Victor A McKusick: From “musical murmurs” to “medical genetics”

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A self-styled medical nomad, Victor A McKusick was a clinical cardiologist who became an interventional cardiologist and then finally went on to become the doyen of medical genetics.

Introduction

Many medical students of the earlier generation must have studied the book on heart sounds and murmurs “Cardiovascular Sound in Health and Disease” during their undergraduate or postgraduate days. That authoritative and comprehensive treatise authored by Victor McKusick has played a major role in molding the knowledge bank and auscultatory skills of scores of physicians.

This article aims to remind readers of this great soul. His career illuminates several outstanding qualities a doctor is expected to possess. His efforts and success in getting a publication early in his career ought to show the way to present young medical professionals in India where research is the urgent need of the day. His versatility in excelling in two major branches of medicine is commendable. His deeds are certainly inspiring, worthy of emulation by our younger generation — medical colleagues and medical students. A noble figure in medicine indeed.

Early schooling

Victor McKusick was born in 1921. Raised on the family dairy farm, he attended a one-room school and graduated from a local high school. In his teens, he considered becoming a minister, but a close encounter with medicine changed his plans: he developed an abscess under one arm, and the infection (caused by an unusual streptococcus strain) spread and would not heal. McKusick spent 10 weeks in hospital at Massachusetts General in Boston, where he was treated successfully with the then new antibiotic sulfanilamide. Inspired by the experience, he chose to pursue a career in medicine.

Medical career

McKusick began his medical studies in 1943 at Johns Hopkins, received his MD in 1946. He joined the faculty at Johns Hopkins in 1951 as an instructor, and was promoted to professor in 1960. From 1973 to 1985, he served as Physician-in-Chief and Chairman of the Department of Medicine. From 1985, he was University Professor of Medical Genetics, and remained active in teaching and research till the end.

McKusick took his first step into medical genetics during his internship. A teenager with intestinal polyps and curious pigmented spots on his lips became his patient. McKusick had seen four other patients with this combination, three of them in the same family, indicating a hereditary condition. Hearing that a Boston physician, Harold Jeghers, had seen five such cases,
McKusick contacted him, and the two wrote up their cases for the New England Journal of Medicine. This became McKusick’s first medical publication. Because the syndrome had been noted many years earlier by Peutz, a Dutch physician, it was named the Peutz-Jeghers syndrome (PJS).

**Cardiology days**

Medical genetics was not a specialty at that time, and in his residency, McKusick was drawn to cardiology. He worked for two years in the cardiovascular unit at the U.S. Marine Hospital, doing cardiac catheterisations and studying the heart borders with a new imaging method called electrokymography.

During his early faculty years at Johns Hopkins, he pursued a study of heart sounds and murmurs using sound spectrography, which had been developed at the Bell Telephone Laboratory for studying speech sounds. The technology could pick up and record the frequency spectrum of heart sounds, allowing physicians to visualise what they were hearing with their stethoscopes. McKusick renamed it spectral phonocardiography, and used the studies as the basis for a comprehensive treatise on heart sounds titled Cardiovascular Sound in Health and Disease, published in 1958. He also published papers on chronic constrictive pericarditis, ventricular fibrillation, the electrocardiographic effects of lithium chloride, and other topics.

He carried out a comprehensive study of Marfan syndrome and four similar disorders, collecting patients and family histories from his own practice and from many other clinical departments at Johns Hopkins. This work produced his first book, Heritable Disorders of Connective Tissue, first published in 1956.

**Genetics**

Medical genetics as a distinct clinical and academic discipline at Johns Hopkins began in 1957, when McKusick was asked to serve as director of the multifaceted chronic disease clinic. McKusick accepted the position, on the condition that he could develop a Division of Medical Genetics within the Department of Medicine.

Arguing that genetic disease is the ultimate chronic disease, he envisioned the new division carrying out teaching, research, and patient care related to hereditary disorders. Increased understanding of rare genetic disorders would vastly improve differential diagnosis and treatment, and enable physicians to better counsel affected patients and their families.

His research focussed on nosology (defining the multiple distinct forms of genetic diseases) and on gene mapping. The centre became a premier post-doctoral training ground for specialists from many areas of medicine, and its alumni helped propagate the new field of medical genetics in the United States and abroad.

Soon after the medical genetics programme was founded, McKusick and his colleagues began doing comprehensive annual reviews of the relevant medical and scientific literature, which were published in the Journal of Chronic Diseases from 1958 to 1963. The catalogue, titled Mendelian Inheritance in Man, was first published in 1966, in collaboration with the National Library of Medicine, the Online Mendelian Inheritance in Man became available in 1987, and remains a standard reference work.

McKusick played a leading role on the committee charged with assessing the feasibility of what became the Human Genome Project. In 1988, he became Founding-President of the Human Genome Organisation (HUGO), an international coordinating agency for the global mapping and sequencing efforts.

**Publications**

McKusick was a prolific writer throughout his career and published over 500 medical articles and 7 books in addition to the ongoing compilation of Mendelian Inheritance in Man. Other projects included a study of haemophilia in colonial New England.

**History of medicine**

He also pursued a lifelong interest in the history of medicine. Many of his clinical publications included historical components, and he wrote a history of medical genetics for a standard textbook in the field.

In a paper in 2005, McKusick was quoted: “I have always told my students, residents, and fellows, if you want to really get on top of some topic, you need to know how it got from where it was to how it is now. I was always strong on eponyms, too – like Marfan syndrome, Freeman-Sheldon syndrome, Down syndrome, Tay-Sachs disease, etc. On rounds, the resident or student would present a patient with some particular condition, and I would always ask, so who is so and so for whom the disease was named. This prompts thought and research into the disease or condition itself to find out who first described it and, therefore, for whom it was named.”

“A historical background is an essential part of research in any field of medicine.”

– Victor McKusick.
Awards

McKusick's work earned him many honours and awards, including the Award of the American College of Physicians for distinguished contributions in internal medicine (1972); election to the National Academy of Sciences (1973); the Lasker Award for Special Achievement in Medical Science (1997); the National Medal of Science (2002), along with more than 20 honorary doctorates.

Victor McKusick died on July 22, 2008, at the age of 86.

“I became known as a cardiologist before I became known as a geneticist. Some thought I was committing professional suicide in leaving cardiology to focus on rare and “unimportant” genetic disorders. They asked why I switched from cardiology to genetics. Actually it was a matter of phasing down cardiology and ramping up genetics after 1957.”

– Victor McKusick.

References


Relaxed Deep-Breathing

Pranayama – the breathing practices of yoga – is found to shift the overall basal autonomic balance to the parasympathetic direction. In actual practice, it involves slow diaphragmatic breathing. This process reduces the adverse psycho-physiological and psychological effects of chronic stress and reactivity in stressful situations. Relaxed deep breath practices have now become a valuable component of many integrated treatment programmes. The patients with heart disease, hypertension, and asthma have found these breathing practices beneficial in stress reduction.

Deep breathing is closely related to the movement of the diaphragm – the dome-shaped muscular structure that separates the chest from the abdomen. When we breathe deeply, our diaphragm moves downward as we inhale, and upward as we exhale. The more the diaphragm moves, the more our lungs can expand. It means that more oxygen can be taken in and more carbon dioxide released with each breath.

Breathing is the rhythm of life. Pranayamic breathing exercises are more than deep-breathing practices. Pranayama allows the body to regain its natural rhythm. It thus promotes holistic healing.

Regular practise of Pranayama throws out physical and mental toxins. We can use our breath to free the mind of blocks. This leads to greater clarity of thinking. Pranayama is considered to be the highest form of purification and self-discipline for our mind.

Says Patanjali (ca B.C. 300), the Indian sage and the foremost exponent of Pranayama, “When we practice Pranayama, the veil of lethargy and ignorance is slowly taken away from the mind.”

Through Pranayama techniques, the blood stream, heart, lungs, brain, tissues, and other organs get enriched with vital energy. It brings about a change in the coordination of physical functions and mental attitudes.

The practice of Pranayama reinforces the findings of modern medical science which has come to accept the role of the mind in physical diseases. With the judicious practice of Pranayama we can attain a sound healthy body and a steady peaceful mind.

A Taoist physician from China, Shen Chai-tsu (17th century A.D) has also pointed out the benefits of breathing practices. "Breathing exercises are a hundred times more effective than any drug," says he.

The Pranayama – the relaxed deep-breathing exercise – is a definite journey to good health.

(Courtesy: Journal of the Science of Healing Outcomes; Vol. 3, No. 10)