

Current Comments

The 1982 NAS Fourth J.M. Luck Award for Excellence in Scientific Reviewing Goes to Victor McKusick for His Mapping of the Literature in Human Genetics

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On April 26, 1982, the National Academy of Sciences (NAS) presented the 1982 James Murray Luck Award for Excellence in Scientific Reviewing^{1,3} to Victor McKusick, William Osler professor and director of the department of medicine at Johns Hopkins University School of Medicine, and physician-in-chief at Johns Hopkins Hospital. McKusick was honored at the awards ceremony at the academy's 119th annual meeting in Washington, DC. This is the fourth year the award has been presented. I was especially glad to attend because I have known McKusick for many years. Incidentally, neither I nor ISI® was consulted about his selection by the academy committee.

The award is named for the founder of Annual Reviews Inc., James Murray Luck, who served as that organization's editor-in-chief until his retirement in 1969. Luck remains on the editorial committee of the *Annual Review of Biochemistry*, which he started in 1932. He was also present at the awards ceremony.

Jointly sponsored by ISI and Annual Reviews, the award carries a \$5,000 honorarium, and is administered by the NAS to honor outstanding authors of scientific reviews. To my knowledge, its inception in 1979 made it the first award of its kind.

The importance of well-written review articles is recognized but needs constant reiteration.⁴ I have pointed out in the past that many review articles become

milestone papers in their fields. I have even proposed that review writing be considered a profession unto itself.⁵ But until recently, reviewers rarely received formal recognition for their contributions to science. The cosponsorship of the NAS award by ISI and Annual Reviews is intended to encourage more scientists, especially younger ones, to write more and better reviews.

The field of the James Murray Luck Award rotates annually. The first award recognized an outstanding reviewer from the life sciences. G. Alan Robison, University of Texas at Houston, was honored for his series of reviews on cyclic AMP.¹ In 1980, a reviewer from the physical sciences, Conyers Herring, Stanford University, received the award for his reviews in solid-state physics, now known as condensed matter physics.² The third award recognized a reviewer in the social sciences, John S. Chipman, University of Minnesota, for his surveys of international trade theory and other aspects of economics.³

This year the award was again given to a reviewer in the life sciences. McKusick was cited by the academy for the "preparation of rigorous and comprehensive reviews which have stimulated and guided the entire field of human genetic research in both its basic and clinical aspects."

McKusick was born on a Maine dairy farm in 1921. Appropriately enough, the boy who would study genetics was an identical twin. His brother Vincent is



Victor McKusick

chief justice of the Supreme Court of the state of Maine. After attending Tufts College, McKusick entered Johns Hopkins University School of Medicine, receiving an MD in 1946. He then completed his internship and residency at the Osler Medical Clinic, Johns Hopkins Hospital.

McKusick has been a member of the faculty of the department of medicine at Johns Hopkins University School of Medicine continuously since 1947. He was appointed full professor in 1960, and was made chairman of the department in 1973. From 1957 to 1973 he served as chief of the division of medical genetics in the department of medicine, and director of the Joseph Earl Moore Clinic at Johns Hopkins Hospital. Since 1960, he has directed a course in medical genetics for medical school faculties at Bar Harbor, Maine, in conjunction with the staff of the Jackson Laboratory.

McKusick belongs to so many professional and honorary societies that we can name only a few here. He is a member of the American Society for Clinical Investigation, the Association of American Physicians, the American Medical Association, and the American Society

of Human Genetics, of which he was president in 1974. He is also a fellow of the American College of Physicians. He was elected to membership in the NAS in 1973, and the American Philosophical Society in 1974. Also in 1974, he was elected a fellow of the Royal College of Physicians of London, and a corresponding member of the National Academy of Medicine of France.

McKusick now serves as an associate editor of eight medical journals, including *American Journal of Medical Genetics*, *Journal of Chronic Diseases*, *Clinical Genetics*, and *Johns Hopkins Medical Journal*. He also is a member of the advisory board for *American Journal of Medicine*. In the past he has served as an associate editor of *Circulation*, *American Journal of Human Genetics*, and *Annals of Internal Medicine*.

McKusick's medical and scientific contributions have been mainly in medical genetics. He has authored over 600 publications. Many of these works describe and classify genetic diseases. For instance, McKusick and colleagues have described the clinical range of homocystinuria (an inborn error in metabolism),⁶ various forms of Ehlers-Danlos syndrome (an enzyme disorder affecting the joints and skin),^{7,8} and mucopolysaccharidosis (an enzyme deficiency in the connective tissues).⁹ McKusick has also performed important studies of the Old Order Amish, illustrating the dynamics of genes in inbred populations.¹⁰⁻¹² McKusick and G.A. Chase, also of Johns Hopkins University, used this latter work to explain the high frequency of Tay-Sachs disease in the Ashkenazi Jews of Eastern Europe.¹³

In addition to his work in genetics, McKusick has made important contributions to the field of cardiovascular sound. Adapting the sound spectrography method of Bell Laboratories—called spectral phonocardiography—he described heart sounds and murmurs in more detail than was previously possible.¹⁴

There is no question that the work of McKusick and his colleagues is of high impact. During the years 1961-1980, McKusick was cited over 6,700 times as a primary author. He appeared on our list of the 250 most-cited authors, 1961-1975.¹⁵ He was also one of 21 geneticists on our list of the 1,000 most-cited authors.¹⁶ He received over 2,700 citations just for articles written during the period 1965-1978.

In much of his work, McKusick combines reviewing and original research. For instance, he frequently illustrates what has previously been documented in the literature with cases he has treated at Johns Hopkins Hospital. McKusick has adopted this method in order to foster new discoveries in his field. McKusick observes, "Reviewers have an obligation to be as accurate—historically and scientifically—as possible. I would like to think, however, that my reviewing has been successful because there has been an important element of synthesis which has had thereafter a catalytic effect on the field. This is what you like to do in reviews—put different pieces of information together into a picture that is revealing to people so that they then can take the next step that is necessary for pushing the field ahead."¹⁷ A bibliogra-

phy of McKusick's reviews appears in Table 1.

I recently asked McKusick which of his own works was his favorite. He noted that Aaron Copland, the American composer, had been asked the same question. Like a parent, Copland feels he has to love all his "children," and should have no favorites among them. While McKusick also cherishes all his "children," he admits that two works especially stand out in his career.¹⁷ The first is his book *Heritable Disorders of Connective Tissue*. This monograph synthesizes the literature on genetically transmitted diseases in various connective tissues, and incorporates McKusick's extensive clinical experience. Originally published in 1956, it has gone through four editions. In 1979 it was a *Citation Classic*¹⁸ in *Current Contents*¹⁹ and it continues to be McKusick's most-cited work, having received over 1,600 citations during the period 1961-1980.

The other work which McKusick considers his most important is also a book, entitled *Mendelian Inheritance in Man: Catalog of Autosomal Dominant, Autosomal Recessive, and X-Linked Phenotypes*. This encyclopedic work is now in its fifth edition, and was cited over 1,100 times during the period 1961-1980. Both

Table 1: Selected review books and articles by Victor McKusick.

| Total Citations 1961-1980 | Bibliographic Data |
|---------------------------|--|
| 1617 | McKusick V A. <i>Heritable disorders of connective tissue</i> . St. Louis: Mosby, 1972. 878 p. |
| 1117 | McKusick V A. <i>Mendelian inheritance in man: catalog of autosomal dominant, autosomal recessive, and X-linked phenotypes</i> . Baltimore, MD: Johns Hopkins University Press, 1978. 975 p. |
| 194 | McKusick V A, Kaplan D, Wise D, Hanley W B, Suddarth S B, Sevik M F & Maumane A E. The genetic mucopolysaccharidoses. <i>Medicine</i> 44:445-83, 1965. |
| 184 | McKusick V A & Ruddle F H. The status of the gene map of the human chromosomes. <i>Science</i> 196:390-405, 1977. |
| 105 | McKusick V A. On the X chromosome in man. <i>Quart. Rev. Biol.</i> 37:69-175, 1962. |
| 22 | McKusick V A. Human genetics. <i>Annu. Rev. Genet.</i> 4:1-46, 1970. |
| 20 | McKusick V A. <i>Medical genetics 1958-1960</i> . St. Louis: Mosby, 1961. 534 p. |
| 17 | McKusick V A & Chase G A. Human genetics. <i>Annu. Rev. Genet.</i> 7:435-73, 1973. |
| 5 | McKusick V A. <i>Medical genetics 1961-1963</i> . New York: Pergamon Press, 1966. 455 p. |
| — | McKusick V A. The anatomy of the human genome. <i>J. Hered.</i> 71:370-91, 1980. |

books are, in McKusick's view, "substantially reviews, but also very important contributions" to genetics.¹⁷

McKusick's highly cited article, "The genetic mucopolysaccharidoses," also combines original research and review. In this paper, McKusick and his colleagues described five and possibly six forms of mucopolysaccharidosis on the basis of cases they had studied at Johns Hopkins Hospital. This article, which was McKusick's most-cited article in our 1,000-author study, 1965-1978,¹⁶ received over 190 citations during the period 1965-1980.

Incidentally, this 1965 article was used as evidence by Williams & Wilkins when that publishing firm sued the National Library of Medicine (NLM) for copyright infringement in 1968. Williams & Wilkins argued that McKusick's article, which appeared in a journal published by that firm, was a classic example of an item frequently photocopied without publisher permission. In fact, McKusick testified (on behalf of NLM) during the trial.¹⁷ Partly in response to this lawsuit, a new copyright law addressing the photocopy issue was enacted in 1978.¹⁹

McKusick suspects that if the award committee were asked to pick out a single review which led to his winning the award, it would be "The status of the gene map of the human chromosomes," published in *Science* in 1977 with coauthor F.H. Ruddle, Yale University.¹⁷ This article reports progress in locating specific genes on human chromosomes. A five-year update is soon to appear in *Science*. McKusick has published another review article on gene mapping in *Journal of Heredity*, called "The anatomy of the human genome."

As little as 15 years ago, human chromosomes were for the most part uncharted territory. So, too, was the vast expanse of scientific research itself. At ISI we have been locating the "genes" of scientific research, which pass on important information from one genera-

Table 2: 1981 *ISI/BIOMED*TM research front specialties in which Victor McKusick's work appears.

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| Alteration of collagen synthesis and metabolism in Ehlers-Danlos syndrome |
| Clinical measurement of plasma HDL cholesterol |
| Collagen characterization |
| Collagenase production and properties |
| Expression, regulation, movement and recombination of transformed cell lines |
| Gene related Hb diseases and thalassemia |
| Genetic studies of congenital adrenal hyperplasia hydroxylase deficiency |
| Molecular genetics of Ig complexes |
| Pathogenesis of rheumatoid arthritis and ankylosing spondylitis |
| Structure of procollagen and collagen types in related diseases |

tion of research to the next. By analyzing which papers cite the same pairs of older papers, we can identify new generations of scientific discovery, what we call "research front specialties."

We recently checked the citation data from our *ISI/BIOMED*TM online retrieval system,^{20,21} and found that McKusick has one or more papers in at least ten of our 1981 research front specialties. The names of these specialties are listed in Table 2. So besides having a powerful impact on research in genetics for over 20 years, McKusick continues to advance the frontiers of his field.

Next year's award will be presented to a reviewer from the physical sciences, including applied physics and engineering. The award is not limited to those disciplines recognized by academy membership categories. Nominations should be submitted to the Office of the Home Secretary, National Academy of Sciences 254, 2102 Constitution Avenue, Washington, DC 20418, before September 10, 1982.

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REFERENCES

1. **Garfield E.** The NAS James Murray Luck Award for Excellence in Scientific Reviewing: G. Alan Robison receives the first award for his work on cyclic AMP. *Essays of an information scientist.* Philadelphia: ISI Press, 1981. Vol. 4. p. 127-31.
2. -----, The 1980 NAS James Murray Luck Award for Excellence in Scientific Reviewing: Conyers Herring receives second award for his work in solid-state physics. *Essays of an information scientist.* Philadelphia: ISI Press, 1981. Vol. 4. p. 512-4.
3. -----, The 1981 NAS James Murray Luck Award for Excellence in Scientific Reviewing: John S. Chipman receives third award for his reviews in economics. *Current Contents* (18):5-8, 4 May 1981.
4. **Herring C.** Distill or drown: the need for reviews. *Phys. Today* 21(9):27-33, 1968.
5. **Garfield E.** Proposal for a new profession: scientific reviewer. *Essays of an information scientist.* Philadelphia: ISI Press, 1980. Vol. 3. p. 84-7.
6. **Schimke R N, McKusick V A, Huang T & Pollack A D.** Homocystinuria: studies of 20 families with 38 affected members. *J. Amer. Med. Assn.* 193:711-9, 1965.
7. **Lichtenstein J R, Martin G R, Kohn L D, Byers P H & McKusick V A.** Defect in conversion of procollagen to collagen in a form of Ehlers-Danlos syndrome. *Science* 182:298-9, 1973.
8. **Pope F M, Martin G R, Lichtenstein J R, Penttinen R, Gerson B, Rowe D W & McKusick V A.** Patients with Ehlers-Danlos syndrome type IV lack type III collagen. *Proc. Nat. Acad. Sci. US* 72:1314-6, 1975.
9. **McKusick V A, Howell R R, Hussels I E, Neufeld E F & Stevenson R E.** Allelism, non-allelism, and genetic compounds among the mucopolysaccharidoses. *Lancet* 1:993-6, 1972.
10. **McKusick V A, Hostetler J A, Egeland J A & Eldridge R.** The distribution of certain genes in the Old Order Amish. *Cold Spring Harbor Symp.* 29:99-114, 1964.
11. **McKusick V A.** Genetic studies in American inbred populations with particular reference to the Old Order Amish. *Isr. J. Med. Sci.* 9:1276-84, 1973.
12. **McKusick V A,** ed. *Medical genetic studies of the Amish: selected papers.* Baltimore, MD: Johns Hopkins University Press, 1978. 525 p.
13. **Chase G A & McKusick V A.** Letter to editor. (Controversy in human genetics: founder effect in Tay-Sachs disease.) *Amer. J. Hum. Genet.* 24:339-40, 1972.
14. **McKusick V A.** *Cardiovascular sound in health and disease.* Baltimore, MD: Williams & Wilkins, 1958. 570 p.
15. **Garfield E.** The 250 most-cited primary authors, 1961-1975. Part 1. How the names were selected. *Essays of an information scientist.* Philadelphia: ISI Press, 1980. Vol. 3. p. 326-36.
16. -----, The 1,000 contemporary scientists most-cited 1965-1978. Part 1. The basic list and introduction. *Current Contents* (41):5-14, 12 October 1981.
17. **McKusick V A.** Telephone communication. 11 March 1982.
18. -----, Citation Classic. Commentary on *Heritable disorders of connective tissue.* St. Louis: Mosby, 1972. 878 p. *Current Contents/Clinical Practice* 7(13):26, 26 March 1979.
19. **Garfield E.** The new Copyright Clearance Center and the doctrine of fair use. *Essays of an information scientist.* Philadelphia: ISI Press, 1980. Vol. 3. p. 387-92.
20. -----, ISI's on-line system makes searching so easy even a scientist can do it: introducing METADEX automatic indexing & ISI/BIOMED SEARCH. *Current Contents* (4):5-8, 26 January 1981.
21. **Institute for Scientific Information.** *Index to research fronts in ISI/BIOMED™, 1981.* Philadelphia: ISI, 1981. 205 p. \$50.00.