A Piebald Cladistic Portray of Mitochondrial DNA Control Region Haplogroups in Khyber Pakhtunkhwa, Pakistan

Authors : Shahzad Bhatti, M. Aslamkhan, Sana Abbas, Marcella Attimonelli, Hikmet Hakan Aydin, Erica Martinha Silva de Souza,

Abstract : Despite being situated at the crossroad of Asia, Pakistan has gained crucial importance because of its pivotal role in subsequent migratory events. To highlight the genetic footprints and to contribute an enigmatic picture of the relative population expansion pattern among four major Pashtun tribes in Khyber Pakhtunkhwa viz., Bangash, Khattak, Mahsuds and Orakzai, the complete mitochondrial control region of 100 Pashtun were analyzed. All Pashtun tribes studied here revealed high genetic diversity; that was comparable to the other Central Asian, Southeast Asian and European populations. The configuration of genetic variation and heterogeneity further unveiled through Multidimensional Scaling, Principal Component Analysis, and phylogenetic analysis. The results revealed that the Pashtun is a composite mosaic of West Eurasian ancestry of numerous geographic origin. They received substantial gene flow during different invasions and have a high element of the Western provenance. The most common haplogroups reported in this study are: South Asian haplogroup M (28%) and R (8%); whereas, West Asians haplogroups are present, albeit in high frequencies (67%) and widespread over all; HV (15%), U (17%), H (9%), J (8%), K (8%), W (4%), N (3%) and T (3%). Herein we linked the unexplored genetic connection between Ashkenazi Jews and Pashtun. The presence of specific haplotypes J1b (4%) and K1a1b1a (5%) point to a genetic connection of Jewish conglomeration with Khattak tribe. This was a result of an ancient genetic influx in the early Neolithic period that led to the formation of a diverse genetic substratum in present day Pashtun.

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Keywords : mtDNA haplogroups, control region, Pakistan, KPK, ethnicity

Conference Title : ICSRD 2020 : International Conference on Scientific Research and Development

Conference Location : Chicago, United States

Conference Dates : December 12-13, 2020