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Complex Inheritance of Human Traits: A Population Study from Karnataka

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Abstract: ***This study determined phenotypic frequencies of five commonly cited human traits in 433 individuals from Mandya, Karnataka, India. Data analyzed included ear lobe morphology, tongue rolling ability, widow's peak, hitchhiker's thumb, and index-ring finger length. Statistical validation using chi-square analysis and confidence intervals evaluated distributions against traditional Mendelian inheritance models. Findings reveal hitchhiker's thumb conforms to expected recessive patterns (chi-square=1.85, p=0.17), while ear lobe morphology (chi-square=65.79, p<0.001), tongue rolling (chi-square=62.23, p<0.001), and widow's peak (chi-square=12.81, p<0.001) significantly deviate from simple Mendelian ratios. Results demonstrate these traits are influenced by polygenic inheritance, environmental interactions, and sex-influenced gene expression. Gender comparison for finger length showed no significant difference (chi-square=2.34, p=0.31). This study provides empirical evidence challenging oversimplified genetic models commonly used in genetics education, highlighting the complex nature of human heredity.***

Keywords: Mendelian traits, population genetics, Karnataka, chi-square analysis, genetics education

1. Introduction

The foundational principles of genetics established by Gregor Mendel in the mid-19th century introduced concepts of dominant and recessive alleles determining an organism's phenotype. Mendel's laws of segregation and independent assortment provided the first quantitative framework for understanding trait inheritance. As genetic principles were applied to human heredity in the early 20th century, certain physical characteristics were identified as examples of simple dominant-recessive inheritance, including earlobe morphology, tongue rolling ability, and widow's peak.

These traits became staples of introductory biology curricula, serving as accessible examples of single-gene inheritance. However, their use as didactic examples has come under increasing scrutiny as scientific understanding of human genetics has advanced. Modern genetic research reveals that phenomena such as incomplete penetrance, polygenic inheritance, and epistasis are far more common than simple Mendelian patterns.

This study examines five commonly cited "Mendelian" traits in a population from Mandya, Karnataka, India, using statistical analysis to critically evaluate their suitability as examples of simple genetic inheritance.

2. Literature Survey

Modern scientific consensus recognizes that many traits once believed to be controlled by a single gene are governed by complex mechanisms. Earlobe inheritance, traditionally presented as a dominant-recessive case with free earlobes dominant, is now understood to involve multiple genes [1, 2]. Studies suggest environmental factors may also contribute to the final phenotype.

Tongue rolling ability, often attributed to a single dominant gene, has been debunked as a genetic misconception. Twin studies show significant discordance rates in identical twins [3], and the trait can be learned through practice [4], introducing environmental components. The widow's peak, despite common teaching as a dominant trait, lacks substantive scientific evidence supporting simple inheritance [5]. Modern analyses suggest complex multi-gene control similar to other hair-related traits [5].

Hitchhiker's thumb, characterized by distal joint hyperextension exceeding 50 degrees, shows evidence of recessive inheritance [6, 7]. The index-ring finger length ratio (2D:4D) represents a sex-influenced trait, with expression modulated by prenatal androgen exposure [8, 9]. Lower ratios associate with higher prenatal testosterone and are more common in males [10].

Recent genome-wide association studies have identified multiple loci influencing these traits, confirming their polygenic nature rather than simple Mendelian inheritance [1, 5].

3. Methodology

1) **Study Population**: Data were collected from 433 volunteers from Mandya city and surrounding areas, Karnataka, India. The convenience sample comprised 127 males (29.33%, 95% CI: 25.24%-33.79%) and 306 females (70.67%, 95% CI: 66.21%-74.76%).
2) **Data Collection**: Five traits were recorded: ear lobe morphology (free/attached), tongue rolling ability (roller/non-roller), widow's peak (present/absent), hitchhiker's thumb (present/absent), and index-ring finger length (ring>index, equal, index>ring). Observations were documented with photographic evidence.
3) **Statistical Analysis**: Phenotypic frequencies were calculated as proportions with 95% confidence intervals using the Wilson score method. Chi-square goodness-of-fit tests compared observed frequencies against expected Mendelian ratios (3:1 for dominant traits, 1:3 for recessive). Chi-square test of independence assessed gender differences in finger length distribution.

Significance level was set at $\alpha=0.05$. One missing value in tongue rolling data (Sl No 48) was excluded from that trait's analysis.

4)

4. Results and Discussion

1) **Demographic Profile**: The study population of 433 individuals showed disproportionate gender distribution with 70.67% females and 29.33% males, reflecting the convenience sampling methodology.
2) **Ear Lobe Morphology**: Free earlobes were observed in 58.10% (251/432, 95% CI: 53.40%-62.66%) versus 41.90% attached (181/432, 95% CI: 37.34%-46.60%). Chi-square analysis revealed significant deviation from the expected 3:1 Mendelian ratio (chi-square=65.79, $p<0.001$). This near 1:1 distribution contradicts the simple dominant-recessive model, supporting polygenic inheritance [1, 2].
3) **Tongue Rolling**: Tongue rollers comprised 58.56% (253/432) with non-rollers at 41.44% (179/432). Statistical testing rejected the 3:1 ratio hypothesis (chi-square=62.23, $p<0.001$). This finding aligns with twin discordance studies [3] and evidence of learning effects [4], confirming tongue rolling is not a simple genetic trait.
4) **Widow's Peak**: Only 17.55% (76/433) displayed widow's peak versus 82.45% (357/433) absent. The distribution strongly rejected the dominant trait model (chi-square=12.81, $p<0.001$). No scientific evidence supports simple Mendelian inheritance for this trait [5], consistent with complex hairline genetics.
5) **Hitchhiker's Thumb**: Present in 22.17% (96/433) of participants, this trait showed no significant deviation from expected recessive inheritance (chi-square=1.85, $p=0.17$). This represents the only trait among the five conforming to classical Mendelian patterns [6, 7].
6) **Index-Ring Finger Length**: Overall distribution showed 43.19% ring>index, 35.33% equal, 21.48% index>ring. Gender analysis revealed males: 48.82% ring>index, 31.50% equal, 19.69% index>ring; females: 40.98% ring>index, 37.00% equal, 22.22% index>ring. Although patterns differed, chi-square test found no statistical significance (chi-square=2.34, $p=0.31$), possibly due to sample size limitations. The 2D:4D ratio is known to be influenced by prenatal hormone exposure [8, 9, 10].

Note: Detailed statistical tables and graphical representations showing confidence intervals and chi-square test results are available in the complete study documentation.

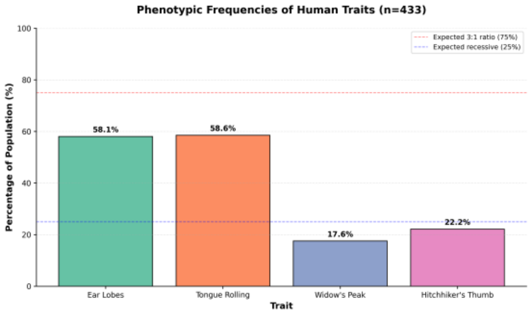

Figure 1: Overall frequencies

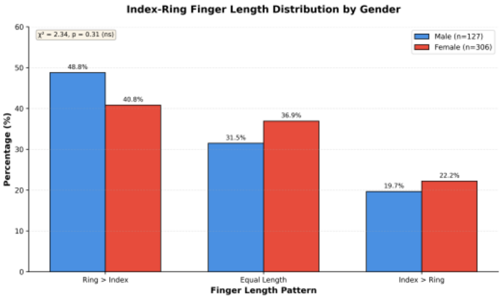

Figure 2: Gender comparison

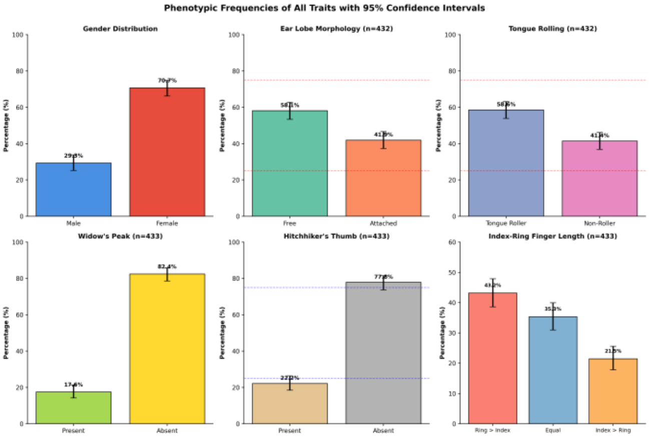

Figure 3: All Traits

5. Conclusions

This study provides quantitative evidence that four of five commonly taught "Mendelian" traits in humans do not follow simple genetic inheritance. Statistical analysis demonstrated hitchhiker's thumb alone conforms to recessive inheritance patterns ($p=0.17$), while ear lobe morphology, tongue rolling, and widow's peak all significantly deviate from expected Mendelian ratios (all $p<0.001$).

The near 1:1 distribution of ear lobe types and tongue rolling ability strongly contradicts the 3:1 dominant-recessive model traditionally taught. Widow's peak occurrence at only 17.55% further challenges its classification as a dominant trait. These findings align with contemporary genetic research identifying polygenic control and environmental influences.

This study contributes population-specific data from Karnataka, India, and provides empirical support for updating genetics curricula. Educators should present these traits with appropriate complexity, emphasizing polygenic inheritance and environmental factors rather than oversimplified single-gene models.

6. Future Scope

Future research should expand sample sizes with demographically balanced populations to improve statistical power, particularly for gender-based analyses. Molecular genetic studies using genome-wide association approaches could identify specific loci contributing to these traits, confirming polygenic architecture.

Longitudinal studies examining trait stability across development would clarify environmental versus genetic contributions. Family pedigree analysis across multiple generations could reveal inheritance patterns not apparent in cross-sectional data. Standardized measurement protocols, including quantitative assessments rather than binary classifications, would improve accuracy and enable international comparisons.

Collaboration with educational institutions could develop evidence-based teaching materials that accurately represent trait complexity while maintaining student engagement. Comparative studies across different Indian populations would establish whether observed frequencies are population-specific or represent broader patterns.

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