

Original Research

Human adaptation in the Andes Mountains

Jessica De Loma ¹, Mário Vicente ^{2,3,4}, Noemi Tirado ⁵, Franz Ascui ⁶, Luis A. Parada ⁷, Jacques Gardon ⁸, Carina Schlebusch ^{2,9,10}, Karin Broberg ^{1,*}

1. Institute of Environmental Medicine, Unit of Metals and Health, Karolinska Institutet, 177 Stockholm, Sweden; Email: jessica.deloma.olson@ki.se
2. Human Evolution, Department of Organismal Biology, Uppsala University, 752 36 Uppsala, Sweden; Emails: mario.vicente@su.se (M.V.); carina.schlebusch@ebc.uu.se (C.S.)
3. Centre for Palaeogenetics, University of Stockholm, 114 18 Stockholm, Sweden
4. Department of Archaeology and Classical Studies, Stockholm University, 106 91 Stockholm, Sweden
5. Genetics Institute, Genotoxicology Unit, Universidad Mayor de San Andrés, La Paz, Bolivia; Email: noemistirado@gmail.com
6. Programa de Salud Familiar Comunitaria e Intercultural (SAFCI), Ministerio de Salud y Deportes Bolivia, La Paz, Bolivia; Email: afranzeduardo@yahoo.com
7. Facultad de Ciencias Exactas and Consejo de Investigación, Universidad Nacional de Salta, A4400 Salta, Argentina; Email: luisantonioparada@gmail.com
8. Hydrosiences Montpellier, Institut de Recherche pour le Développement, CNRS, University of Montpellier, 34093 Montpellier, France; Email: jacques.gardon@ird.fr
9. Palaeo-Research Institute, University of Johannesburg, P.O. Box 524, Auckland Park, 2006, South Africa
10. SciLifeLab Uppsala, 752 37 Uppsala, Sweden

* **Correspondence:** Karin Broberg; Email: karin.broberg@ki.se

Supplementary Materials

Figure S1. Population structure of the three Andean study populations (marked in bold) and comparative populations.

Figure S2. Runs of homozygosity of the Andean study populations and comparative populations.

Figure S3. Genome-wide XP-EHH selection scans for the three Andean study populations.

Figure S4. Genome-wide LSBL selection scans for the three Andean study populations.

Table S1. Putatively selected genes overlapping across the Andean populations according to any selection scan method.

Table S2. Gene symbol mapping by IPA from the list of putatively selected genes shared across the three Andean populations ($n = 116$).

Table S3. List of pathways (IPA; Canonical) enriched in the putatively selected genes across the three Andean populations. Top 50 results.

Table S4. List of pathways (IPA; Disease or Function) enriched in the putatively selected genes across the three Andean populations. Top 50 results.

Table S5. List of pathways (WebGestalt; KEGG) enriched in the putatively selected genes across the three Andean populations. Top 10 results.

Table S6. List of pathways (WebGestalt; PANTHER) enriched in the putatively selected genes across the three Andean populations. Top 10 results.

Table S7. Putatively selected genes overlapping across selection scan methods in the SAC study population.
Table S8. Putatively selected genes overlapping across selection scan methods in the Aymara-Quechua study population.
Table S9. Putatively selected genes overlapping across selection scan methods in the Uru study population.
Table S10. Genes within the top five peaks for potential positive selection in the iHS selection scans.
Table S11. Genes within the top five peaks for potential positive selection in the XP-EHH selection scans.
Table S12. Genes within the top five peaks for potential positive selection in the LSBL selection scans.

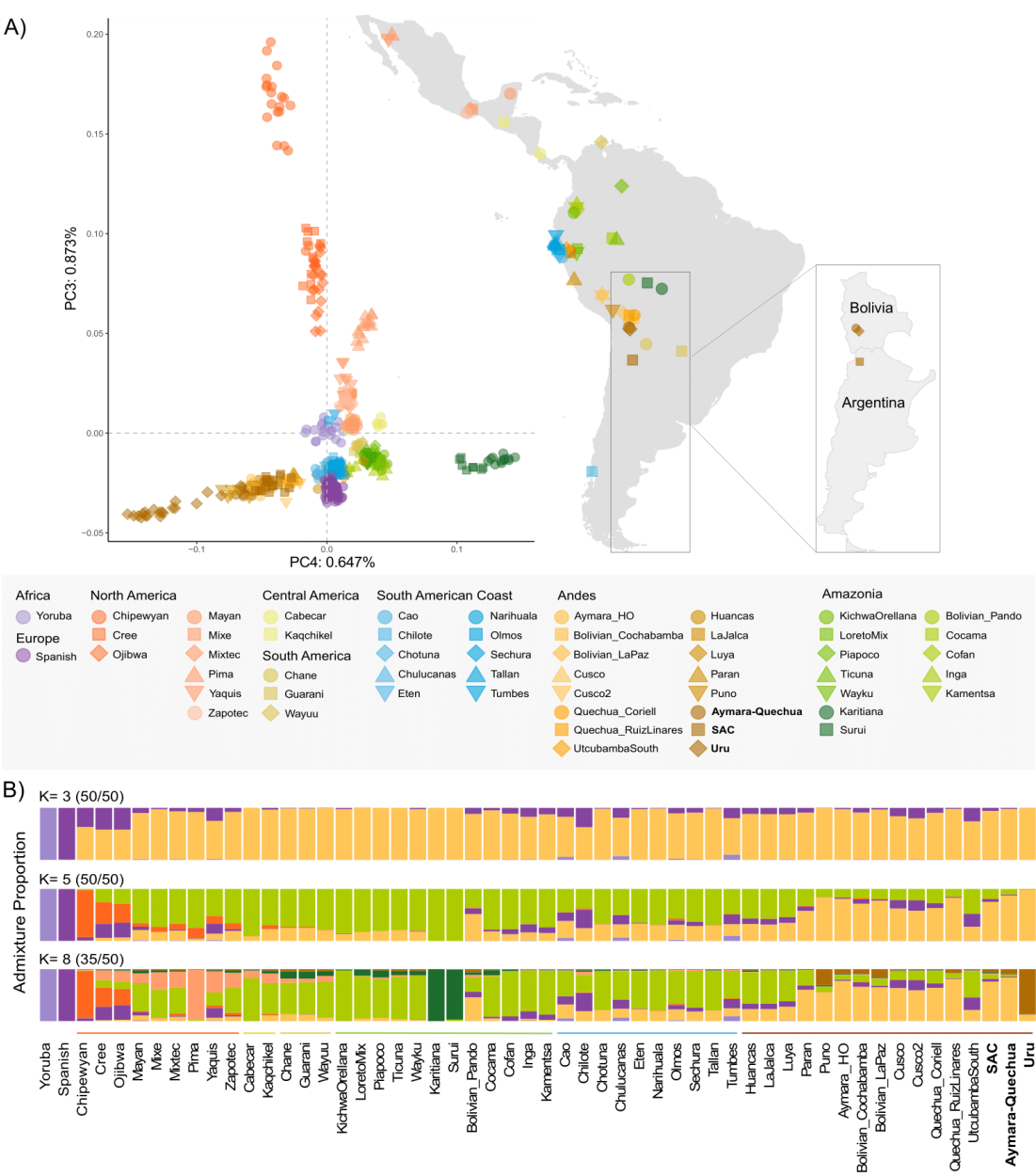


Figure S1. Population structure of the three Andean study populations (marked in bold) and comparative populations. (A) Principal component analysis and approximate geographical locations of the populations. **(B)** Population-averaged cluster analysis for K = 3, 5, and 8. Figure reproduced from De Loma et al. [1] with permission of Elsevier.

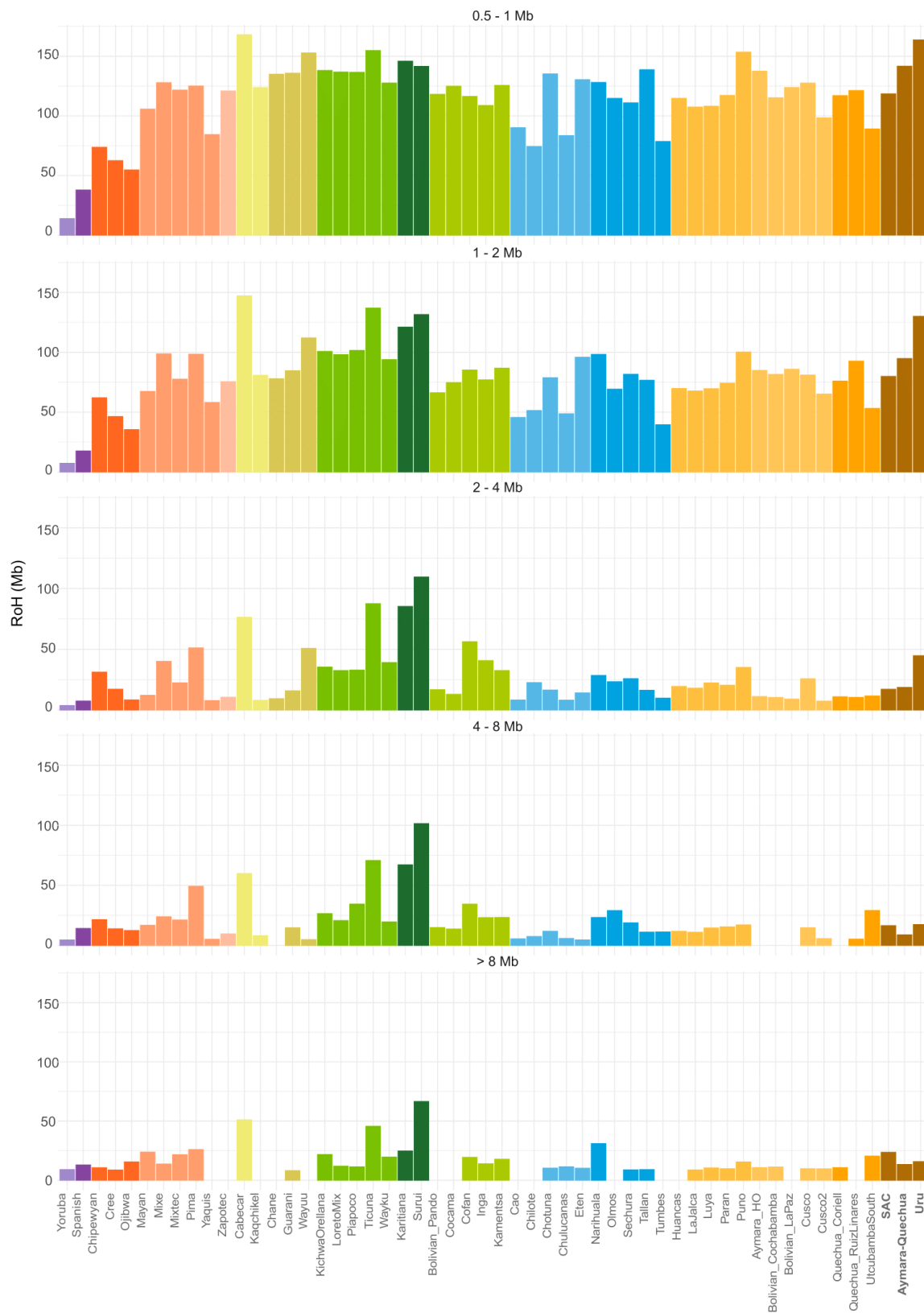


Figure S2. Runs of homozygosity of the Andean study populations and comparative populations.

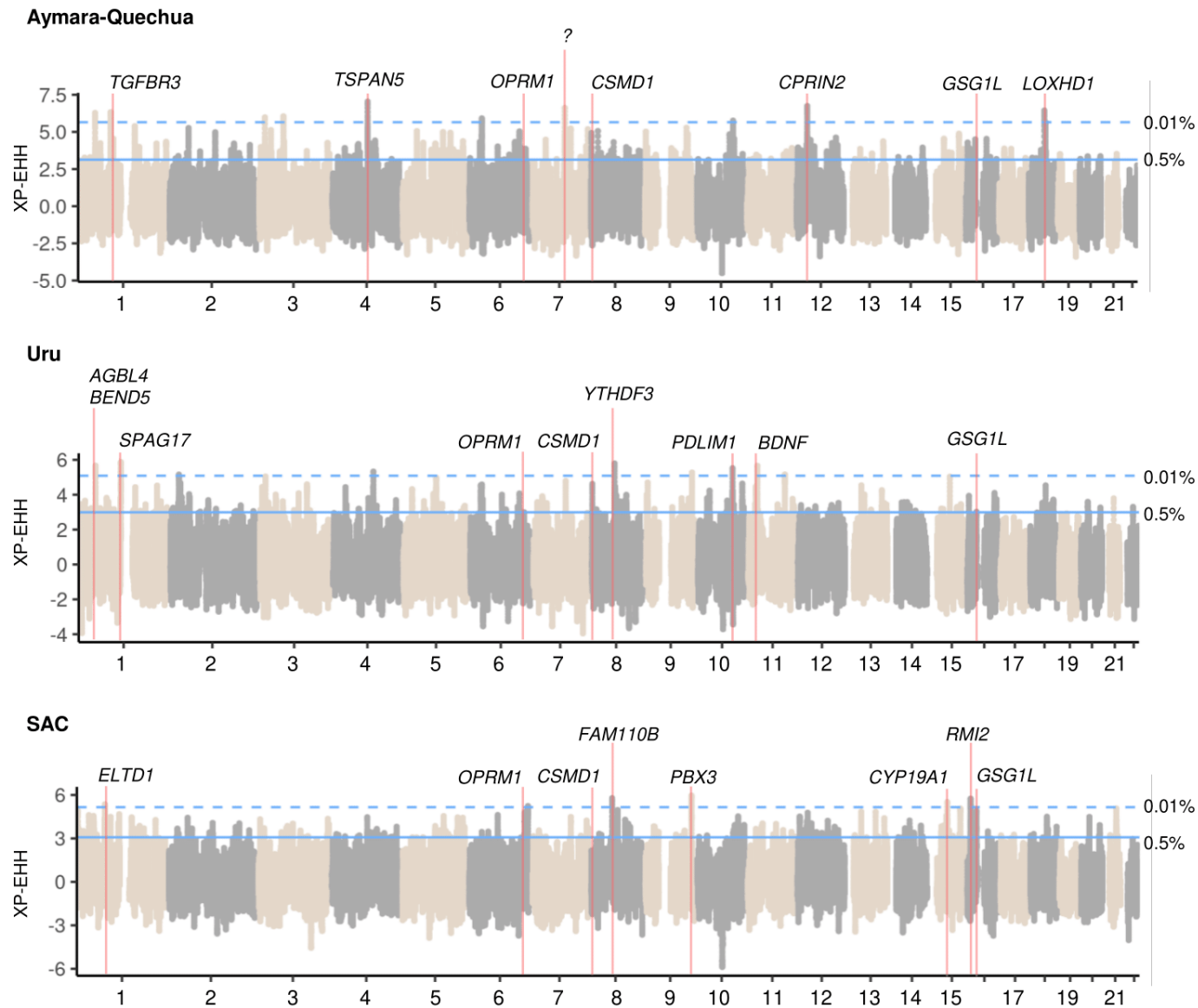


Figure S3. Genome-wide XP-EHH selection scans for the three Andean study populations. To avoid spurious results due to single SNPs, we averaged the XP-EHH values with a sliding window of 10 SNPs. The top five peaks in each population are marked with red lines, as well as the location for *OPRM1*, *CSMD1*, and *GSG1L*. A further description of the genes within these top peaks is included in Table S11. The top 0.5% (solid line) and top 0.01% (dashed line) are shown.

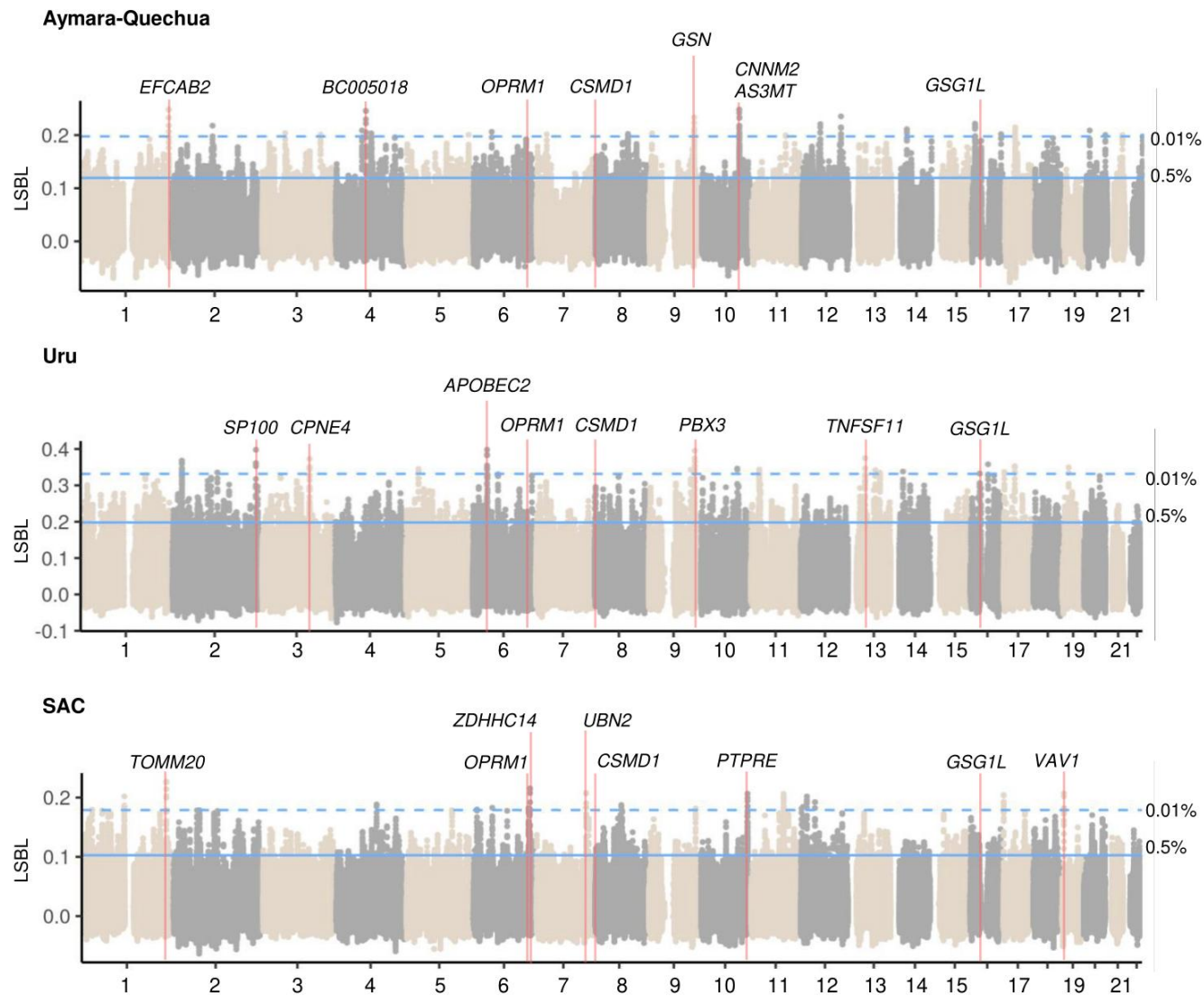


Figure S4. Genome-wide LSBL selection scans for the three Andean study populations. To avoid spurious results due to single SNPs, we averaged the LSBL values with a sliding window of 10 SNPs. The top five peaks in each population are marked with red lines, as well as the location for *OPRM1*, *CSMD1*, and *GSG1L*. A further description of the genes within these top peaks is included in Table S12. The top 0.5% (solid line) and top 0.01% (dashed line) are shown.

References

1. De Loma J, Vicente M, Tirado N, Ascui F, Vahter M, Gardon J, et al. Human adaptation to arsenic in Bolivians living in the Andes. *Chemosphere*. 2022;301:134764.