Sequencing seizures: Discovering new genetic mutations behind epilepsy

Neurologist Prof Sam Berkovic and molecular geneticist Prof David Goldstein describe their work uncovering chance mutations that cause childhood epilepsy. Presented by Dr Dyani Lewis.

"... about 20 years ago we had a fairly simple view that as the genetics tumbled out, we'd have one syndrome that we recognise as clinicians and that there'd be one gene for that. How wrong we were." -- Prof Sam Berkovic

Prof Sam Berkovic

Sam Berkovic ([http://www.brain.org.au/epilepsyresearch/staff/profiles/s_berkovic.htm](http://www.brain.org.au/epilepsyresearch/staff/profiles/s_berkovic.htm)) is Laureate Professor in the Department of Medicine, University of Melbourne. He is a clinical neurologist and clinical researcher with strong basic science collaborations. His group, together with molecular genetic collaborators in Adelaide and Germany, discovered the first epilepsy gene in 1995 and subsequently have been involved the discovery of many of the known epilepsy genes. This has changed the of the causes of epilepsy and is having a major impact on epilepsy research, and on strategies for diagnosis and development of new treatments. Currently he a co-PI on “Epi4K” a major NIH funded project to sequence 4,000 epilepsy genomes. He was elected a Fellow of the Royal Society (London) in 2007.

Prof David Goldstein

David Goldstein ([http://humangenome.duke.edu/people/david-goldstein](http://humangenome.duke.edu/people/david-goldstein)) is Professor of Molecular Genetics & Microbiology, Professor of Biology and Director of the Center for Human Genome Variation at Duke University. He received his Ph.D. in Biological Sciences from Stanford University in 1994, and from 1999-2005 was Wolfson Professor of Genetics at University College London. In April 2007, he was appointed Honorary Professor, Institute of Neurology, University College London.

London, UK. Dr. Goldstein is the author of over 200 scholarly publications in the areas of population and medical genetics. His principal interests include human genetic
diversity, the genetics of disease, and pharmacogenetics. He is the recipient of one of the first seven nationally awarded Royal Society / Wolfson research merit awards in the UK for his work in human population genetics. Dr. Goldstein was awarded the Triangle Business Journal Health Care Heroes Award: Innovator/Researcher Award, March 2008.

Credits

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