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Review Article

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The role of ear morphology in forensic DNA phenotyping: A Comprehensive review

Abstract

Forensic genetics employs molecular methods to aid in the investigation of unresolved cases. Conventional forensic identification relies on comparing DNA profiles from crime-scene samples with reference DNA obtained from potential suspects. Advances in genomic technologies have expanded this capability, enabling the prediction of externally visible characteristics (EVCs) directly from DNA. Traits such as eye, skin, and hair color, as well as specific facial features, can now be inferred using single nucleotide polymorphism (SNP) markers. Recently, ear morphology has emerged as an additional EVC of interest due to its distinctive anatomical structure and relative stability against environmental influences.
The human ear, characterized by its complex and highly individualized morphology, represents a promising biometric feature for forensic applications. Forensic DNA phenotyping (FDP) incorporating ear-related genetic markers may complement traditional DNA analyses by narrowing investigative leads. This review examines the anatomical structure of the ear, the genetic factors underlying its morphological variation, and the potential integration of ear-specific phenotypic prediction into forensic practice. Furthermore, genetic variants associated with ear morphology and their relevance for FDP are summarized.

Keywords: Ear morphology, single nucleotide polymorphisms, forensic DNA phenotyping, genome-wide association studies

INTRODUCTION

Forensic genetics plays a central role in criminal investigations by assisting in the identification of perpetrators, supporting the exoneration of innocent individuals, and reconstructing events at crime scenes. Its importance becomes especially pronounced in cases where no suspects have been identified or when information about the perpetrator is limited (1).

The discipline has advanced substantially through major milestones in genetic research. Foundational discoveries—including the characterization of ABO blood groups, the elucidation of the DNA double-helix structure, and Alec Jeffreys' identification of variable number tandem repeats (VNTRs)—have profoundly shaped modern approaches to human identification (2). Although VNTRs represented a breakthrough in early forensic applications, their reliance on relatively large quantities of intact DNA and the labor-intensive nature of their analysis revealed the need for more sensitive and efficient methodologies (3,4).

A transformative shift occurred in the early 1990s with the widespread adoption of short tandem repeats (STRs) for paternity testing and forensic identification, a development that ultimately established STR profiling as the gold standard in forensic DNA analysis (4,6). However, STR-based methods remain limited when examining highly degraded or low-template samples and, importantly, do not inherently provide phenotypic information about an unknown individual. Recognizing these challenges, the early 2000s witnessed rapid technological advancements that facilitated the incorporation of single nucleotide polymorphisms (SNPs), insertion–deletion polymorphisms (InDels), and mini-STRs into forensic genetic workflows (7,8).

Over the past two decades, NGS technologies have undergone

transformative advancements, enabling high-throughput, rapid, and precise forensic genetic analyses. NGS platforms facilitate the analysis of minimal DNA quantities while providing highly accurate and reproducible results (8). Current research increasingly focuses on leveraging NGS to predict complex phenotypic traits—such as age, skin pigmentation, and facial morphology—thereby expanding the scope of traditional genetic identification (8).

Within this evolving landscape, forensic DNA phenotyping (FDP) has emerged as a promising subdiscipline. FDP aims to infer an individual's externally visible characteristics (EVCs), including eye, hair, and skin color; age; facial shape; ear morphology; baldness; and biogeographical ancestry, directly from DNA. In cases where the suspect's identity is unknown, FDP can provide investigative leads by offering predictive information about an individual's physical appearance (9,10). While robust and validated panels exist for predicting eye, hair, and skin color, ongoing research is extending these capabilities to more complex phenotypic traits, including detailed ear morphology and specific facial structures (10,11).

Ear morphology, characterized by its highly individualized and structurally complex features, has gained increasing attention for identification purposes across multiple scientific disciplines, particularly within forensic science. Unlike other externally visible traits, the external ear maintains a relatively stable form throughout life and is minimally affected by environmental factors, rendering its morphology largely determined by genetic influences. Recent advancements now integrate SNP-based genetic analyses with sophisticated three-dimensional (3D) facial modeling and machine learning algorithms to predict various dimensions of ear shape (12).

This review provides a comprehensive examination of the historical and contemporary developments in ear morphology prediction, spanning traditional visual and anthropometric techniques as well as emerging genomic approaches. It highlights significant methodological advancements and discusses their implications for enhancing forensic investigative capabilities.

The Role of Ear Morphology Across Different Disciplines

Although the primary function of the human ear is auditory perception, its externally visible structure exhibits highly distinctive morphological variability, making it a valuable feature in forensic and biometric applications. The considerable inter-individual differences in auricular shape and dimensions can be systematically assessed and used for personal identification. The stability, accessibility, and inherent uniqueness of ear morphology underpin its utility across multiple disciplines, including forensic casework, biometric verification, and surveillance technologies (14).

Ear shape and measurements also serve as essential anatomical landmarks in facial reconstruction. Notably, earprints — recognized for their individuality and comparability in uniqueness to fingerprints—can be recovered at crime scenes and used as potential biometric evidence for suspect identification (13). Moreover, ear morphology provides a critical alternative identifying traits in situations where facial recognition is limited, such as in profile-view imagery or when individuals are partially masked in security footage. As a result, ear-based features are increasingly incorporated as complementary components to facial recognition in both two-dimensional (2D) and three-dimensional (3D) biometric systems (14). The continued application of metric and morphological analyses of the ear reinforces its significance across multiple disciplines by enhancing accuracy, reliability, and interpretive value. Ear morphology has demonstrated utility in forensic identification, biometric security, genetic research, clinical diagnostics, and evolutionary studies.

- Forensic anthropology: Morphometric analysis of auricular structures can assist in estimating demographic attributes such as age and sex, particularly when examining severely compromised human remains, including those from fire scenes or fragmented bodies (15).
- Biometric security and law enforcement: Ear morphology extracted from surveillance imagery provides a robust biometric identifier, especially when facial features are partially or fully obscured. The auricle's relatively stable shape—less affected by hairstyle, eyewear, or facial expressions—makes it highly suitable for recognition algorithms (14). Notably, auricular evidence obtained from video recordings has been accepted in court proceedings in countries such as the Netherlands, the United Kingdom, Germany, Austria, and the United States (16).
- Genetic research: Ear morphology serves as a valuable phenotype for examining the relationship between genetic architecture and environmental influences on human variation. Recent advances in genome-wide association studies (GWAS) have identified numerous candidate genes linked to ear shape, size, and structural variation, thereby contributing to a more comprehensive understanding of auricular development (17).
- Clinical genetics: Distinctive ear anomalies function as important diagnostic markers for various congenital syndromes. Characteristic auricular features often serve as early clinical indicators in conditions such as Down syndrome, Treacher Collins syndrome, and CHARGE syndrome (18,19).
- Human evolutionary biology: Population-level differences in ear morphology provide critical insights into evolutionary forces, including adaptation, selection, and genetic drift, contributing to broader reconstructions of human evolutionary history (20).
- Facial reconstruction: Ear morphology provides essential anatomical reference points for reconstructing missing or damaged facial structures, thereby supporting visual identification in forensic and archaeological contexts (21).

Human Ear Anatomy

Embryologically, development of the external ear begins within the first six weeks of gestation and progresses rapidly throughout

early fetal growth, achieving its definitive morphological form by approximately the 18th gestational week (22). Ear morphology displays extensive phenotypic variability among individuals and, due to its exposed position on the face, serves as a highly discriminative marker for personal identification. The emergence of inter-individual variation reflects a multifactorial interplay of genetic, epigenetic, demographic, and evolutionary determinants (23).

Anatomically, the human ear is a complex organ consisting of three major regions: the outer ear, middle ear, and inner ear (Figure 1A). The auricle, or pinna, represents the externally visible portion of the outer ear and is composed primarily of elastic cartilage. Its principal function is to capture and channel sound waves toward the auditory canal. The auricle comprises several distinctive substructures that exhibit substantial morphological diversity.

The concha forms the deep, concave central depression of the auricle and serves as a primary acoustic collector. The helix, the prominent outer rim, displays considerable inter-individual variation in curvature, thickness, and continuity. Parallel to it lies the antihelix, an inner cartilaginous fold that contributes to the ear's characteristic contours. These features—along with their numerous measurable dimensions and shape variations—constitute critical parameters for precise morphological assessment and biometric or forensic analysis (24).

The middle ear is an air-filled cavity that houses the tympanic membrane and three auditory ossicles: the malleus, incus, and stapes. The tympanic membrane converts incoming sound waves into mechanical vibrations, which are transmitted through the ossicular chain—beginning with the malleus, which is directly attached to the membrane, followed by the incus, and ultimately the stapes. The stapes interface with the oval window of the inner ear, where these mechanical forces are conveyed to the fluid-filled cochlea. The Eustachian tube, which links the middle ear to the nasopharynx, is essential for maintaining pressure equilibrium across the tympanic membrane and ensuring optimal auditory function.

The inner ear, situated deep within the petrous portion of the temporal bone, mediates both hearing and balance. It comprises the cochlea, a spiral, fluid-filled structure that converts mechanical energy into neural impulses transmitted via the auditory nerve, and the vestibular apparatus, which includes the semicircular canals and otolithic organs and serves as the primary sensory system for balance and spatial orientation (25,26).

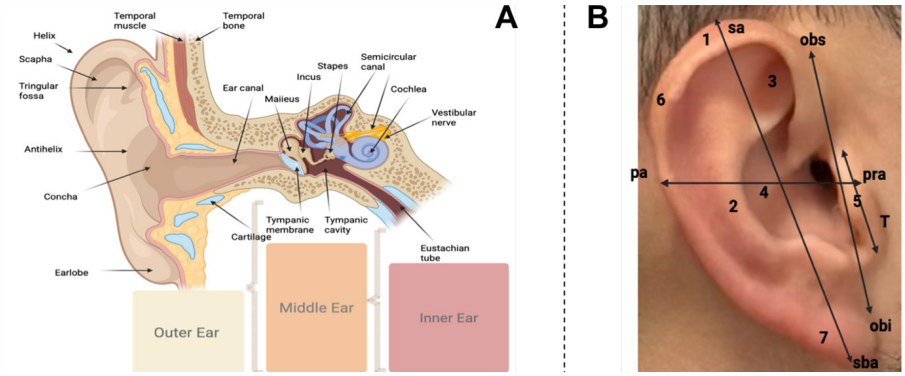

Figure 1. Ear Anatomy and Key Morphological Landmarks. (A) Anatomical structure of the human external ear (created with BioRender). (B) Principal auricular regions and landmark points commonly used in ear morphology and phenotyping studies (created with BioRender). 1. Helix – the prominent outer rim of the auricle. 2. Antihelix – the curved elevation of cartilage situated parallel and medial to the helix. 3. Superior crus of the antihelix – the upper branch of the antihelix, extending toward the superior portion of the auricle. 4. Inferior crus of the antihelix – the lower branch of the antihelix, forming part of the boundary of the conchal bowl. 5. Tragus – a small cartilaginous projection anterior to the external auditory meatus. 6. Darwin's tubercle – a minor nodular thickening located on the posterior–superior portion of the helix, present variably among individuals. 7. Earlobe (lobule) – the soft, pendulous, non-cartilaginous inferior portion of the auricle.

Phenotypic Analysis of Ear Morphology

Ear morphology encompasses a range of genetically influenced phenotypic traits, including variations in ear shape, size, and specific anatomical features that contribute to individual distinctiveness. These traits often display notable variation across different populations, rendering ear phenotypes valuable in contemporary forensic science, anthropology, and genetic research. In forensic investigations, earprints recovered from crime scenes serve as highly informative biometric markers due to the uniquely individualized nature of auricular structure. In anthropological studies, comparative analyses of ear morphology among diverse populations provide insights into human variation and population history. Within genetic research, identifying the genes and regulatory mechanisms that shape ear structure is essential for understanding the complex hereditary factors underlying auricular development (27).

Among the various traits contributing to overall ear morphology, the structure of the earlobe (lobule) represents one of the most visually distinctive phenotypic features. Located at the inferior aspect of the auricle, the lobule is a soft, non-cartilaginous structure that typically appears in three principal phenotypic forms: free, partially attached, and attached (26). A free earlobe—characterized by its lower margin hanging independently from the cheek—is traditionally considered

the dominant genetic trait. In contrast, an attached earlobe, in which the lower margin is continuous with the facial skin, is less prevalent. These phenotypic distinctions can aid the identification of disaster victims, support facial reconstruction in cases involving severe trauma or thermal injury, and provide supplementary forensic information in contexts such as drowning investigations (28).

Phenotypic characterization of ear morphology relies on accurate anthropometric measurements obtained from standardized anatomical reference points. As illustrated in Figure 1B, these landmarks are essential for quantitative assessment and include:

- **Otobasion superius (Obs):** The superior point at which the helix attaches to the temporal region, marking the upper boundary of auricular–facial junction.
- **Otobasion inferius (Obi):** The inferior point of attachment of the lobule to the cheek, defining the lower limit of the auricular–facial junction.
- **Superaurale (Sa):** The most superior point of the auricle.
- **Subaurale (Sba):** The most inferior point of the auricle.
- **Postaurale (Pa):** The most posterior projecting point along the curvature of the auricle.
- **Preaurale (Pra):** The most anterior point of the auricle, located near the level where the helix attaches to the temporal region (29).

Ear Morphology in Identification

Due to its highly individual-specific anatomical configuration, ear morphology has long been recognized as a valuable biometric feature for human identification. As early as the 18th century, the Swiss physiognomist Johann Caspar Lavater (1741–1801) proposed that the human ear possessed uniquely distinguishing characteristics suitable for identifying individuals (30). By the late 19th century, French forensic scientist Alphonse Bertillon incorporated ear morphology into his anthropometric identification system, systematically analyzing auricular contours, lobular shape, folds, and overall configuration. With the emergence of photographic documentation, Bertillon further standardized the recording of suspect images, emphasizing the ear as a reliable distinguishing feature (31). Later, Edmond Locard expanded on this concept through his foundational principle —"every contact leaves a trace"—highlighting the evidentiary potential of earprints, which, like fingerprints, may be deposited at crime scenes and used in forensic identification (32).

Contemporary approaches to utilizing ear morphology for identification rely primarily on three established techniques: visual comparison, anthropometric measurement (originating from the Bertillonage system), and photogrammetry (33). Visual comparison involves detailed examination of earprints or ear images, emphasizing overall auricular configuration, characteristic helical and antihelical folds, distinct depressions and protrusions, lobular form, and other individualized anatomical features. Anthropometric measurement provides a quantitative assessment by determining dimensions such as ear length, width, and lobular size through standardized linear distances between defined anatomical landmarks (Figure 1B) (34). Photogrammetry further expands forensic capability by enabling high-resolution digital imaging and three-dimensional reconstruction of earprints or ear images, thereby allowing for precise morphological analysis and improved reliability in forensic identification (35).

Sex-related variation in auricular dimensions has also been well documented, and several ear measurement parameters exhibit statistically significant differences between males and females. In a study of 191 healthy Turkish adults (106 females and 85 males) aged 18–25 years, males demonstrated significantly greater ear length, ear width, earlobe length, and earlobe width compared with females (22).

The forensic relevance of the auricle has been substantiated through both earprint analysis and image-based identification, with pioneering research conducted in Germany, the Netherlands, the United Kingdom, and the United States. In numerous cases, earprints collected from crime scenes—particularly those involving theft and homicide—have facilitated suspect identification through comparative analysis, notably in Germany, Italy, the Netherlands, and the United Kingdom (36). Beyond earprint evidence, the distinctive and relatively stable morphology of the auricle has proven valuable for image-based forensic identification, especially from surveillance footage. Unlike many facial features, the ear is less susceptible to occlusion by hair, glasses, or facial hair, making it a comparatively robust biometric marker. Courts in several countries, including Austria, Germany, the United Kingdom, and the United States, have accepted auricle-based video evidence, and studies report correct identification rates of up to 65% when using surveillance images (37,38).

Genetic Determination of Ear Phenotypes

The development of ear morphology is governed by a complex interplay of multiple genetic factors. These genes collectively shape individual-specific auricular characteristics, including the contours of the helix, antihelix, tragus, and antitragus, as well as earlobe configuration (e.g., attached or free) and the overall architecture of the external ear. Population-level variation in these traits reflects underlying genetic diversity, reinforcing the polygenic basis of auricular morphology (9). In this context, elucidating the genetic determinants of ear phenotypes is critical for advancing forensic identification. Improved understanding of the molecular and developmental pathways involved in ear morphogenesis not only enhances the precision of biometric recognition systems but also strengthens the scientific rigor and reliability of forensic analyses.

Table 1. Gene regions and SNP markers associated with ear morphology

Chromosome Region	rs number	Gene Region	Affect Region	Reference
1:118849460	rs868157	TF (transcription factor) binding site	Tragus size, antitragus size	Noreen et al., 2023 (40)
1:119333107	rs17023457	TBX15 (T-box transcription factor)	Antitragus size, antihelix folding	Adhikari et al., 2015 (39) Noreen et al.,2023 (40)
2:106457045	rs62152530	RGPD3 (Ran-binding protein–related gene)	Ear size	Wu et al.,2019 (47)
2:108897145	rs3827760	EDAR (Ectodysplasin A receptor)	helix, earlobe size, earlobe attachment	Adhikari et al., 2015 (39) Noreen et al.,2023 (40)
2:108927596	rs13397666	EDAR	Earlobe size, helix, Darwin tubercule	Noreen et al., 2023 (40)
2:108936422	rs13427222	EDAR	Earlobe size, earlobe attachment, antitragus size, antihelix superior crus	Noreen et al., 2023 (40)
2:108982800	rs260674	EDAR	Antihelix folding, antihelix superior crus	Noreen et al., 2023 (40)
2:109142712	rs7567615	SH3RF3 (SH3 domain-containing ring finger 3)	Helix, antihelix superior crus	Noreen et al., 2023 (40)
2:15166294	rs1619249	LINC01630 (Long Intergenic Non-Protein Coding RNA 1630)	Antihelix folding	Adhikari et al., 2015 (39)
2:170677711	rs10192049	MYO3B (Myosin IIIB)	Antihelix superior crus	Noreen et al., 2023 (40)
2:170684313	rs2080401	ENSG00000213981	Earlobe size, earlobe attachment	Adhikari et al., 2015 (39) Noreen et al.,2023 (40)
2: 85318367 3	rs7428	TGOLN2 (Trans-Golgi Network Integral Membrane Protein 2)	Tragus size	Noreen et al., 2023 (40)
3:139278846	rs9866054	FOXL2 (Forkhead Box L2)	Earlobe size	Noreen et al., 2023 (40)
3:139286078	rs10212419	MRPS22 (Mitochondrial Ribosomal Protein S22)	Earlobe size, antihelix superior crus	Adhikari et al., 2015 (39)
4:150301114	rs1960918	LRBA (LPS-Responsive Beige-Like Anchor Protein)	Helix	Adhikari et al., 2015 (39)
6:142586378	rs263156	GPR126 (G protein–coupled receptor)	Earlobe attachment, earlobe size.	Adhikari et al., 2015 (39)
7:51831003	rs684523	FLJ20021	Tragus size, helix	Noreen et al., 2023 (40)
8:121878655	rs7812632	HAS2-AS1 (HAS2 Antisense RNA 1)	Darwin tubercule	Li et al., 2023 (41)
9:31394277	rs7873690	SLC4A1 (Solute Carrier Family 4 Member 1)	Antitragus size, tragus size, earlobe attachment	Noreen et al., 2023 (46)
10:109875454	rs3818285	XPNPEP1 (X-Prolyl Aminopeptidase 1)	Antihelix	Noreen et al., 2023 (40)
11:47656077	rs1878495	SULT1C2P1 (Sulfotransferase Family 1C pseudogene)	Antihelix superior crus	Noreen et al., 2023 (40)

The relationship of these features with ear helix shape, earlobe type, preauricular tubercle (Darwin's tubercle)—which are the main phenotypic characteristics used in the determination of ear phenotype—and their associated gene regions is discussed in detail below

Genetic investigations into ear morphology have increasingly relied on GWAS and analyses of publicly available datasets within the National Center for Biotechnology Information (NCBI). These studies have systematically examined SNPs that contribute to phenotypic variation in ear structure. Collectively, this body of research confirms that ear morphology is a polygenic trait shaped by the coordinated effects of numerous genes rather than a single genetic locus (31). Several SNPs have been repeatedly identified as significantly associated with specific auricular features. For example, the rs3827760 SNP has been linked to variations in auricular folding, earlobe size, and the attached versus free earlobe phenotype. Similarly, rs263156 has shown associations with earlobe size and attachment type. The rs17023457 locus has been implicated in determining antitragus size, while rs2080401 has been associated with both earlobe size and the attached/free earlobe characteristic (39,40). Table 1 provides a summary of these gene regions and their corresponding SNP markers. The following subsections discuss the genetic determinants of the most prominent ear phenotypic traits, including the auricle, the earlobe, and Darwin's tubercle.

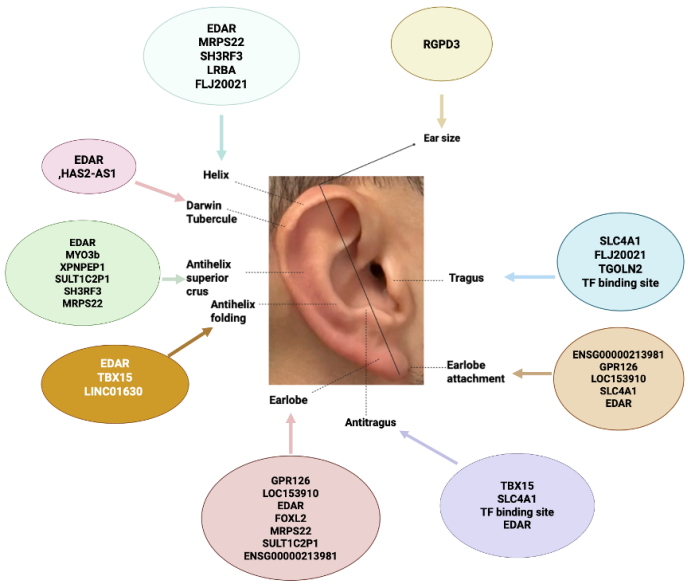

Figure 2. Genes associated with ear morphology (created using BioRender).

Auricle (External Ear)

The human auricle (pinna) is an anatomically complex structure composed of elastic cartilage covered by skin (Figure 1B). This cartilaginous framework is anchored to the temporal bone by intrinsic and extrinsic auricular muscles as well as fibrous connective tissue. The auricle displays substantial morphological variation among individuals, shaped by a combination of age, sex, population ancestry, and—most importantly—genetic factors. These genetic determinants are central to understanding individual variation and hold significant relevance for forensic identification and biometric applications (15).

Several genes have been implicated in the embryological development and final morphological configuration of the auricle. The coordinated interactions among these genes contribute to the diverse range of ear shapes and sizes observed in human populations (13). Key genes involved in auricular morphogenesis are summarized below.

• HOXA2 (Homeobox A2):

HOXA2 is a pivotal transcription factor required for proper auricular development. During embryogenesis, it regulates the differentiation of cranial neural crest cells that give rise to the structural components of the external ear. Pathogenic variants in HOXA2 are strongly associated with severe congenital anomalies, most notably microtia, characterized by a hypoplastic or malformed auricle (42, 43).

• EDAR (Ectodysplasin A Receptor):

EDAR plays a central role in the development of ectodermal derivatives, including hair, teeth, and the external ear. Numerous studies have demonstrated robust associations between EDAR variants and distinct ear morphological traits such as earlobe configuration and ear protrusion. Polymorphisms in EDAR contribute to characteristic ear features particularly prevalent in East Asian populations (5,41).

• PAX1:

PAX1 functions as an essential regulatory gene in craniofacial and auricular development. Evidence from murine models indicates that loss-of-function mutations in PAX1 result in Otofaciocervical Syndrome (OFCS), a disorder marked by craniofacial dysmorphism and pronounced external ear abnormalities (44).

• SH3RF3 (SH3 Domain–Containing Ring Finger 3):

SH3RF3 participates in intracellular signaling pathways and has been statistically associated with variation in external ear morphology. Although its precise developmental role remains to be fully elucidated, current findings suggest its involvement in the broader genetic network that modulates ear shape.

• TBX15 (T-Box Transcription Factor 15):

TBX15 is a transcription factor crucial for craniofacial patterning during embryogenesis. Variants in TBX15 have been linked to a spectrum of auricular anomalies and are implicated in shaping overall facial morphology (45,46).

• PRRX1 (Paired Related Homeobox 1):

PRRX1 is involved in the differentiation of craniofacial mesenchyme and contributes to defining the shape of both the auricle and the surrounding facial structures. Mutations in PRRX1 can give rise to marked auricular malformations (5).

• GSC (Goosecoid):

Although not exclusively specific to auricular shape, GSC influences the developmental pathways of the external ear and auditory canal. Variants, particularly in exons 2 and 3, have been associated with microtia-like phenotypes and are implicated in SAMS syndrome, characterized by the absence of the external auditory canal (47).

Pioneering GWAS on auricular morphology began with the work of Adhikari et al. (2015), which analyzed facial photographs from 5,062 individuals from diverse Latin American populations. This study identified significant associations between auricle shape variation and genetic loci in SH3RF3, EDAR, and TBX15, highlighting their key roles in external ear morphology (39).

Subsequently, a GWAS conducted by Shaffer et al. in individuals of European ancestry reported significant associations between auricular dimensions and variants within BARX1 and PAX9 — both transcription factors essential for craniofacial patterning and morphogenesis (5).

Further insights were provided by the Latin American Diversity and Evolution Analysis (CANDELA) consortium, which examined facial morphology in 6,282 volunteers across Brazil, Chile, Colombia, Mexico, and Peru. This large-scale analysis

identified SNPs within the WARS2/TBX15 genomic region as strongly associated with helix morphology, reinforcing the functional relevance of this locus in auricular development (48).

Earlobe

Earlobe configuration—commonly categorized as attached or free—was historically regarded as a classic example of Mendelian inheritance. However, recent GWAS have demonstrated that the genetic architecture of this trait is substantially more complex and best explained by a polygenic model. Early investigations into its inheritance began in the 1920s. Carrière, examining 15 families, proposed that the attached earlobe phenotype was inherited as a dominant trait, whereas Hilden reported conflicting findings, sparking one of the earliest scientific debates on the mode of inheritance for this feature (20). A major conceptual shift occurred with the work of Wiener et al. (1937), who suggested that earlobe morphology was likely influenced by multiple genes or possibly by a single gene with more than two alleles (49).

Modern GWAS have further clarified the genetic determinants of earlobe morphology. Variants within TBX15 and EDAR have been repeatedly identified as significantly associated with earlobe configuration. Polymorphisms in these genes influence whether the earlobe presents as attached or free, underscoring their central role in the broader genetic network shaping external ear morphology (5).

The SP5 gene also plays a pivotal role in the development of external ear structures, particularly during critical phases of embryogenesis. Its interaction with the Wnt signaling pathway is central to the regulation of craniofacial patterning, and this regulatory axis has been closely linked to both the formation of auricular morphology and the inter-individual variation observed in human populations (5,41).

Recent findings have further expanded the genetic landscape of earlobe morphology by identifying a novel association with ZFHX3. Although not universally implicated in ear development, ZFHX3 participates in several fundamental biological processes, including neuronal and myogenic differentiation, facial morphogenesis, chromatin remodeling, and mRNA processing (50). Relevance is the rs74030209 intronic variant within ZFHX3, which has been significantly associated with the attached versus free earlobe phenotype in a GWAS of 26,806 individuals from the Chinese population (50).

Another noteworthy genetic contributor is the rs10211400 SNP, located within the LINC01107 gene on chromosome 2. A 2022 GWAS conducted in a Chinese cohort identified a significant association between this variant and earlobe morphology, further underscoring the polygenic nature of this trait (50).

Categorical phenotypic traits such as the attached versus free earlobe configuration, which capture inter-individual genetic differences and population-specific variation, represent valuable hereditary markers within population genetics. In a genome-wide association study, Adhikari et al. (2015) identified four genomic loci —2q12.3, 2q31.1, 3q23, and 6q24.2—as being significantly associated with earlobe size, with the 2q12.3 and 2q31.1 regions specifically linked to the attached earlobe phenotype (39).

A large multi-ancestry GWAS conducted by Shaffer et al. further expanded the genetic landscape of earlobe attachment. Drawing from four cohorts and a combined sample of 74,660 individuals of European American, Latin American, and Chinese ancestries, the study identified several genes—EDAR, SP5, MRPS22, ADGRG6, KIAA1217, and PAX9—as significantly associated with earlobe attachment (5). These associations corresponded to specific SNPs, including rs3827760, rs6756973, rs9866054, rs58122955, rs7096127, and rs1950357, respectively (5).

Darwin's Tubercle

Darwin's Tubercle is a distinctive morphological feature located on the superior or middle portion of the helix, typically appearing as a small cartilaginous projection (22). Although evidence generally supports a polygenic mode of inheritance, some family-based studies have reported patterns consistent with autosomal dominant transmission (51).

A pivotal GWAS by Li et al. (41) greatly advanced current understanding of the genetic determinants of this trait. Using digital facial photographs from 14,921 individuals across five populations in Europe, Asia, and Latin America, the study identified multiple genes—EDAR, CART1, SP5, MRPS22, LRBA, LOC153910, and LOC100287225—as being associated with variation in ear morphology. Importantly, 16 SNP loci were linked not only to ear-related characteristics but also to male-pattern baldness. Within this dataset, the rs7812632 locus emerged as specifically associated with the presence or morphological variation of Darwin's Tubercle.

Despite these advancements, the accurate assessment and genetic prediction of Darwin's Tubercle remain challenging. As with other traits of the external ear, variability in expression, subtle phenotypic presentation, and potential population-specific effects continue to complicate both morphological classification and predictive modeling.

Applications of Ear Phenotype Prediction in Forensic Genetics

Significant progress has been made in FDP over the past decade, particularly in linking visible morphological traits with underlying genetic variants. However, the prediction of ear phenotypes remains comparatively underexplored. Given the substantial inter-individual variability and pronounced inter-population differences observed in auricular morphology, ear-based phenotyping represents a promising yet underutilized biometric parameter in forensic investigations (40).

A major contribution to this growing area is the Earplex FDP panel developed by Noreen et al. (40). In this study, 33 distinct ear phenotypes were assessed using digital ear images from 300

Pakistani volunteers, followed by genotyping of 21 phenotype-associated SNPs via SNaPshot™ multiplex assays and capillary electrophoresis. Multivariate logistic regression analyses yielded encouraging predictive performance, particularly for key traits such as antihelix morphology, Darwin's tubercle, and earlobe size (40).

The study identified multiple SNP loci with statistically significant associations to specific auricular traits. For example:

- Earlobe size was associated with rs17023457, rs1339766, rs1960918, rs161249, and rs9866054.
- The attached earlobe trait correlated with rs7428, rs7873690, and rs1960918.
- Antihelix morphology was linked to rs2080401.
- The superior crus of the antihelix was associated with rs17023457, rs1960918, rs9866054, rs1012049, and rs1347222 (40).

Collectively, these findings highlight the feasibility of integrating SNP-based predictions of ear phenotypes into forensic workflows. As additional population-specific datasets emerge, ear morphology could become an increasingly informative component of FDP-driven investigative intelligence.

Challenges and Limitations in Genetic Prediction of Ear Phenotypes

Despite substantial progress in the genetic prediction of external ear morphology, several challenges continue to impede its broader application in forensic and biomedical settings. A major limitation stems from the anatomical complexity of the auricle, which displays considerable inter-individual variation shaped by both genetic and environmental influences. This multifactorial basis complicates efforts to precisely disentangle and quantify the genetic determinants of ear morphology (52).

In addition, the pronounced morphological heterogeneity observed across global populations presents significant barriers to developing robust and generalizable prediction models. Population-specific variation necessitates rigorous standardization of phenotyping protocols, as inconsistencies in trait definitions or scoring criteria can markedly reduce cross-study comparability and undermine the validity of predictive models (52).

The polygenic nature of ear morphology further complicates predictive modeling. Numerous genes—each exerting small individual effects—collectively shape the phenotype, necessitating the use of advanced statistical or machine-learning approaches requiring large, diverse datasets for effective training and validation (53). This challenge is compounded by the fact that many existing GWAS in this domain are underpowered or lack replication in independent cohorts, thereby limiting the reliability and reproducibility of reported associations (5,48).

Age-related changes introduce additional variability, particularly in more elastic regions such as the earlobe. Features including elongation, sagging, or alterations in cartilage flexibility may progressively modify external ear morphology, reducing prediction accuracy when reference datasets predominantly comprise younger individuals (54).

Methodological inconsistencies in ear measurement practices also present significant limitations. Whether phenotypes are captured via direct anthropometry, 2D photography, or 3D imaging, discrepancies in landmark identification and measurement precision can produce subtle yet impactful variation. Such heterogeneity complicates cross-study comparisons and reinforces the need for standardized, validated phenotyping protocols (33).

Collectively, these challenges highlight the importance of interdisciplinary collaboration, greater methodological standardization, and the development of larger, ethnically diverse datasets to improve the accuracy, robustness, and generalizability of genetic prediction models for ear phenotypes (55).

CONCLUSION

In the context of rising global crime rates, forensic DNA analysis has become an indispensable component of modern judicial systems. Advances in forensic genetics have accelerated the identification of novel genetic markers that enable the prediction of EVCs, thereby enhancing individual identification when conventional reference samples are unavailable. DNA-based inference of physical traits is particularly valuable in suspect profiling, as SNPs associated with phenotypic variation can support reliable predictions of features such as hair, skin, and eye color, facial morphology, stature, and ear morphology.

Among these traits, ear morphology holds special relevance for biometric technologies incorporating facial recognition. Because each facial substructure must be precisely characterized both metrically and morphologically, the unique anatomical features of the ear provide an additional, highly discriminative dimension to identification efforts.

Moreover, the ear's anatomical position on the lateral aspect of the skull, coupled with its relatively low susceptibility to postmortem degradation —even in mass disaster settings— highlights its utility in victim identification. Despite substantial forensic and biometric attention directed toward frontal facial features, research specifically examining ear morphology remains comparatively limited.

Predictive models incorporating ear morphology have the potential to strengthen existing forensic DNA analysis pipelines and facilitate the identification of unknown human remains. The development of robust ear phenotype prediction models would not only enhance suspect profiling in criminal investigations but also support anthropological research and mass disaster victim identification by serving as valuable and systematically curated data resources.

At present, no commercially available forensic DNA panels are

dedicated to the prediction of ear phenotypes. Nevertheless, ongoing GWAS continues to uncover genetic variants influencing ear morphology, providing an increasingly solid foundation for future model development. Genotyping SNPs associated with ear structure—particularly those validated across diverse populations—could significantly improve phenotypic reconstruction and further advance the field FDP.

Conflict of Interests

The authors declare that there is no conflict of interest in the study.

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