

2 Multiethnic GWAS reveals polygenic architecture of earlobe attachment.

Shaffer JR, Li J, Lee MK, Roosenboom J, Orlova E, Adhikari K, 23andMe Research Team, Gallo C, Poletti G, Schuler-Faccini L, Bortolini MC, Canizales-Quinteros S, Rothhammer F, Bedoya G, González-José R, Pfeiffer PE, Wollenschlaeger CA, Hecht JT, Wehby GL, Moreno LM, Ding A, Jin L, Yang Y, Carlson JC, Leslie EJ, Feingold E, Marazita ML, Hinds DA, Cox TC, Wang S, Ruiz-Linares A, Weinberg SM

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Earlobe attachment, like tongue rolling, is one of the supposedly simple genetic traits that are still (incorrectly) taught in school and even university classes as autosomal dominant. The paper by Shaffer et al. uses genomic and phenotypic data from 65,000 customers of the gene-testing company 23andMe to perform a genome-wide association study (GWAS) of earlobe attachment in combination with three smaller cohorts (with a total of 10,000 subjects). A total of 49 gene loci were identified, including confirmation of the six loci already known from the existing three cohorts. There might be more interesting and more urgent scientific questions than the complex genetics of earlobe attachment, but in its harmlessness, the trait has been skillfully chosen to demonstrate the increasing power of genetic research from customer data bases, which can amass extreme numbers of subjects, unheard of in other settings. The question will be, if the quality and depth of phenotyping, which is obviously rather shallow in the case of earlobe attachment, can keep that pace.

Disclosures

None declared

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Abstract:

ABSTRACT

The genetic basis of earlobe attachment has been a matter of debate since the early 20th century, such that geneticists argue both for and against polygenic inheritance. Recent genetic studies have identified a few loci associated with the trait, but large-scale analyses are still lacking. Here, we performed a genome-wide association study of lobe attachment in a multiethnic sample of 74,860 individuals from four cohorts (three with the trait scored by an expert rater and one with the trait self-reported). Meta-analysis of the three expert-rater-scored cohorts revealed six associated loci harboring numerous candidate genes, including EDAR, SP5, MRPS22, ADGRG6 (GPR128), KIAA1217, and PAX9. The large self-reported 23andMe cohort recapitulated each of these six loci. Moreover, meta-analysis across all four cohorts revealed a total of 49 significant ($p < 5 \times 10^{-8}$) loci. Annotation and enrichment analyses of these 49 loci showed strong evidence of genes involved in ear development and syndromes with auricular phenotypes. RNA sequencing data from both human fetal ear and mouse second branchial arch tissue confirmed that genes located among associated loci showed evidence of expression. These results provide strong evidence for the polygenic nature of earlobe attachment and offer insights into the biological basis of normal and abnormal ear development.

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