

HOW TO MEASURE PHENOTYPIC VARIATION IN HUMAN DEVELOPMENT AND EVOLUTION?



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MEASURING SHAPE VARIATION

Ontogenetic development determines how genetic variation translates into phenotypic variation, which in turn influences a population's response to natural selection. The study of phenotypic variance-covariance patterns hence is central for bridging developmental and evolutionary biology, and it is a valuable alternative to experimental approaches in studies of human development. Characterized by a large number of variables, modern morphometrics is highly effective for detailed exploratory studies, but a biologically meaningful analysis of variance-covariance patterns is difficult in high-dimensional data spaces.

What makes a statistic or a conclusion derived from a statistic biologically meaningful? One necessary criterion for scientific meaningfulness is the invariance to arbitrary decisions in the course of the analysis (Narens 2002; Huttegger & Mitteroecker 2011): For example, a conclusion should not depend on the units of the variables (whether a measurement is expressed in mm or in cm), or on the exact count and spatial distribution of measurements (small changes in the landmark scheme should not affect the conclusions).

In geometric morphometrics all variables are of the same unit, but the equal weighting of all variables is both statistically and biologically arbitrary. Furthermore, the number and degree of redundancy of landmarks, which is likewise often arbitrary, can be considered as a form of linear weighting of the corresponding shape features or traits (Figure 1). In order to warrant a meaningful biological interpretation, a measure of shape variation hence should be unaffected by linear scaling of the variables (shape coordinates).

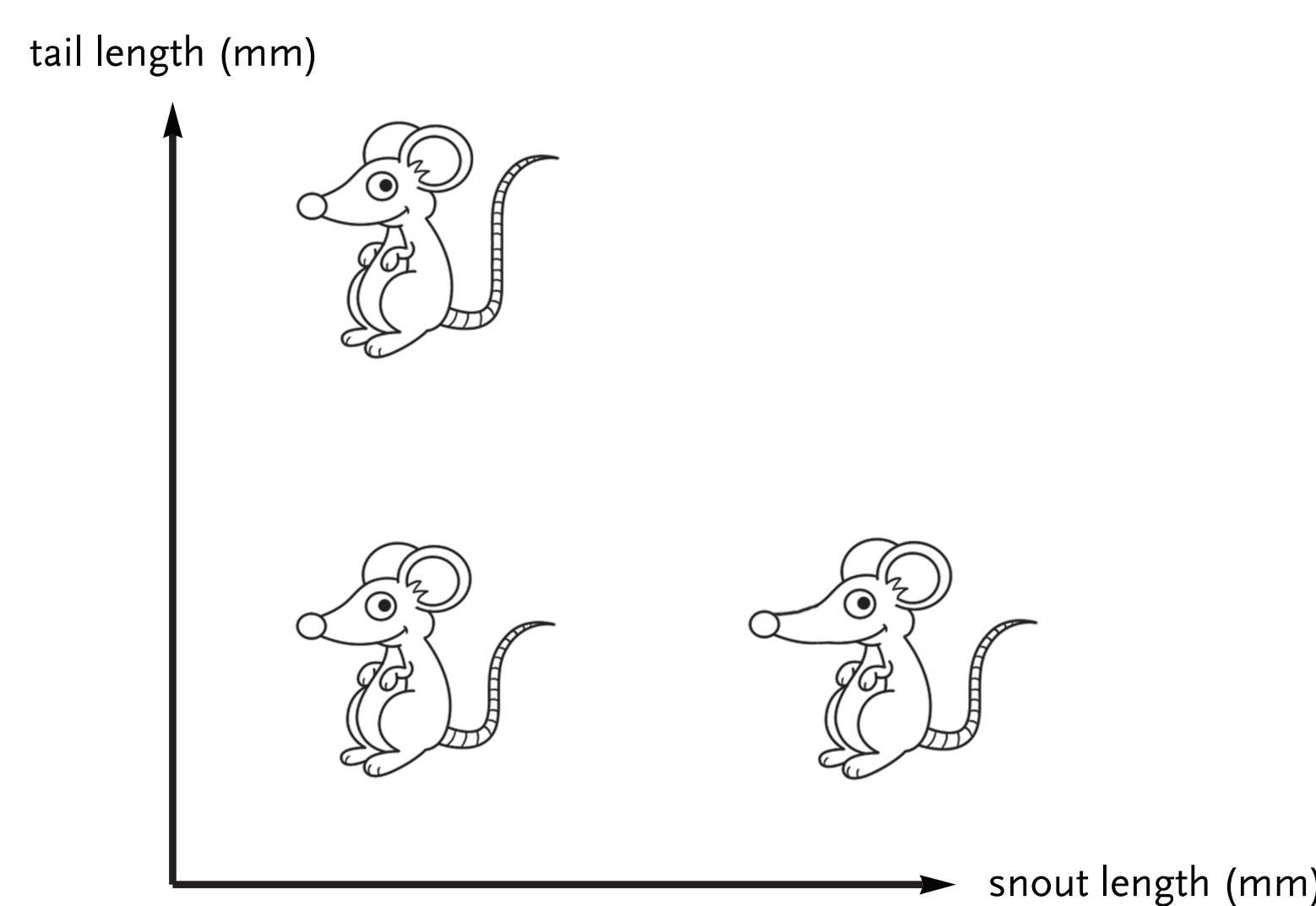


Figure 1. Is 1 mm difference in tail length comparable to 1 mm difference in snout length? What if the tail was described by ten measurements and the snout by one measurement?

The equal weighting of variables in multivariate statistics is arbitrary from a biological perspective, whereas the – often arbitrary – number and spatial distribution of landmarks is a form of linear weighting of traits in many statistics. A biologically meaningful statistic hence should not depend on linear scaling or weighting of the variables.

To most multivariate statistics all variables contribute independently from all other variables, yet variables may be geometrically dependent (e.g., through common size correction or common landmark superimposition) or be subject to spatial autocorrelation (closely adjacent landmarks cannot be considered as independent). It is thus also desirable that a statistic does not depend on assumptions about linear (in)dependence, as long as they are not explicitly modeled. For certain variables, such as for landmark coordinates, the origin of the coordinate system is arbitrary, so that statistics should further be unaffected by changes of the origin. The class of all linear transformations of the variables (linear scaling and changes of linear dependence) together with translation of the origin is called the class of affine transformations. Hence, in the most general case, a biologically meaningful statistic should be *affine invariant*.

A useful way to compare variances in two groups A and B is in terms of a ratio $\text{var}(x_A)/\text{var}(x_B)$, because this ratio is invariant (unchanged) under linear transformations of the variables, such as changes of the unit. One way to quantify the overall amount of variation in a multivariate data set is the *generalized variance*, which is computed as the determinant of the variance-covariance matrix or as the product of the eigenvalues of this matrix. Ratios of generalized variances are invariant under all affine transformations of the variables.

DEVELOPMENTAL VARIATION OF HUMAN CRANIOFACIAL SHAPE

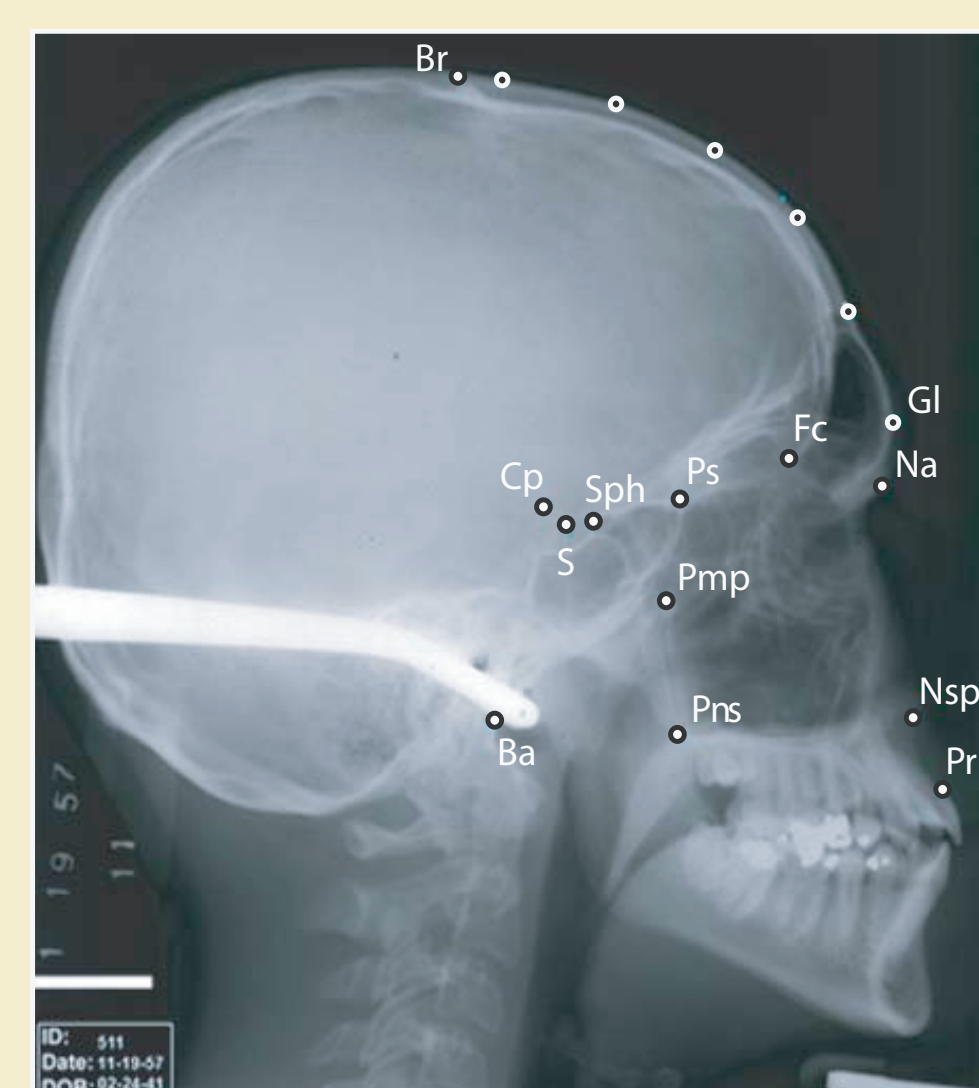


Figure 2. The sample consists of longitudinal cephalograms from the Denver Growth Study of 13 males and 13 females, beginning at age one month approximately every year up to the age of 18 years. On every radiograph, 18 landmarks (including six semilandmarks) on the face and the cranial base were digitized by Ekaterina Stansfield (Bulygina et al. 2006). The landmark configurations were superimposed by Generalized Procrustes analysis (Rohlf & Slice 1990). All further analyses were based on the first 7 principal components of these shape coordinates, accounting for 90% of total variance.

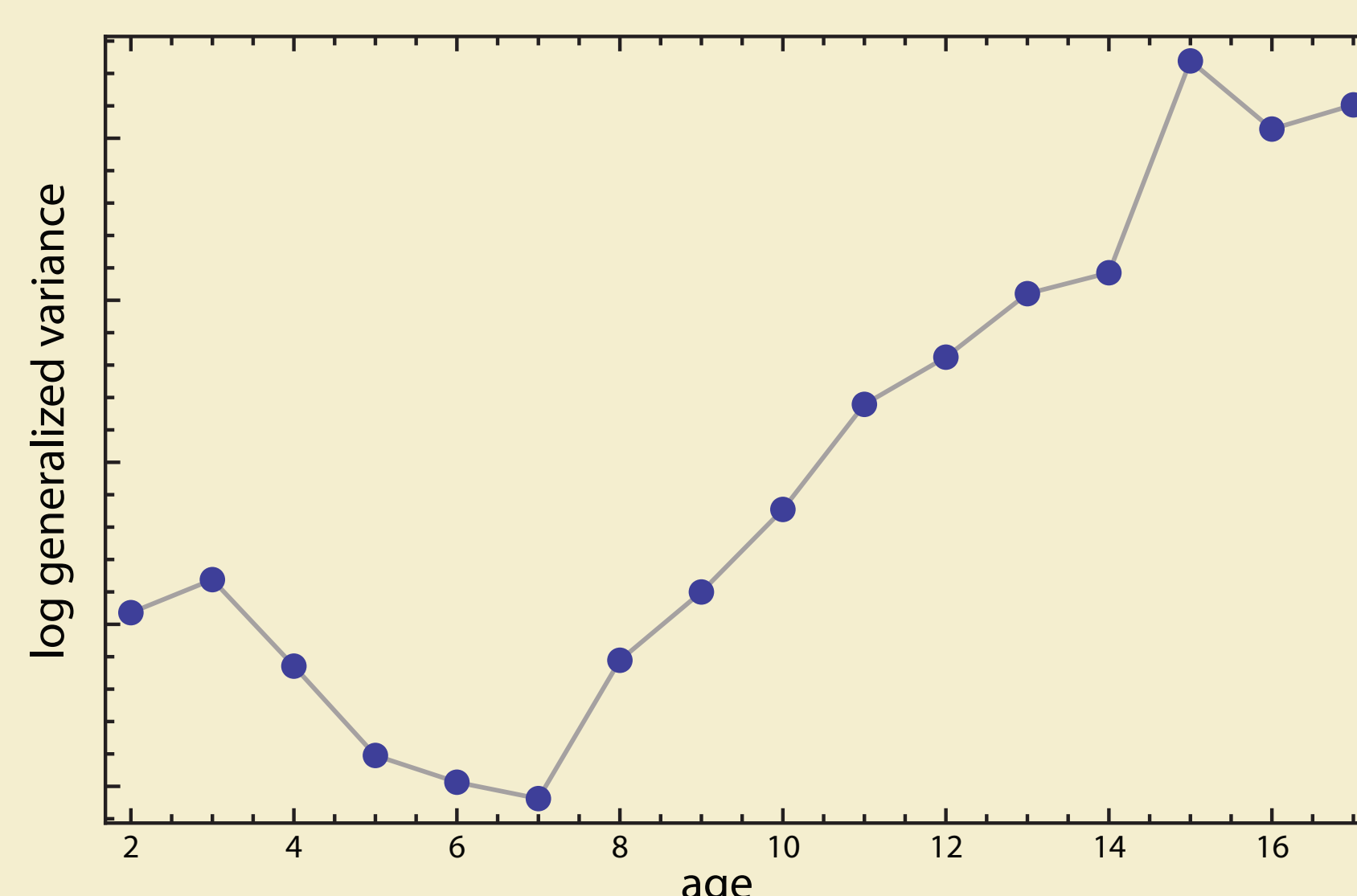


Figure 3. Overall craniofacial shape variation as measured by the generalized variance is decreasing until about 6-7 years of age and increasing again thereafter. The peak at about 15 years reflects variation in the onset of the pubertal growth spurt.

Despite this general ontogenetic trend, different shape features might show different dynamics. Figure 4 thus separately assesses the two shape features (linear combinations of shape coordinates) with maximum increase and maximum decrease of variance in the observed age range. Note that maximum ratios of variance are also affine invariant (Flury 1985; Mitteroecker & Bookstein, in prep.).

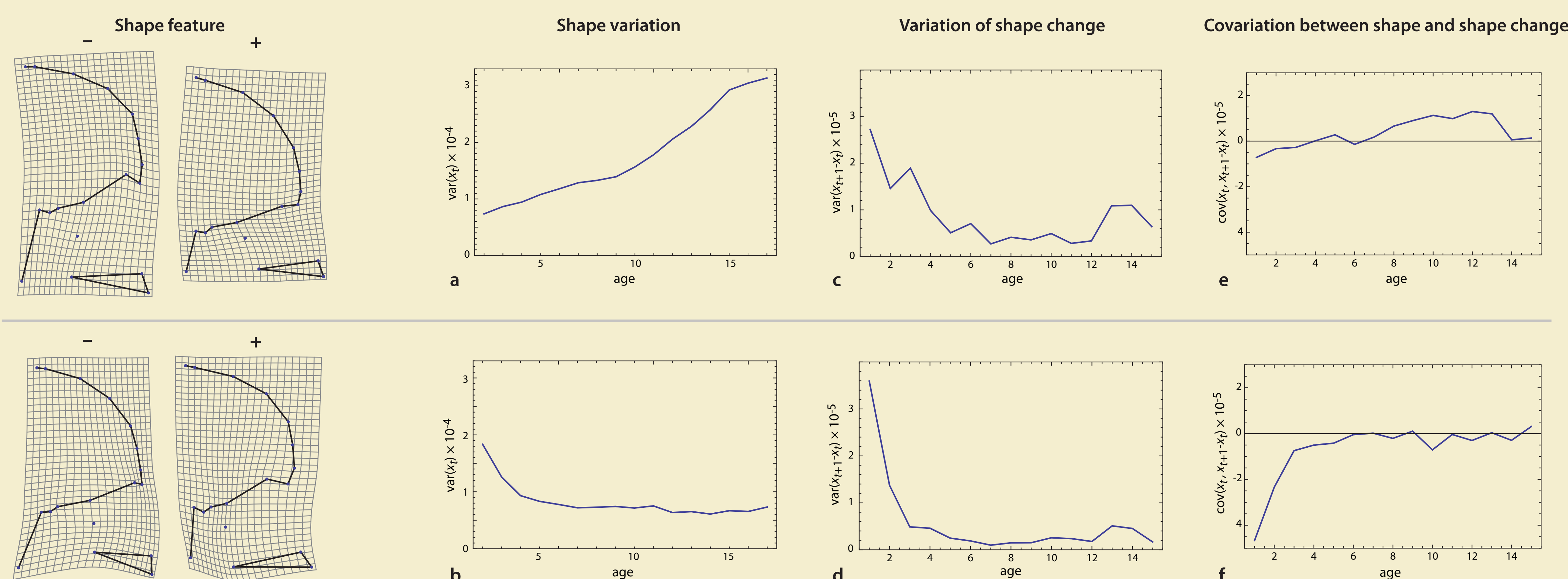


Figure 4. Relative facial height is the shape feature with maximum increase in variance during postnatal ontogeny (a), whereas the relative size of the pharynx decreases most in variance (b).

For both features, the amount of ontogenetic change varies across individuals, particularly within the first 7 years of age (c, d).

For facial height, the ontogenetic variance accumulates, leading to increasing shape variation throughout postnatal ontogeny. For relative pharyngeal size, by contrast, ontogenetic change is negatively correlated with age-specific shape (f), which is referred to as targeted growth or developmental canalization, and hence shape variation decreases.

SUMMARY

Pooled over all landmarks, age-specific shape variation decreases until about 6 years of age and increases again thereafter until early adulthood. But different shape features have different developmental dynamics: The variance of relative facial height increases throughout postnatal ontogeny, whereas the development of the pharyngeal region is canalized, leading to a decrease of shape variance within the first 5 years. Given the apparent functional importance of pharyngeal size, the develop-

mental canalization might be an evolved property to avoid obstruction of airways within the first few years of life (Coquerelle et al. 2013). Relative facial height, by contrast, is of less functional relevance and variation in its development accumulates. These different developmental dynamics might also affect evolution, as a developmentally canalized trait might also be constrained throughout phylogeny.

Because of the affine invariant statistics, these conclusions are independent of

any assumptions about equal weighting and geometric or spatial independence of measurements, and they are unaffected by small changes of the number and spatial distribution of landmarks. Affine invariance is of course not a sufficient criterion for scientific meaningfulness, but the *consilience* of temporal, spatial, and functional lines of evidence warrants a biological interpretation of the multivariate statistical results (Bookstein, in press).

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