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Variation and signatures of selection on the human face

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ABSTRACT

There has been much debate about why humans throughout the world differ in facial form. Previous studies of human skull morphology found levels of among-population differentiation that were comparable to those of neutral genetic markers, suggesting that genetic drift (neutral processes) played an important role in influencing facial differentiation. However, variation in soft-tissue morphology has not been studied in detail. In this study, we analyzed high-resolution 3D images of soft-tissue facial form in four Eurasian populations: Han Chinese, Tibetans, Uyghur and Europeans. A novel method was used to establish a high-density alignment across all of the faces, allowing facial diversity to be examined at an unprecedented resolution. These data exhibit signatures of population structure and history. However, among-population differentiation was higher for soft-tissue facial form than for genome-wide genetic loci, and high-resolution analyses reveal that the nose, brow area and cheekbones exhibit particularly strong signals of differentiation (Q_{st} estimates: 0.3–0.8) between Europeans and Han Chinese. Our results suggest that local adaptation and/or sexual selection have been important in shaping human soft-tissue facial morphology.

Introduction

Humans are characterized by variation in many external features, including the shape of the brow area and nose, skin and eye color, various properties of body hair, and body size and proportions. The extent to which such variation can be explained by genetic drift (neutral processes), local adaptation, or sexual selection has been much debated, going back to Darwin (1871). One approach to unraveling the relative influence of neutral versus selective processes on the differentiation of human traits is to compare the relative amounts of within-population and between-population variance. It is well known that in humans the majority of the genetic variance (~90%) is found within continental regions, whereas only a minor portion (~10%) is accounted for by differences between regions (Weir et al., 2005; Barreiro et al., 2008). Such apportionment of diversity is generally accepted as the amount of differentiation expected under neutral evolution (Relethford, 2002). In genetic data, differentiation is mainly measured using Wright's fixation index (F_{st}) (Wright, 1950), while an analogous statistic has also been defined for phenotypic variation contributed to by genetic factors, usually called Q_{st} (Spitze, 1993). Consequently, the direct comparison of F_{st} and Q_{st} constitutes a useful neutrality test for phenotypic traits, where strong deviations of Q_{st} from neutral F_{st} levels are suggestive of non-neutral evolution (Miller et al., 2008).

In humans, studies of phenotypic diversity and apportionment have mainly focused on features that can be measured on skeletal

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remains (in particular, skulls). Based on 57 inter-landmark distances, Relethford (1994) first reported that the variation among continental regions accounted for ~10% of overall craniometric variation (Relethford, 1994), in good agreement with the apportionment based on neutral genetic loci. Later studies that utilized various sets of measurements (Harvati and Weaver, 2006; von Cramon-Taubadel, 2009a), different methods for partitioning variation (e.g., principal components analysis (Roseman and Weaver, 2004)), 3D landmark data (Harvati and Weaver, 2006; von Cramon-Taubadel, 2009b) or different samples (Harvati and Weaver, 2006; Hubbe et al., 2009; von Cramon-Taubadel, 2009b), have repeatedly come to the conclusion that the morphological variation in the human skull has been largely shaped by neutral evolution (Roseman and Weaver, 2007; von Cramon-Taubadel and Weaver, 2009; Relethford, 2010). The finding of a close correspondence between phenotypic distance and geographic distance is also consistent with the idea that human skull variation has been shaped by neutral evolutionary processes (Relethford, 2004a,b; 2009). In addition, as with genetic diversity, human craniometric variation can be used to infer population structure and history (Harvati and Weaver, 2006; Gunz et al., 2009; von Cramon-Taubadel, 2009b).

This notwithstanding, the relatively low levels of differentiation of craniometric features strongly contrasts with the situation for skin pigmentation, which exhibits the most variation (~80%) among populations (Relethford, 2002). It also contradicts the intuitive notion that there exists extensive population variation in facial features across the world (Nei and Roychoudhury, 1982; Wright, 1992; Howells, 1995; Gill, 1998; Hennessy and Stringer, 2001). Indeed, adaptive hypotheses have been proposed for a number of craniofacial features. For example, the shape of the nose has long been hypothesized to play an important role in climatic adaptation (Thomson and Buxton, 1923; Coon et al., 1950, 1955). Consistently strong correlations have repeatedly been found between the nasal index (ratio of nose breadth/height) and temperature and humidity (Thomson and Buxton, 1923; Davies, 1932; Weiner, 1954; Wolpoff, 1968; Hiernaux and Froment, 1976; Crognier, 1981; Franciscus and Long, 1991). Recent studies have also reported higher among-population differentiation values (maximum Q_{st} ~ 0.4) than expected under neutrality for several nasal measurements (Roseman, 2004; Roseman and Weaver, 2004; Hubbe et al., 2009). Nonetheless, these studies were all based on the skeletal elements of the nose, leaving the soft-tissue external nose poorly studied.

To date there has been no systematic study of the variation in soft-tissue facial form even though soft-tissue facial form may have experienced greater selection pressures than the underlying skull due to the direct exposure to the environment. Selection might shape the skin, cartilage or adipose tissue distribution, rather than the skull bones. We therefore applied a new approach to analyze variation in soft-tissue facial form. In brief, high-resolution 3D facial images were taken from individuals from four Eurasian populations: Han Chinese from East China (HAN), Tibetans (TIB), Uygur (UYG) (an admixed population with European and Chinese ancestry) and Europeans (EUR). A novel 3D facial surface alignment approach was applied to automatically annotate 15 facial landmarks, and to subsequently establish a dense point-to-point correspondence for ~30,000 3D point markers, with a resolution of one point per 1 mm × 1 mm surface. The high-density data were then aligned to the same Cartesian coordinate system using generalized partial Procrustes analysis (pGPA) (Dryden and Mardia, 1998). Analyses of population structure and variance apportionment were carried out on both the whole face and specific facial features. We find that variation in the soft tissue morphology of the human face has been influenced by both population history and selection.

Materials and methods

Ethics statement

Sample collection for this study was carried out with the approval of the ethics committee of the Shanghai Institutes for Biological Science and in accordance with the standards of the Declaration of Helsinki. Written informed consent was obtained from every participant.

Data and sample collection

The 3dMDface® system (www.3dmd.com/3dMDface) was used to collect high-resolution 3D facial images from volunteers who took part in this study. Four hundred Han Chinese (200 females and 200 males) who were 17–25 years old were sampled in Taizhou, Jiangsu Province. Three hundred and three Uyghur (200 females and 103 males) who were 17–25 years old were sampled in Kashi, Xinjiang. One hundred sixty-nine Tibetans (100 females and 69 males) who were 15–22 years old were sampled in Shigatse. All participants were required to have the same ancestry over three generations. Finally, 89 individuals of self-reported European ancestry (32 females and 57 males) between 16 and 57 years old were collected in Shanghai. They were required to have complete European ancestry over the last three generations. The country of origin of all three generations is shown in Appendix A, Supplementary Online Material (SOM), Fig. S1. Eighty-one percent of the individuals studied have the same place of origin as their parents, and 79% have the same place of origin as their parents and grandparents. The age distributions of all four samples are shown in SOM, Fig. S2. Individuals with obvious health problems or any history of facial surgery were excluded from the study.

High-density 3D facial image alignment

We developed a novel approach for aligning a dense set of quasi-landmarks (Rohr, 2001), evenly distributed on the facial surface, to enable facial comparisons (Guo et al., 2013). First, 15 salient facial landmarks (SOM, Fig. S3) are automatically recognized. In brief, the automatic landmark recognition starts with identifying the location of the pronasale by searching a semi-sphere centered on the nose, followed by pose normalization, which is to align all sample faces to a uniform frontal view. Shape depth (z axis) values and surface texture are then projected to the x–y 2D plane (the frontal portrait plane), where highly specific texture/shape signatures of endo/ecto-canthions and cheilions are identified by a principal components analysis (PCA) based approach. The remaining 10 landmarks are recognized by heuristic methods using geometric relations and texture constraints (Guo et al., 2013). Next, a reference face is chosen and the surface mesh is re-sampled to achieve an even density of one point per vertex of a 1 mm × 1 mm grid. In total, ~30,000 points are used to construct the reference mesh. Third, this reference facial mesh is warped to each face in the sample to ensure the proper matching of all of the 15 landmarks, via a thin-plate spline (TPS) transformation (see SOM, Fig. S4). Fourth, the grid points of the reference face are projected onto each face in the sample. The resulting points of projection, which have a one to one correspondence with the grid points of the reference, are used to define a mesh that describes the surface of each of the faces in the sample (SOM, Fig. S4). Finally, these sample grids are aligned to achieve a common coordinate system by pGPA, in which scaling is not used and size information is preserved. We did not remove size information so that potential differentiation involving size changes could be examined. Details of this alignment method are described elsewhere (Guo et al., 2013) and the corresponding software is