Thomson Reuters 2014 Citation Laureates: Their Impact on the World

Chemistry

Graeme Moad
Ezio Rizzardo
San H. Thang

Synthesizing polymers with specifically tailored chemical properties, for adhesives, cosmetics, hydraulic fluids and in drug delivery and biotechnology

Charles T. Kresge
Ryong Ryoo
Galen D. Stucky

Nanomaterials with pores that offer large internal surface area for delivering drug compounds, creating bio- sensors, and other biotechnological uses

Ching W. Tang
Steven Van Slyke

Invention of the first organic light-emitting diode, technology in the displays of smartphones, tablets, digital cameras, and high-definition televisions

Physics

Charles L. Kane
Laurens W. Molenkamp
Shoucheng Zhang

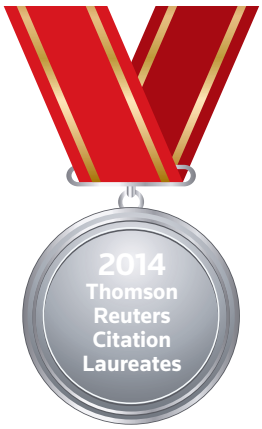
Accelerated the speed of computing

Ramamoorthy Ramesh
James. F. Scott
Yoshinori Tokura

Increased memory storage and power usage in computers and other electronic devices

Peidong Yang

Nanowires with varied optical properties: from generating lasers to capturing the sun's energy



Annually, Thomson Reuters analysts from IP & Science study scientific literature citation data to identify scientists whose work is worthy of recognition with a Nobel Prize. These individuals are of "Nobel Class," producing highly cited papers, often at levels exceeding 1,000 citations, and are typically authors of multiple highly cited papers.

The Thomson Reuters Citation Laureates have been cited so often in the last two or more decades that they typically rank in the top 0.1% by citations in their research areas. Our top picks for 2014 can be found at **sciencewatch.com/nobel**. And, the technologies they've influenced are shown in this infographic.

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Medicine

James E. Darnell
Robert G. Roeder
Robert Tjian

Insights into the actions and regulation of genes promise improved treatment for diabetes and other diseases

David Julius

Understanding the molecular workings of how we perceive temperature, leading to advances in pain management

Charles Lee
Stephen W. Scherer
Michael H. Wigler

Clarifying how specific genetic variations link to disease — knowledge that is improving diagnosis and treatment

Economics

Philippe M. Aghion
Peter W. Howitt

Advancing the theory of Creative Destruction: innovation & transformation constantly alter the economic and business landscape to the advantage of those capable of adapting

William J. Baumol
Israel M. Kirzner

Advancement of the study of entrepreneurism

Mark S. Granovetter

Economic Sociology: economic activity is "embedded" within social relationships and social networks, enabling complex & consequential economic interactions

For more information on the 2014 Thomson Reuters Citation Laureates, please go to http://sciencewatch.com/nobel

MEDICINE

FOR THEIR DISCOVERY OF LARGE-SCALE COPY NUMBER VARIATION AND ITS ASSOCIATION WITH SPECIFIC DISEASES

Michael H. Wigler
Professor and Head, Mammalian Cell Genetics Section, Cold Spring Harbor Laboratory, Cold Spring Harbor, NY USA

Charles Lee
Professor and Scientific Director, Jackson Laboratory for Genomic Medicine, Farmington, CT USA

Stephen W. Scherer
Senior Scientist and Director, The Centre for Applied Genomics, The Hospital for Sick Children; Professor and Director, McLaughlin Centre, University of Toronto, Toronto, ON CANADA

ESSAY

GENOMIC VARIATION AND DISEASE

The publication of the first drafts of the human genome led many scientists to proclaim that the difference in genetic sequence between any two humans was around 1%. The normal genome was assumed to be 99% identical in all humans, and the differences—underlying all the differences not only in simple things like eye color but also susceptibility to diseases—was down to around 3 million single-nucleotide polymorphisms, or SNPs. Lee and Scherer together and Wigler independently blew this assumption apart.

In 2004, shortly after publication of the human genome, Scherer and Lee showed that the human genome contained what they described as "large-scale variation." Earlier that year, Wigler had described "large-scale copy number variation." Both groups had found that there are large areas of the genome where long stretches, often encompassing several adjacent genes, are either duplicated or deleted. These differences between genomes are now called copy number variants (CNVs). Far from genomes being 99% identical, CNVs cover around 12–13% of the genome and about 2,900 genes, roughly one in ten of all known genes. CNVs tend to be stable and inherited from the parents, although they can also arise spontaneously, and identical twins may differ in copy number variants.

Although most CNVs do not seem to be associated with disease, some are. Scherer and Wigler have both shown an association between CNVs and autism spectrum diseases, and CNVs have also been identified in schizophrenia, systemic lupus erythematosus, some cancers, and even reduced susceptibility to HIV infection. The severity of diseases such as lupus and muscular dystrophy has been linked to differences in the number of copies of the DNA sequences in question.

While the role of CNVs in disease may have driven much of the research, evolutionary biologists have also taken a keen interest, because gene duplication (and most CNVs seem to be duplications rather than deletions) "frees" genes to evolve in different directions. If an organism has but a single copy of a gene, then changes to that gene might well be harmful. If, however, there are multiple copies, then one copy can keep on performing the original function while others are under less constraint and can evolve new kinds of functionality. Many genes belong to families that could have arisen in this way.

Quite apart from their role in disease and evolution, copy number variants raise another fundamental question. If so much of the genome may differ between two individuals, much of it with no obvious consequences, what does it mean even to refer to the "normal" genome?

Commentary on the Medicine Laureates written by Jeremy Cherfas, Biology correspondent, ScienceWatch. A former editor at New Scientist, Dr. Cherfas is a science writer based in Rome, Italy.

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