Blood group system polymorphisms in Papua New-Guinea

Benjamin Tournan [1], Adeline Morez [1], Stéphane Mazières [2], Christopher Kinipi [3], Matthew Leavesley [4], Nicolas Brucato [1], François-Xavier Ricaut [1]

[1] Laboratoire Évolution and Diversité Biologique (EDB UMR5174), Université de Toulouse Midi-Pyrénées, CNRS, IRD, UPS, Toulouse, France ; [2] UMR7268 ADES Anthropologie Bioculturelle Droit Ethique et Sante, Aix-Marseille Université - Campus Timone ; [3] Medical Clinic, University of Papua New Guinea, Port Moresby, Papua New Guinea ; [4] Strand of Anthropology, Sociology and Archaeology, School of Humanities and Social Sciences, University of Papua New Guinea, National Capital District, Papua New Guinea

Context

The settlement of Papua New Guinea (PNG) is the result of multiple waves of migration that have contributed to the high genetic diversity of the country [1].

This diversity is particularly notable in blood systems, which were used by early anthropologists as markers of diversity [2]. Today, blood systems are characterized in detail due to their specific epidemiological interest. This is particularly useful in PNG, which is prone to high rate of malaria.

However, the relationship between blood polymorphisms and malaria in PNG is still understudied [3]. We aim to describe blood group genomic profiles in PNG, their geographic distribution, their link to pathogenic pressure (i.e. malaria) and Archaic inheritance.

Methods

- The genomes of 150 individuals were obtained by high-throughput next-generation sequencing (NGS) from salivary samples [4]. Each individual was assigned to a location based on their father's birthplace and mapped with QGIS [5].
- Genetic typing of blood groups was performed using the RBCeq algorithm and 43 blood group systems were identified [6].
- Comparison with 20th century immunological studies based on 6500 individuals [7-9].
- Comparison between malaria incidence rate [3] and presence/absence of alleles of interest.
- Wilcoxon, Student test and PCA were performed with R [10].

Diversity

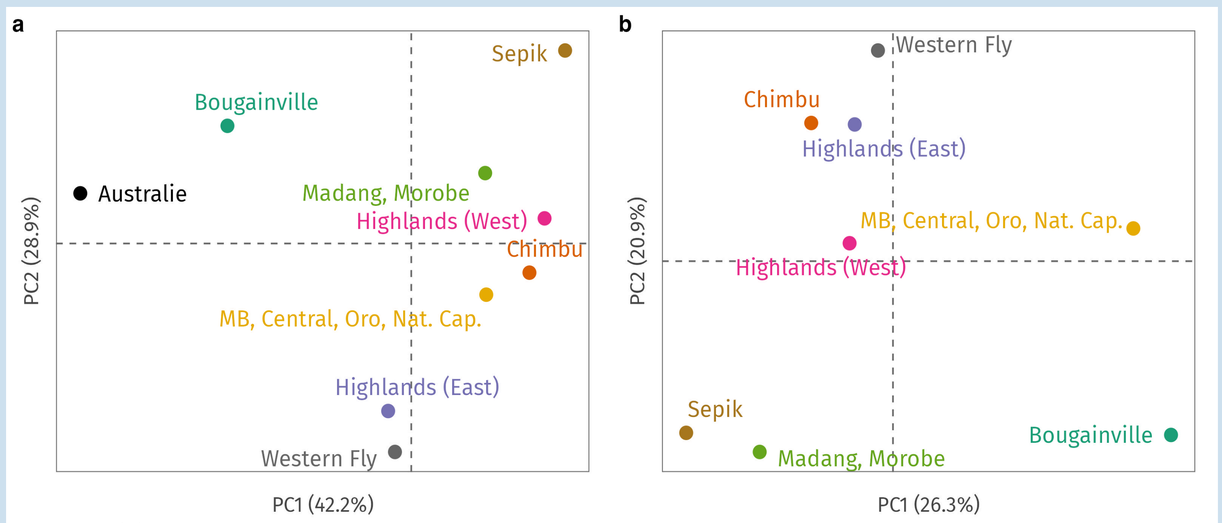

Figure 1 (a) First and second PCA components on genetic frequencies obtained by phenotypic typing in the second half of the 20th century, **(b)** First and second PCA components on allelic frequencies from our database.

- Phenotypic data from the 20th century shows a structuration between Australia and PNG, but no clear structuration within PNG (Fig. 1a).
- Data show geographical structuration between PNG groups (Fig. 1b).

ABO

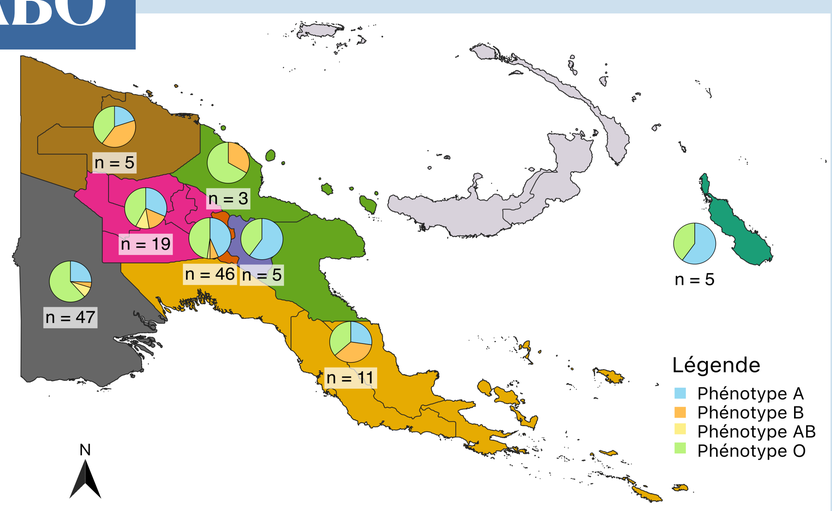

Figure 2 ABO phenotypic frequencies for each geographical area.

ABO*B phenotype is predominant on the northern coast and is probably the result of the Austronesian migration.

This hypothesis is supported by the fact that the B phenotype is mainly present in Asia and nowhere else in the world.

Malaria

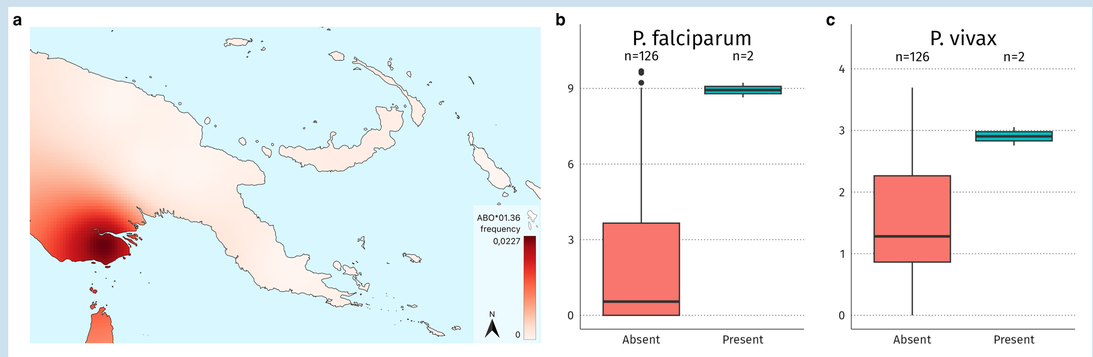

Figure 3 (a) ABO*0.01.36 allelic frequency interpolation map, **(b)** *P. falciparum* incidence rate when ABO*0.01.36 is present/absent for an individual, **(c)** *P. vivax* incidence rate when ABO*0.01.36 is present/absent for an individual.

Rare allele ABO*0.01.36, found in the Western Fly province, is significatively linked to a greater *P. falciparum* ($p<0.05$) and marginally *P. vivax* ($p=0.06$) incidence rate.

Archaic

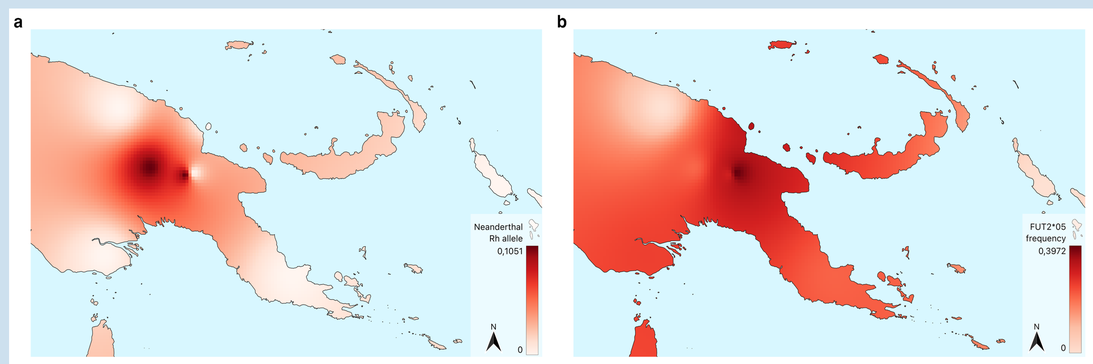

Figure 4 (a) Neanderthal Rh allelic frequency interpolation map, **(b)** FUT2*05, from possible Denisovian introgression, allelic frequency interpolation map.

Neanderthal Rh allele [11] is found only in the highlands (Fig. 4a) and is significatively linked to a lower *P. falciparum* ($p<0.05$) incidence rate. Allele FUT2*05, identified in Denisova, is also found primarily in the highlands (Fig. 4b).

Conclusion

- Current blood systems in PNG are inherited from the populations history and by their interaction with their ecosystem, e.g. pathological pressure.
- New genomic tools allow for a finer definition of blood systems in PNG, completing those obtained since the 20th century.
- Preliminary analyses suggest links between certain blood groups and malaria, some being potentially inherited from Neanderthal/Denisova.

References

[1] Brucato et al. 2018. [2] Heydon and Murphy, 1924. [3] Hay and Snow, 2006. [4] Brucato et al. 2022. [5] QGIS Association, 2023. [6] Jadhao et al. 2022. [7] Cavalli-Sforza et al. 1995. [8] Ivinskis et al. 1956. [9] Simmons et al. 1965. [10] R, 2023. [11] Condemi et al. 2021.

Acknowledgments

We thank the University of Papua New Guinea and the National Research Institute (PNG) for their help and support. This work was also supported by the French National Research Agency (ANR), and the French Embassy in PNG.

More information on https://papuanpast.hypotheses.org/

Contacts: nicolas.brucato@univ-tlse3.fr, matthew.leavesley@gmail.com