

RESEARCH HIGHLIGHTS

Selections from the scientific literature

HUMAN GENETICS

Not-so-rare gene variants

By sequencing more human genomes more thoroughly than ever before, researchers have shown that most of the genetic variation in the human population is due to rare variants. These rare versions of genes are more likely to affect human health than are their common counterparts — which could make it more difficult to link gene variants to disease than previously thought.

Joshua Akey and Michael Bamshad at the University of Washington, Seattle, and their colleagues sequenced 15,585 genes in 2,440 people. The researchers found that 86% of the gene variants that they detected were rare, as were more than 95% of variants that were predicted to have a medical or biological effect.

Meanwhile, a team including Matthew Nelson at GlaxoSmithKline in Research Triangle Park, North Carolina, and John Novembre at the University of California, Los Angeles, report that more than 74% of variants identified by sequencing 202 genes in 14,002 people were carried by only one or two of the individuals.

Andy Clark and Alon Keinan of Cornell University in Ithaca, New York, used data from previous sequencing studies to show that rapid growth of the human population, starting 1,400 years ago, helped to seed the human genome with numerous rare variants.

Science <http://dx.doi.org/10.1126/science.1219240>; <http://dx.doi.org/10.1126/science.1217876>; 336, 740–743 (2012)

For a longer story on this research, see go.nature.com/lssahs



CALIFORNIA DEPT OF WATER RESOURCES

HYDROLOGY

Groundwater down, sea level up

Groundwater depletion and other human-induced changes in terrestrial water storage were responsible for almost half of the increase in global sea level observed between 1961 and 2003.

Thermal expansion of the oceans, the melting of polar ice and changes in terrestrial water reservoirs all contribute to rising sea levels. To single out the impact of changes in land-based water use on sea level, Yadu Pokhrel at the University of Tokyo and his colleagues ran a simulation of global terrestrial water stocks and flows, accounting for human activities such as irrigation (pictured), which often uses

groundwater, and dam-building.

The model suggests that about 42% of the almost eight-centimetre rise in sea level observed over the study period has resulted from changes in terrestrial water storage, particularly groundwater use. This human contribution could explain the discrepancy between observed and expected sea levels, the authors say.

Nature Geosci. <http://dx.doi.org/10.1038/ngeo1476> (2012)

For a longer story on this research, see go.nature.com/damx2m

NEUROSCIENCE

Brain stimulation zaps fear

Electrical stimulation of certain brain regions has been used to treat obsessive–compulsive disorder (OCD), but how it works is unknown. Researchers suggest that the stimulation could work by acting on brain circuits involved in dampening fear responses.

Gregory Quirk and his team at the University of Puerto Rico in San Juan implanted electrodes in the

brains of rats. The authors conditioned the animals to expect a footshock whenever they heard a tone and then, the next day, exposed the rats to the tone without the shock to extinguish the conditioned fear. When, on the second day, the researchers stimulated a specific area of a brain region similar to that targeted during OCD treatment, the rats later showed less fear on hearing the tone than did control rats. The stimulation also boosted the expression of a marker of plasticity — the ability to reorganize neural

pathways — in regions involved in extinguishing fear. This suggests that the brain stimulation enhances fear extinction.

Proc. Natl Acad. Sci. USA <http://dx.doi.org/10.1073/pnas.1200782109> (2012)

BIOENGINEERING

Viruses as power generators

Thin films of viruses can generate an electrical charge when subjected to mechanical stress — a form of energy