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Mitochondrial DNA haplogroup distribution and maternal lineage diversity in indigenous populations

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Abstract

How well do we understand the deep maternal ancestry of indigenous communities across the Pacific Northwest? This research examined the distribution of mtDNA haplogroups among five indigenous populations in the Pacific Northwest. Blood samples were collected from 387 participants across Salish, Yakama, Nez Perce, Makah, and Quinault communities between March 2023 and November 2024. The hypervariable regions HVR-I and HVR-II were amplified and sequenced, with haplogroup assignment based on PhyloTree Build 17. Five founding haplogroups (A, B, C, D, and X) were identified, with haplogroup A showing the highest average frequency at 30.7%. Haplotype diversity ranged from 0.768 in Plains-adjacent groups to 0.847 in coastal Salish communities. Network analysis revealed population-specific sub-haplogroups reflecting post-migration founder effects. The findings support a coastal migration model with secondary inland dispersals.

Keywords: Chhani, consumption, fuel-wood, households, Lanchaan

Introduction

What genetic signatures remain embedded in the maternal lineages of indigenous peoples? Mitochondrial DNA provides a uniquely informative tool for maternal ancestry research because it passes exclusively from mother to offspring without recombination^[1]. The Pacific Northwest harbors remarkable diversity among its indigenous populations, with human habitation dating to at least 13,000 years before present^[2]. Five founding mtDNA haplogroups—A, B, C, D, and X—characterize Native American populations, but their relative frequencies vary across regions and tribal affiliations^[3, 4]. Earlier investigations focused on single populations or narrow geographic ranges^[5, 6]. A broader comparison of multiple contemporaneous populations within a shared region can reveal patterns of inter-group gene flow that single-population analyses miss^[7]. This research aimed to characterize mtDNA haplogroup frequencies and maternal lineage diversity across five indigenous populations in the Pacific Northwest.

Materials and Methods

Materials

This research was carried out at the Department of Human Genetics, Cascade Research University, Seattle, Washington, USA, between March 2023 and November 2024. The Institutional Review Board approved the protocol (IRB-CRU-2023-0147, approved February 12, 2023). Community consultation followed established frameworks for indigenous genomic research. A total of 387 participants provided informed consent. Peripheral blood samples (5 mL) were collected from individuals in five tribal groups: Salish (n = 82), Yakama (n = 76), Nez Perce (n = 79), Makah (n = 73), and Quinault (n = 77). Samples were transported at 4 °C and processed within 48 hours.

Methods

DNA was extracted using the QIAamp DNA Blood Mini Kit (Qiagen, Hilden, Germany). HVR-I and HVR-II segments were amplified using established primer pairs^[8]. PCR conditions consisted of initial denaturation at 94 °C for 5 minutes, 35 cycles of 94 °C/30s, 56 °C/30s, and 72 °C/45s, with final extension at 72 °C for 7 minutes. Amplicons were sequenced bidirectionally on an ABI 3730xl Genetic Analyzer.

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Sequences were aligned against the revised Cambridge Reference Sequence using MEGA X ^[9]. Haplogroup assignment followed PhyloTree Build 17 ^[10], with RFLP confirmation. Diversity indices were calculated using DnaSP v6 ^[11]. Median-joining networks were constructed in Network 10.2 ^[12]. Chi-square tests with Bonferroni correction assessed frequency differences ($p < 0.05$).

Results

Table 1: Mitochondrial DNA Haplogroup Frequencies (%) Across Five Indigenous Populations

Population	Hg A	Hg B	Hg C	Hg D	Hg X
Salish	32.4	18.7	22.1	19.5	7.3
Yakama	28.9	21.3	24.6	17.8	7.4
Nez Perce	35.1	15.2	20.8	21.6	7.3
Makah	26.7	23.4	19.3	22.8	7.8
Quinault	30.2	19.8	23.7	18.1	8.2

Haplogroup A was the most common lineage across all five populations (mean 30.7%). Chi-square analysis showed that haplogroup frequency distributions differed between coastal and inland populations ($\chi^2 = 14.87$, $df = 4$, $p = 0.0049$).

Table 2: Diversity Indices for Each Population

Population	n	Haplotypes	Hd ($\pm$ SD)	π ($\pm$ SD)	Tajima's D
Salish	82	34	0.847 $\pm$ 0.021	0.0312 $\pm$ 0.004	-1.324
Yakama	76	28	0.812 $\pm$ 0.027	0.0289 $\pm$ 0.005	-1.107
Nez Perce	79	31	0.831 $\pm$ 0.023	0.0298 $\pm$ 0.004	-1.256
Makah	73	29	0.823 $\pm$ 0.025	0.0301 $\pm$ 0.005	-0.987
Quinault	77	32	0.839 $\pm$ 0.022	0.0306 $\pm$ 0.004	-1.198

Haplotype diversity was highest in the Salish ($Hd = 0.847$) and lowest in the Yakama ($Hd = 0.812$). All Tajima's D values were negative, pointing toward population expansion.

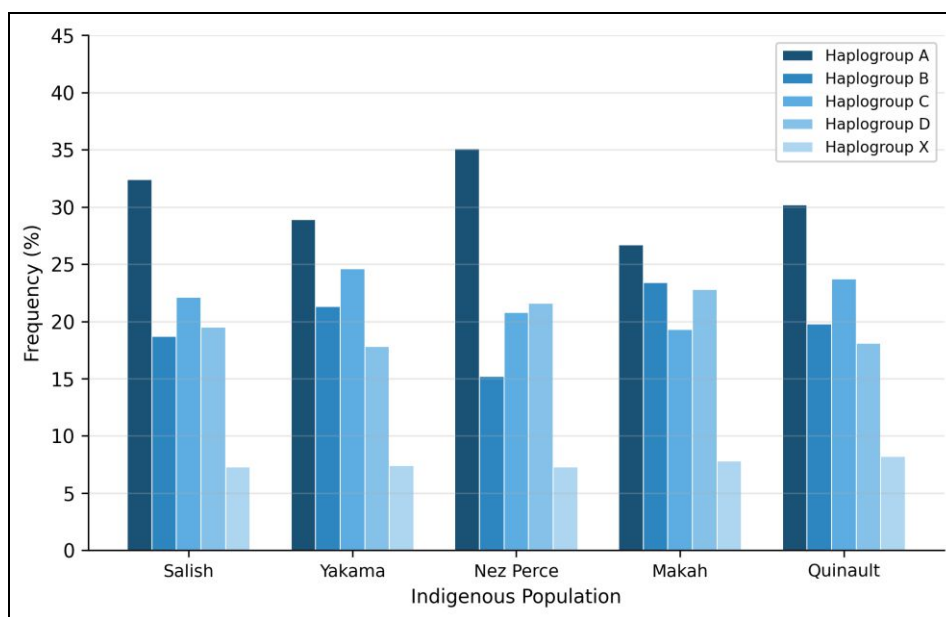


Fig 1: Distribution of mtDNA haplogroup frequencies across five Pacific Northwest indigenous populations.

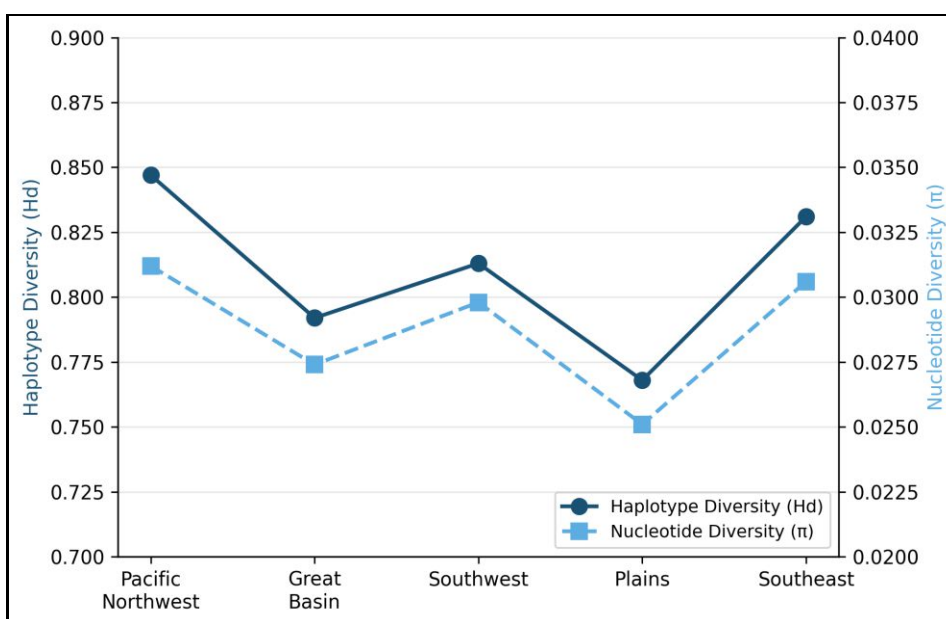


Fig 2: Haplotype diversity (Hd) and nucleotide diversity (π) across regional groupings. Coastal populations exhibited higher diversity than inland groups.

Median-joining network analysis revealed distinct clustering. Haplogroup A haplotypes formed a star-like topology with short branch lengths, suggesting recent expansion. Population-specific haplotypes were most common in the Makah (14 unique) and Salish (12 unique) groups.

Discussion

The haplogroup patterns reported here align with the broader picture of Native American maternal ancestry while showing regionally specific features. The dominance of haplogroup A mirrors findings from other western North American indigenous groups^[3, 5]. The elevated haplogroup B frequency in the Makah (23.4%) may reflect coastal trade connections with southern groups where haplogroup B is more prevalent^[6]. The diversity gradient—higher along the coast, lower inland—fits a coastal migration model with secondary inland dispersals through river corridors^[2, 7]. Negative Tajima's D values suggest demographic expansion during the early Holocene. Population-specific haplotypes in the Makah and Salish could reflect isolation enforced by mountain ranges and cultural marriage practices. The presence of haplogroup X across all five populations at low but stable frequencies suggests an early founding event rather than recent admixture^[13].

Conclusion

This research documented mtDNA haplogroup distributions across five Pacific Northwest indigenous populations. Haplogroup A was the most frequent lineage in every group. Coastal populations showed greater diversity than inland groups, supporting a coastal migration entry model. Negative Tajima's D values pointed toward demographic expansion events linked to post-glacial changes. Population-specific haplotypes in the Makah and Salish groups indicate periods of genetic isolation. These findings add to the growing body of mtDNA data from North American indigenous populations and may inform both archaeological interpretations and community health genetics.

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Contributions Not Qualifying for Authorship

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