes (average depth of
76 60X, range 20X-100X) from 83 Aboriginal Australian individuals representing a wide geographical
77 distribution and a broad range of Pama-Nyungan languages (Figure 1, Extended Data Table 1, S02,
78 S03, S04). Additionally, we sequenced 25 Highland Papuan genomes (38X-53X; S03) from five
79 linguistic groups, and generated genotype data for 45 additional Papuans living or originating in the
80 highlands (Figure 1). These datasets were combined with previously published genomes and SNP-chip
81 genotype data, including Aboriginal Australian data from Arnhem Land and from a human diversity
82 cell line panel from the European Collection of Cell Cultures⁴⁴ (ECCAC, Figure 1, S04).

83 We explored the extent of admixture in the Aboriginal Australian autosomal gene pool by estimating
84 ancestry proportions with an approach based on sparse nonnegative matrix factorization (sNMF)⁴⁵. We
85 found that the genomic diversity of Aboriginal Australian populations is best modelled by a mixture of
86 four main different genetic ancestries that can be assigned to four geographic regions based on their

87 relative frequencies: Europe, East Asia, New Guinea and Australia (Figure 2, Extended Data Figure 1,
88 S05). The degree of admixture varies among groups (S05) with the Ngaanyatjarra speakers from
89 central Australia (WCD) having a significantly higher “Aboriginal Australian component” (median
90 value = 0.95) in their genomes compared to the median value of other Aboriginal Australian groups
91 (median value = 0.64; Mann-Whitney rank sum test, one tail p-value = 3.55e-07). The “East Asian”
92 and “Papuan” components are mostly present in northeastern Aboriginal Australian populations (Figure
93 2b, Extended Data Figure 1, S05), while the “European component” is widely distributed across
94 groups. In most of the subsequent analyses, we either selected specific samples or groups according to
95 their level of Aboriginal Australian ancestry, or masked the data for the non-Aboriginal Australian
96 ancestry genomic component (S06).

97 **Colonisation of Sahul and diversification of Australians and Papuans**

98 The origins of Aboriginal Australians is a source of much debate, as are the nature of the relationships
99 among Aboriginal Australians and between Aboriginal Australians and Papuans. Using f_3 statistics,
100 estimates of genomic ancestry proportions and classical multi-dimensional scaling (MDS) analyses, we
101 find that Aboriginal Australians and Papuans are closer to each other than to any other present-day
102 worldwide population included in our study (Figure 2a, Figure 3a, S05). This is consistent with
103 Aboriginal Australians and Papuans being derived from a common ancestral population, which initially
104 colonised Sahul. Moreover, comparing outgroup f_3 statistics we do not find significant differences
105 between Papuan populations (highland Papuan groups and HGDP-Papuans) in their genetic affinities to
106 Aboriginal Australians (Figure 3b), suggesting that the Papuan groups share a common ancestor after
107 or at the same time as the divergence between Aboriginal Australians and Papuans.

108 To investigate the number of founding waves into Australia, we contrasted alternative models of
109 settlement history through a composite likelihood method that compares the observed joint Site
110 Frequency Spectrum (SFS) to that predicted under specific demographic models^{46,47}(Figure 4a, S07).
111 We compared the HGDP-Papuans to four Aboriginal Australian populations with low levels of
112 European admixture (Extended Data Figure 1) from both northeastern (CAI and WPA) and
113 southwestern (WON and WCD) Australia. We compared one and two-wave models where each
114 Australian region was either colonized independently, or by descendants of a single Australian
115 founding population after its divergence from Papuans. The one-wave model resulted in a better fit to

116 the observed SFS, suggesting that the ancestors of the sampled Aboriginal Australians diverged from a
117 single ancestral population. This scenario is also supported by MDS analyses, even when masking
118 Eurasian tracts, as well as by estimation of ancestry proportion analyses where all Aboriginal
119 Australians form a cluster distinct from the Papuan populations (Figure 2, S05). Additionally, it is
120 supported by f_3 analyses where all Aboriginal Australians are largely equidistant from Papuans when
121 adjusting for recent admixture (Figure 3c). Thus, our results based on 83 Pama-Nyungan speakers, do
122 not support earlier claims of multiple ancestral migrations into Australia giving rise to contemporary
123 Aboriginal Australian diversity⁹⁻¹¹.

124 The SFS analysis suggests that there was a bottleneck in the ancestral Australo-Papuan population ~50
125 kya (95% CI 35-54 kya, S07), which overlaps with archaeological evidence for the earliest occupation
126 of both Sunda and Sahul, between 47.5-55 kya^{4,5,48}. We further infer that the ancestors of Pama-
127 Nyungan speakers and Highland Papuans diverged ~37 kya (95% CI 25-40 kya, Figure 4a, S07), which
128 is in close agreement with results of an MSMC analysis (Figure 4b, S08), a method estimating cross
129 coalescence rates between pairs of populations based on individuals' haplotypes⁴⁹. It is also in
130 agreement with previous estimates based on SNP array data³⁹ and the distribution of *Helicobacter*
131 *pylori* strains⁵⁰. These results imply that the divergence between sampled Papuans and Aboriginal
132 Australians is older than the disappearance of the land bridge between New Guinea and Australia about
133 8 kya, and suggest ancient genetic structure in Sahul. Such structure may be related to palaeo-
134 environmental changes leading up to the onset of the LGM. Sedimentary studies show that the vast
135 Lake Carpentaria (500 x 250 km, Figure 1) began to form ~40 kya, when sea-levels fell below the 53m-
136 deep Arafura Sill⁵¹. Therefore, although Australia and New Guinea remained connected until the early
137 Holocene, the flooding of the Carpentaria basin and its increasing salinity⁵¹ may have promoted
138 population isolation.

139 **Archaic admixture**

140 We characterised the number, timing and intensity of archaic gene flow events using three
141 complementary approaches: SFS-based (Figure 4a, Figure 5c, S07), a goodness-of-fit analysis
142 combining D-statistics (S09), and a method that directly infers putatively derived archaic 'haplotypes'
143 (S11). Aboriginal Australians and Papuan genomes show an excess of putative Denisovan-derived

144 variants (Extended Data Figure 2d, S10), as well as substantially more putative Denisovan-derived
145 haplotypes (PDH) than other non-Africans (Extended Data Figure 3). The number and total length of
146 those putative haplotypes varied considerably across samples. However, the estimated number of PDH
147 correlates almost perfectly ($r^2 = 0.96$) with the estimated proportion of Australo-Papuan ancestry in
148 each individual (Extended Data Figure 3). We also estimated that the values of F_{ST} between autosomal
149 SNPs or PDHs assigned to WCD and Papuans were both around 0.12. Moreover, we found no
150 significant difference in the distribution of the number of PDHs or the average length of PDHs between
151 putatively unadmixed Australians and Papuans (Mann-Whitney U test, $p > 0.05$). Taken together, these
152 observations provide strong evidence for a single Denisovan admixture event that predates the
153 population split between Australians and Papuans (see also⁵²) and widespread recent Eurasian
154 admixture in Aboriginal Australians (Figure 2, S05). Furthermore, using the SFS-based approach and
155 constraining Denisovan admixture to have occurred before the Aboriginal Australian-Papuan
156 divergence results in an admixture estimate of ~4% (95% CI 3-5%, Figure 5c, S07), similar to the
157 estimates using D-statistics (~5%, S09). The SFS analyses further suggest that Denisovan/Australo-
158 Papuan admixture took place ~44 kya (95% CI 31-50 kya, S07). We note that the point estimate for the
159 age of the bottleneck overlaps with the confidence interval for the age of admixture, and that a
160 bottleneck could have occurred anywhere along the dispersal route of Australo-Papuan populations
161 from the ancestral source.

162 The SFS analysis also provides evidence for a primary Neanderthal admixture event (~2%, 95% CI 1-
163 3%, Figure 5c, S07) taking place in the ancestral population of all non-Africans ~60 kya (95% CI 55-
164 84 kya, Figure 5c, S07). Note that, although we cannot estimate absolute dates of archaic admixture
165 from the lengths of PDHs and putative Neanderthal-derived haplotypes (PNHs), we can obtain a
166 relative date. We found that for 20 putatively unadmixed Australians and 12 putatively unadmixed
167 HGDP-Papuans, the average PNH length is 33.8 Kb and the average PDH length is 37.4 Kb. These are
168 significantly different from each other ($p = 9.65 * 10^{-6}$ using a conservative sign test), and suggest that
169 the time since Neanderthal admixture was roughly 11% greater than the time since Denisovan
170 admixture roughly in line with our SFS based estimates for Denisovan pulse (31-50 kya) versus the
171 primary pulse of Neanderthal admixture (55-84 kya). The SFS analysis also suggests that the main
172 Neanderthal pulse was followed by a further 1% (95% CI: 0.2-2.7%, Figure 5c, S07) pulse of

173 Neanderthal gene flow into the ancestors of Eurasians, and a smaller pulse into the ancestors of Asians
174 (0.2%, 95% CI 0.1-1.0%, Figure 5c, S07), but there is little evidence for Neanderthal introgression
175 private to Australo-Papuans, potentially limited to ~0.2% (95% CI 0.05-1.3%, Figure 5c, S07). In
176 addition, the fact that the number of Neanderthal-specific introgressed sites increases from Europe to
177 Australia (Extended Data Figure 2d, S10), and then decreases in Amerindians is consistent with
178 recurrent Neanderthal (or Neanderthal-related archaic) gene flow during the waves of expansion into
179 Eurasia. Our results are thus indicative of several pulses of Neanderthal gene flow into modern
180 humans, as inferred previously⁵³⁻⁵⁵. Note however, the apparent high levels in Neanderthal-specific
181 introgressed sites in Australo-Papuans can be explained by the expected number of misclassified
182 Neanderthal introgressed sites resulting from the shared ancestry of these two archaic hominins (S10).
183 Finally, using our SFS and haplotype based approaches, we explored additional models involving
184 complex structure among the archaic populations. We found suggestive evidence that the archaic
185 contribution could be more complex than a model involving discrete Denisovan and Neanderthal
186 admixture pulses^{20,21} (S07, S11), supporting the view that the archaic contribution in Australo-Papuans
187 is likely more complex than was previously assumed^{20,21} (S07).

188 **Out of Africa**

189 To investigate the relationship of Australo-Papuan ancestors to other world populations, we computed
190 D-statistics^{56,57} of the form ((H1=Aboriginal Australian,H2=Eurasian), H3=African) and
191 ((H1=Aboriginal Australian,H2=Eurasian), H3=Ust'-Ishim). Several of these were significantly
192 positive (S09), suggesting that Africans and Ust'-Ishim – a ~45 kya modern human from Asia⁵⁸ - are
193 both closer to Eurasians than to Aboriginal Australians. These findings are in agreement with a model
194 of Eurasians and Australo-Papuan ancestors dispersing from Africa in two independent waves.
195 However, when correcting for a moderate amount of Denisovan admixture, Aboriginal Australians and
196 Eurasians become equally close to Ust'-Ishim, as expected in a single OoA scenario (S09). Similarly,
197 the D-statistics for ((H1=Aboriginal Australian, H2=Eurasian), H3=African) becomes much smaller
198 after correcting for Denisovan admixture. Additionally, a goodness-of-fit approach combining D-
199 statistics across worldwide populations indicates stronger support for two waves OoA, but when taking
200 Denisovan admixture into account, a one-wave scenario fits the observed D-statistics equally well
201 (Figure 5a, S09).

202 To further investigate the timing and number of OoA events giving rise to present-day Australo-Papuan
203 and Eurasians (Sardinians and Han Chinese) we used the observed SFS in a model based composite
204 likelihood framework. When considering only modern human genomes, we find evidence for two
205 waves OoA, with a dispersal of Australo-Papuans ~14 ky before Eurasians (S07). However, when
206 explicitly taking into account archaic Neanderthal and Denisovan introgression into modern
207 humans^{44,59}, the SFS analysis supports a single origin for the OoA populations marked by a bottleneck
208 ~72 kya (95% CI 60-104 kya, S07). This scenario is reinforced by the observation that the ancestors of
209 Australo-Papuan and Eurasians share a Neanderthal admixture event (95% CI 1.1-3.5%). Our analyses
210 suggest that this single OoA ancestral population underwent two expansions at approximately the same
211 time: one involving the ancestors of Australo-Papuan (51-72 kya) and the other, possibly slightly more
212 recent, involving the ancestors of Eurasians (48-68 kya) (Figure 5c). Furthermore, modern humans have
213 both an LD decay rate and a number of predicted deleterious homozygous mutations (recessive genetic
214 load) that correlates with distance from Africa (S05, S10, and Extended Data Figure 2 a-c), again
215 consistent with a single African origin. Aboriginal Australians also show levels of recessive load and
216 LD that are intermediate between East Asians and Amerindians as expected if they all derive from the
217 same OoA dispersal event.

218 The model estimated from the SFS analysis also suggests an early divergence of Australo-Papuans
219 from the ancestors of all non-Africans, in agreement with two colonisation waves across Asia^{20,21,39}.
220 Under our best model, Australo-Papuans began to diverge from Eurasians ~58 kya (95% CI 51-72 kya,
221 Figure 5c, S07), whereas Europeans and East Asians diverged from each other ~42 kya (95% CI 29-55
222 kya, Figure 5c, S07) in agreement with previous estimates^{19,39,60,61}. We find evidence for high levels of
223 gene flow between the ancestors of Eurasians and Australo-Papuans, suggesting that, after the
224 fragmentation of the OoA population (“Ghost” in Figure 5c) 57-58 kya, the groups remained in close
225 geographical proximity for some time before Australo-Papuan ancestors dispersed eastwards.
226 Furthermore, our results show multiple gene flow events between sub-Saharan Africans and Western
227 Eurasians after ~42 kya. This supports previous findings of extensive contact between African and non-
228 African populations⁶⁰⁻⁶².

229 Our MSMC analyses suggest that the Yoruba/Australo-Papuans and the Yoruba/Eurasians cross-
230 coalescence rates are distinct, implying that the Yoruba and Eurasian gene trees across the genome

231 have on average a more recent common ancestor (Figure 5b, S08). We show through simulations that
232 these differences cannot be explained by archaic admixture. Moreover, the expected difference in
233 phasing quality is not sufficient to fully explain this pattern either (see S08). While a similar separation
234 in cross coalescence rate curves is obtained when comparing Eurasians or Australo-Papuans with
235 Dinka, we find that, when comparing the Australo-Papuans or the Eurasians with the San, the cross
236 coalescence curves are overlapping (S08). We also find that the change in effective population size
237 through time of Aboriginal Australians, Papuans, and East Asians is very similar until around 50 kya,
238 including a deep bottleneck around 60 kya (Extended Data Figure 7). Taken together, these MSMC
239 results suggest complex population structure in Africa preceding a split of a single non-African
240 ancestral population, combined with gene flow between the ancestors of Yoruba or Dinka (but not San)
241 and the ancestors of Eurasians, which is not shared with Australo-Papuans. These results are
242 qualitatively in line with the SFS-based analyses (see e.g., Figure 5b).

243

244 **Genetic structure of Aboriginal Australians**

245 Uniparental haplogroup diversity in this dataset (Extended Data Table 1, S12) is consistent with
246 previous studies of mitochondrial DNA (mtDNA) and Y chromosome variation in Australia and
247 Oceania, including the presence of typically European, Southeast and East Asian lineages⁶³⁻⁶⁸. The
248 combined results provide important insights into the social structure of Aboriginal Australian societies.
249 Aboriginal Australian groups exhibit greater between-group variation for mtDNA (16.8%) than for the
250 Y chromosome (11.3%), in contrast to the pattern for most human populations^{69,70}. This result suggests
251 higher levels of male than female migration between Aboriginal Australian groups and may reflect the
252 complex marriage and post-marital residence patterns among Pama-Nyungan Australian groups⁷¹.
253 Moreover, the inferred European ancestry for the Y chromosome is much greater than that for mtDNA
254 (31.8% vs. 2.4%), reflecting male-biased European gene flow into Aboriginal Australian groups during
255 the colonial era.

256 Based on the genome sequences, we find genetic relationships within Australia that mirror geography,
257 with a significant correlation ($r_{\text{GEN,GEO}} = 0.59$, p-value < 0.0005) when comparing the first two
258 dimensions in an MDS analysis (S14). This correlation is higher when genomic regions of putative

259 recent European and East Asian (i.e., Han Chinese) origin are “masked” ($r_{\text{GEN,GEO}} = 0.77$, p-value <
260 0.0005, Extended Data Figure 5). The main axis of genetic differentiation in the masked Aboriginal
261 Australian genomes was determined using the Bearing correlogram approach. We found that an axis of
262 angle = 65° compared to the equator (i.e., in the southwest to northeast direction) explains most of the
263 genetic differentiation (S14).

264 Populations from the centre of the continent occupy positions genetically intermediate to this axis
265 (Extended Data Figure 5). A similar result is observed with an F_{ST} -based tree for the masked data
266 (Figure 6a, S05) as well as in analyses of genetic affinity based on the f_3 statistic (Figure 3b),
267 suggesting a population division between northeastern and southwestern groups. Such structure is
268 further supported by the SFS analyses showing that populations from southwestern desert and
269 northeastern regions diverged as early as ~31 kya (95% CI 10-32 kya), followed by limited gene flow
270 (estimated $2Nm < 0.01$, 95% CI $2 < Nm < 11.25$). The analysis of the major routes of gene flow within the
271 continent supports the idea that the Australian interior has acted as a barrier to gene flow. Indeed, using
272 a model inspired by principles of electrical engineering where gene flow is represented as a current
273 flowing through the Australian continent and observed F_{ST} values are a measure of connectivity, we
274 find that gene flow occurred preferentially along the coasts of Australia (Extended Data Figure 6, S14).
275 These findings are consistent with a model of expansion followed by population fragmentation when
276 and the extreme aridity in the interior of Australia²⁵ formed barriers to population movements during
277 the LGM²².

278 We used MSMC based on autosomal data and mtDNA Bayesian Skyline Plots⁷²(BSP) to estimate
279 changes in effective population sizes within Australia. The MSMC analyses show evidence of a
280 population expansion starting ~10 kya in the northeast, while both MSMC and BSP suggest a
281 bottleneck in the southwestern desert populations taking place during the past ~10 kya (Extended Data
282 Figure 7 , S08, S12). This is consistent with archaeological evidence for a population expansion
283 associated with significant changes in socio-economic and subsistence strategies in the Holocene^{73,74}.

284 European admixture almost certainly had not occurred before the late 18th century, but earlier East
285 Asian and/or Papuan gene flow into Australia could have taken place. We characterized the mode and
286 tempo of gene flow into Aboriginal Australians using three different approaches (S06, S07, S13). We
287 used approximate Bayesian computation (ABC) to compare the observed mean and variance among

288 Aboriginal Australian individuals in the proportion of European, East Asian and Papuan admixture, to
289 that computed from simulated datasets under various models of gene flow. We estimated the European
290 and East Asian admixture to have occurred on the order of ten generations ago (S13), consistent with
291 historical and ethnographic records. Consistent with this, the local ancestry approach based on RFMix
292 suggests that the European and East Asian admixture is more recent than the Papuan admixture
293 (Extended Data Figure 4a). In addition, both the ABC and SFS analyses suggest that the best fitting
294 model for the Aboriginal Australian-Papuan data is one of continuous but modest gene flow, mostly
295 unidirectional from Papuans to Aboriginal Australians, and geographically restricted to northeast
296 Aboriginal Australians ($2Nm=0.4$, 95% CI 0.0-20.4, Figure 4a, S07).

297 To further investigate Papuan gene flow, we conducted follow-up analyses on the Papuan ancestry
298 tracts obtained from the local ancestry analysis. We inferred local ancestry as the result of admixture
299 between four components: European, East Asian, Papuans and Aboriginal Australian (S06). We chose
300 WCD as the representative of Aboriginal Australian ancestry, because it is the least admixed
301 population among our Australian samples (Figure 2, S05). Papuan tract length distribution show a clear
302 geographic pattern, with “younger tracts” (higher median length and variance) in individuals closer to
303 New Guinea and “older” (lower median length and variance) in individuals closer to WCD (Extended
304 Data Figure 4b); there is a strong correlation of Papuan tract length variance with distance from WCD
305 to other Aboriginal Australian groups ($r=0.64$, $p\text{-value}<0.0001$). The prevalence of short ancestry tracts
306 of Papuan origin, compared to longer tracts of East Asian and European origin, suggests that a large
307 fraction of the Papuan gene flow is much older than that from Europe and Asia, which is consistent
308 with the ABC analysis (S13). We also investigated possible South Asian (Indian related) gene flow into
309 Aboriginal Australian, as reported by a recent study³⁹. However, we found no evidence of a component
310 that can be uniquely assigned to Indian populations in the Aboriginal Australian gene pool using either
311 admixture analyses or f_3 and D-statistics (S05), even when including the original Aboriginal Australian
312 genotype data from Arnhem Land. The different nature and size of the comparative datasets may
313 account for the discrepancy in the results.

314 **Pama-Nyungan languages and genetic structure**

315 To investigate if linguistic relationships reflect genetic relationships among Aboriginal Australian
316 populations, we built a Bayesian phylogenetic tree for the 28 different Pama-Nyungan languages

317 represented in this sample⁷⁵ (Figure 6b, S15). The linguistic and F_{ST} -based genetic trees obtained
318 (Figure 6) share several well-supported partitions. For example, both trees indicate that the northeastern
319 (CAI and WPA), and southwestern groups (ENY, NGA, WCD and WON) each form a cluster, while
320 PIL, BDV and RIV are found between them. A distance matrix between pairs of languages, computed
321 from the language-based tree, is significantly correlated with geographic distances ($r_{GEO,LAN} = 0.83$,
322 Mantel test two-tail p-value on 9,999 permutations = 0.0001). This suggests that differentiation among
323 Pama-Nyungan languages in Australia follows geographic patterns, as observed in other language
324 families elsewhere in the world^{15,76}. Furthermore, we find a correlation between linguistics and genetics
325 ($r_{GEN,LAN} = 0.43$, Mantel test p-value < 0.0005) that remains significant when controlling for geography
326 ($r_{GEN,LAN,GEO} = 0.26$, Mantel test p-value < 0.0005). This is consistent with language differentiation after
327 populations lose (genetic) contact with one another⁷⁷. The correlation between the linguistic and genetic
328 trees is all the more striking given the difference in time scales: the Pama-Nyungan family is generally
329 accepted to have diversified within the last 6 ky⁷⁸, while the genetic estimates are two to five times that
330 age. The linguistic tree thus cannot simply reflect initial population dispersals, but rather reflects a
331 genetic structure that has a complex history, with initial differentiation 10-32 kya, localised population
332 expansions (northeast) and bottlenecks (southwest) ~10 kya, and subsequent limited gene flow from the
333 northeast to the southwest. The latter may be the genetic signature that tracks the divergence of the
334 Pama-Nyungan language family.

335 **Selection in Aboriginal Australians**

336 To identify any selection specific to Aboriginal Australians, we used two different methods based on
337 the identification of SNPs with high allele frequency differences between Aboriginal Australians and
338 other groups, similar to the often used Population-Branch Statistics⁷⁹ (PBS, S16). First, we scanned the
339 Aboriginal Australian genomes for loci with an unusually large change in allele frequencies since the
340 divergence from Papuans, taking recent admixture with Europeans and Asians into account. Among the
341 top ranked genomic regions (Extended Data Table 2), we identified candidate loci that might be related
342 to cold tolerance and dehydration resistance. One peak of high differentiation (the 7th highest peak) is
343 located near the *NETO1* gene, which harbours alleles that have previously been shown to be associated
344 with thyroid hormone levels. Interestingly, it has been suggested that thyroid hormone levels are
345 associated with Aboriginal Australian specific adaptations to desert cold⁸⁰. We investigated this

346 potential thermoregulatory adaptation further by identifying genomic regions showing high
347 differentiation associated with different ecological regions in Australia (S16). The top candidate gene
348 in this scan is *KCNJ2*, encoding a potassium channel protein harbouring alleles associated with
349 thyrotoxic periodic paralysis⁸¹. This disease results from complications related to hyperthyroidism,
350 providing additional support for the thyroid hormone system as a target of desert-related natural
351 selection in Aboriginal Australians⁸⁰.

352 Another locus of interest close to the 8th highest peak of differentiation, *SLC2A12*, is associated with
353 serum urate levels⁸². The pathophysiology of dehydration includes elevated serum urate levels.
354 Therefore, these results are suggestive of a locus that may be involved in tolerance to dehydration in
355 Aboriginal Australians. Although further studies are needed to associate putative selected genetic
356 variants in Aboriginal Australians with specific phenotypic effects, the current selection scan provides
357 candidate genes for such future efforts.

358 **Discussion**

359 Our findings shed light, but also raise new questions, concerning on the population history of
360 Aboriginal Australians. They suggest an early population structure in Sahul likely dating back ~37 kya
361 (25-40 kya), when the ancestors of Highland Papuans and Pama-Nyungan Aboriginal Australians
362 diversified. Intriguingly, despite this, our results also indicate that the population that diverged from
363 Papuans was the ancestor of all the Aboriginal Australian groups sampled in this study; yet,
364 archaeological evidence shows that by 40-45 kya, humans were widespread within Australia (Figure 1).
365 Three non-exclusive demographic scenarios can account for this observation: 1) the Aboriginal
366 Australian ancestral population prior to the divergence from Papuans was widespread, maintaining
367 gene flow across the continent; 2) it was deeply structured, and only one group among the early settlers
368 survived to give rise to Aboriginal Australians; and 3) other groups survived, but the descendants are
369 not represented in our sample. Additional modern genomes, especially from Tasmania and the Non-
370 Pama-Nyungan regions of the Northern Territory and Kimberley (both regions highly distinct
371 linguistically⁸³ and not represented in our sample), as well as ancient genomes pre-dating European
372 contact in Australia and other expansions across South East Asia³⁸, should help resolve these questions
373 in the future.

374 To add to this already complex picture, our estimates of ~44 kya (31-50 kya) for the time of admixture
375 between the Australo-Papuan ancestors and an archaic hominin distantly related to Denisovans are very
376 young. In the absence of paleontological evidence that archaic hominins crossed the Wallace Line,
377 combined with evidence of much lower levels of Denisovan ancestry across East Asia and the
378 Americas^{52,86}, it is likely that the admixture occurred in Southeast Asia or even further to the west,
379 constraining the age when the ancestors of living Australo-Papuan colonised Sahul and/or the actual
380 timing of Denisovan admixture. In this context, it is noteworthy that our SFS based time estimates
381 relies on the use of recently suggested molecular clock (1.25×10^{-8} , see⁸⁴) and generation time for
382 humans (29 years⁸⁵). Should any of these parameters change, our genetic-based time estimates will
383 need revisions too.

384 Interestingly, our results also show that southwestern and northeastern Pama-Nyungan populations
385 diverged 10-32kya. Together with the evidence for selection in genes that may have provided an
386 advantage in extreme desert environments, such as those experienced in Western Desert populations
387 during the LGM, these results point to a long-standing genetic structure among Pama-Nyungan
388 Aboriginal Australians that survived post-glacial demographic changes. In other parts of the world,
389 including South East Asia, Pleistocene demographic patterns were overlaid by post-glacial and
390 Holocene expansions that left both genetic and linguistic regional signatures⁸⁷. In Australia, the
391 archaeological record also shows post-glacial expansions^{73,74}, while the spread of Pama-Nyungan
392 languages across the continent is generally accepted to be mid-to-late Holocene³⁵. Our genetic findings
393 indicate an early Holocene demographic expansion localized to northeast Aboriginal Australians, as
394 well as gene flow spreading from the northeast across the continent. These observations are consistent
395 with a possible origin and spread of the Pama-Nyungan languages from the northeast of Australia to the
396 rest of the continent. Thus, evidence from genetics may add to the linguistic and cultural evidence -
397 such as the spread of large ceremonial gatherings, trade and exchange intensification, broad alliance
398 networks, cross-group male ritual induction, new plant foods, among several others³⁵ – that the
399 dispersal of Pama-Nyungan languages has been driven by both cultural diffusion and demic expansion.

400 **Data access**

401 The whole genome sequence data and SNP array data generated in this study are available upon request
402 from E.W (ewillerslev@snm.ku.dk) and D.M.L. (d.lambert@griffith.edu.au). The Papuan whole

403 genome sequence data generated in this study are also available under managed access through the
404 EGA database (<https://www.ebi.ac.uk/ega>) under study accession number EGAS00001001247.

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- 574 Supplementary Information (see annex)
- 575 S01 Ethical approvals in relation to sampling in Australia
- 576 S02 Ethnography and linguistics for the Aboriginal Australian individuals
- 577 S03 Sample collection, DNA extraction, array genotyping, whole-genome sequencing and processing
- 578 S04 Reference panels, relatedness and runs of homozygosity
- 579 S05 Linkage disequilibrium (LD) and population structure within Australia
- 580 S06 Local ancestry
- 581 S07 Demographic inferences
- 582 S08 MSMC analysis
- 583 S09 D-statistic based tests using sampled reads from sequencing data
- 584 S10 Mutation load analysis

- 585 S11 Archaic gene flow
- 586 S12 Uniparental markers
- 587 S13 ABC analysis to characterize recent European, East Asian and Papuan gene flow
- 588 S14 Spatial analyses
- 589 S15 Computational phylogenetics: Pama-Nyungan languages
- 590 S16 Scan for positive selection
- 591