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Antihelical Ossified Granule: A Recently Identified Potentially Inherited Human Trait in the Auricular Concha - A Case Study

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Abstract

In the early stages of research on the genetics of human morphological traits, particular attention was given to the details of the physical complexities of the face. In this field, prominent focus was placed on the characteristics of the auricle, especially the types of the earlobe (*lobulus auriculae*). This paper presents the first data on the previously undescribed possibility of inheriting the ossified granules of the antihelix. Based on genealogical analysis of the presence/absence of the granule across five successive generations of a Bosnian family, evidence is provided for the autosomal recessive inheritance of this dimorphism.

Keywords

human genetics, auricular antihelix, newly described polymorphism, autosomal recessive inheritance, Bosnia and Herzegovina

Introduction

The first known data on the genetic characteristics of the population of Bosnia and Herzegovina were collected by Austrian military doctors as part of standardized systematic examinations of candidates for recruitment into the Bosniaks regiments of the Austro-Hungarian army. In addition to the usual anthropometric indicators, they also classified phenotypic traits of highly heritable anthroposcopic characteristics, such as eye, hair, and skin color (Weissbach, 1895).

Thanks to primarily individual, and less frequently organized institutional research, the genetic structure of local populations in Bosnia and Herzegovina is better understood than that of other countries in the Western Balkans. This is equally true for the knowledge of the genetic structure of representative metapopulation samples, as well as for the investigation of potential factors contributing to genetic differentiation among local populations.

The observed trait is not included in any of the reference databases on inherited traits of individual variation such as OMIM database (Hunter & Yotsuyanagi. 2005).

Material and methods

The pedigree of the index case includes 71 individuals from five generations of a single family in Central Bosnia, with affected individuals ranging in age from 0 to 80 years. The control sample consisted of 126 student volunteers. A rough image of the granule was captured using a camera, after thoroughly illuminating the observed area on the auricle (Figure 1). The ultrasound recordings were performed at the SANASA polyclinic, Sarajevo (Figure 2).

Results and discussion

The comparison of test proportions was conducted using MedCalc online software. Sample 1 consisted of 3 individuals (2.38%) exhibiting the given trait, while Sample 2 included 123 individuals (97.62%) without the observed trait. The Chi-squared test revealed a statistically significant difference between the two groups (227.67), with a corresponding *p*-value of <0.0001. In the control sample of 126 student volunteers, only 3 individuals (1 male and 2 females) exhibited the trait (Figure 3; Table 1).

Figure 1. Personal camera: posterior surface of the helix (high-intensity personal camera with thorough lighting)

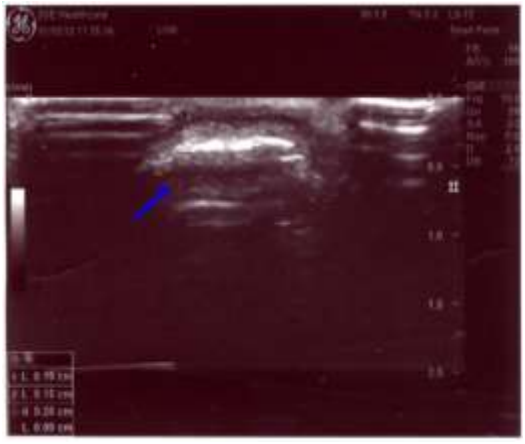

Figure 2. Ultrasound image of the given trait

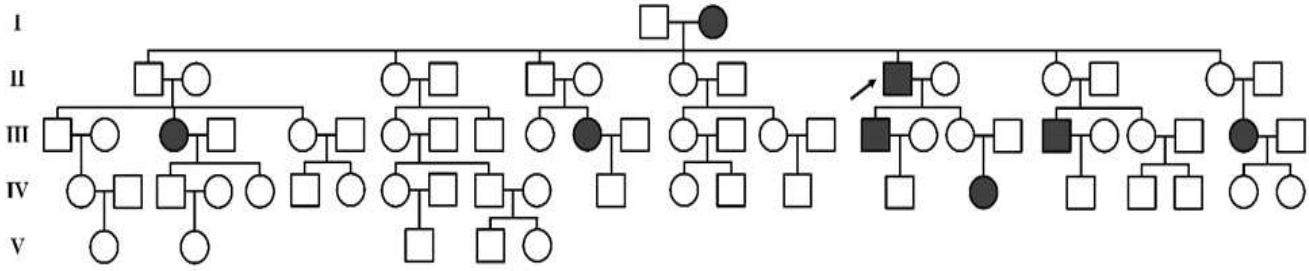

Figure 3. The proband's phenotypes for selected polymorphisms

Table 1. List of traits and their inheritance patterns

Trait	Recessive phenotype	Proband's phenotype
ABO blood group system	O	B
ABH antigens secretion	Nonsecretor	Secretor
PTC tasting	Nontaster	Taster
Green color vision	Blindness (protan)	Normal
Red color vision	Blindness (deutan)	Normal
Arm folding	Right/Left type	R
Hand clasping	Right/Left type	R
Ear lobe types	Attached	Free
Tongue rugosity (Fissured tongue) (Geographic tongue)	„Normal“	Fissured
Chin cleftness (Dimple in chin)	Smooth chin	Smooth
Midphalangeal hair (mid-digital hair)	Hairless	Hairy
Nail shape (Short nails)	Long nails	Long
Crooked fifth finger (Streblodactly)	„Normal“ phalanx	Normal
Digital index	Short (♀♀) and long (♂♂) index finger	Short
Tongue rolling (Tongue curling)	Non-roller	Roller
„Hitch-hiker’s” thumb	„Hitch-hiker’s” thumb	Hitch-hiker’s” thumb
Hyperextensibility of the proximal joint	„Hitch-hiker’s” thumb	Normal
ABO blood group system	O	B
ABH antigens secretion	Nonsecretor	Secretor
PTC tasting	Nontaster	Taster

Available literature (Quelprud, 1934, 1935; Lai & Walsh, 1966; Nakamura et al., 1995) and major databases, including OMIM, provide no records of comparable structural variations involving the antihelix complex of the external ear. Previously described hereditary auricular anomalies predominantly affect the helix, lobule, or general ear shape, highlighting the novelty of the present finding. The distribution of affected individuals, balanced by gender and age, indicates that the trait is neither sex-linked nor age-dependent. Its higher frequency among related individuals, compared with a control sample, supports a genetic basis. The consistent vertical transmission across multiple generations, coupled with the absence of the phenotype in presumed heterozygous carriers, is most consistent with an autosomal recessive inheritance pattern (Dutta & Ganguly, 1965; Bondenson & Miles, 1993; Canun et al., 2003). Developmentally, the antihelix derives from the second pharyngeal arch, where chondrogenic and morphogenetic processes shape its structure. Subtle alterations in cartilage differentiation or ossification could account for the observed morphological variant. Although specific genes cannot yet be identified, candidate pathways include those regulating auricular cartilage patterning (HOXA2, PAX1, BMP5) and matrix organization (COL2A1, ELN), as well as ossification regulators such as SOX9 and RUNX2. Future molecular studies could clarify whether this trait reflects a local variant of chondrogenic gene expression or a novel mutation affecting auricular development. From a population-genetic perspective, the presence of this recessive trait in a Bosnian family may reflect the region's historical isolation and occasional endogamy, which can increase the expression of rare alleles. Broader population surveys in the Western Balkans would help determine whether this represents a unique familial variant or a low-frequency regional polymorphism. Limitations include the small sample size, focus on a single pedigree, and reliance on morphological assessment without molecular or imaging confirmation. Future studies incorporating genetic analyses, morphometric methods, and comparative data from other populations will be essential to validate and characterize this trait further.

Conclusion

This study reports a previously undescribed hereditary morphological variant of the auricular antihelix, documented across five generations of a Bosnian family. The balanced distribution of the trait between sexes and its transgenerational occurrence support an autosomal recessive pattern of inheritance. Although the molecular basis remains unknown, the finding highlights potential involvement of genes regulating auricular cartilage development and ossification. The identification of this rare trait expands current knowledge of human auricular variation and underscores the importance of detailed morphological and genetic investigations within regional populations such as those of the Western Balkans. Broader population and molecular studies will be essential to confirm the genetic mechanism and to assess the prevalence and evolutionary significance of this newly characterized auricular feature.

Authors' contributions

R. Hadziselimovic:

Conceptualization, Methodology, Investigation, Data curation, Visualization, Writing – original draft, Writing – review & editing.

Conflict of interest

Author declares no conflict of interest.

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