

IN BRIEF

GENOMICS

The diploid genome sequence of an individual human.

Levy, S. *et al. PLoS Biol.* **5**, e254 (2007)

J. Craig Venter has made public the first entire, diploid genome sequence of a single individual, himself. Such a sequence — obtained by a modified version of random shotgun sequencing — allows a comparison of the variation between the two haploid chromosome sets. This within-individual variation, mainly in the form of SNPs and insertion and deletion polymorphisms (indels), is surprisingly high: chromosomal copies are only 99.5% identical, leading the authors to estimate that that variation between individuals is about five times higher than expected. The authors' hope is that such efforts will aid disease-gene mapping and stimulate personalized genome sequencing.

DEVELOPMENTAL BIOLOGY

Wnt signaling mediates regional specification in the vertebrate face.

Brugmann, S. A. *et al. Development* **134**, 3283–3295 (2007)

This paper reports that the regional specification of the vertebrate face and the changes in face morphology that occur during evolution depend on the distribution of Wnt signalling during embryonic development. During embryogenesis, Wnt signalling is active in face regions that protrude at later stages, corresponding to areas of cell proliferation. Mice in which Wnt signalling was disrupted had a flatter midface and more widely set eyes. The fact that a similar treatment had comparable consequences in chicks suggests that the distribution of Wnt signalling determines species-specific facial features.

HUMAN GENETICS

A common variant of *HMGA2* is associated with adult and childhood height in the general population.

Weedon, M. N. *et al. Nature Genet.* 2 September 2007 (doi:10.1038/ng2121)

The authors report the first genetic variant to contribute to variation in adult and childhood height in humans. The link between stature and a SNP in the 3' untranslated region of the high-mobility group AT-hook 2 gene (*HMGA2*) emerged from an association study involving nearly 5,000 individuals, and was confirmed in a larger study of >19,000 individuals. This gene is a good candidate for height, as *HMGA2* mutations cause size reduction in mice and humans. This study should spur further genetic investigations into visible traits in humans.

GENOMICS

Human genome ultraconserved elements are ultraselected.

Katzman, S. *et al. Science* **317**, 915 (2007)

Deletion of ultraconserved elements yields viable mice.

Ahituv, N. *et al. PLoS Biol.* **5**, e234 (2007)

Ultraconserved elements are largely non-coding stretches of >200 bp that are identical among humans, mice and rats. Rather than being mutational cold spots, Katzman *et al.* show that these stretches are under strong negative selection, more so than protein-coding sequences. Ahituv *et al.* report the unexpected finding that removing four ultraconserved elements from mice has no apparent phenotypic consequence, implying that these elements are not as essential as their conservation suggests.

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