

OBITUARY



Human Variome Project/Robert Levy

Richard Graham Hay Cotton 1940–2015

Tim Smith & Graham Taylor

Dick Cotton passed away peacefully on 14 June 2015, in Melbourne, following a stroke. He is survived by his wife, Elizabeth; two sons, James and Michael; and a daughter, Caroline.

Science needs visionaries, but it also needs team-builders who can achieve consensus across disparate groups. With his first-rate intellect and easy charm, Dick was both. His footprints can be seen across the international clinical genomics landscape, from the idea of locus-specific databases and the HGVS mutation nomenclature standards through to international data-sharing projects such as the BRCA Challenge and the Global Globin 2020 initiative. Countless geneticists and technology-oriented scientists attended his famous biannual International Mutation Detection Workshops and HUGO Mutation Detection Training Courses.

A country boy at heart, Dick often described himself as “just an orange farmer” and spent much of his spare time on his beloved family farm in Wangaratta, Victoria. He was very interested in the impact of government policy on the economics and sustainability of farming and on the environment and was an active campaigner for both. Friends and colleagues have mentioned many fond memories; for example, Al Brown (Edinburgh) remarked, “Dick introduced himself to me during the coffee break at my first HUGO MDI meeting, then asked me to give a two-minute talk about my poster immediately after coffee with just an overhead projector and a felt-tip pen!” Thus was born the “mutation database in a box” concept that enables community annotation of the human genome.

Following a PhD in genetics at the University of Melbourne, Dick worked as a post-doctoral fellow in human genetics at the John Curtin School of Medical Research, the Scripps Clinic and the Laboratory of Molecular Biology in Cambridge. After his pioneering work on cell fusion with César Milstein at the Laboratory of Molecular Biology he returned to the University of Melbourne, where as DSc and professor, he switched his attention to the challenge of identifying mutations, developing chemical and enzymatic mutation detection assays. In 1986, this work contributed to the founding of The Murdoch Childrens Research Institute at the Royal Children’s Hospital in Melbourne, a major step in introducing molecular methods to human genetic research to Australia. In 2005, he was admitted as a Member of the Order of Australia.

Dick soon realized that molecular diagnoses create a challenge for data sharing, and he therefore founded the scientific journal *Human Mutation* in 1992 and the HUGO Mutation Database Initiative (MDI) in 1994. Over time the HUGO MDI led to his establishment of the Human Genome Variation Society (HGVS), which still thrives today. Crucially,

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Dick Cotton with H3Africa PIs and Nigeria’s UNESCO Ambassador, H.E. Mrs. Mariam Y. Katagum.

the HGVS is the custodian of the nomenclature system used to describe genetic variation that is the *de facto* standard for clinical reports. Most modern variation annotation software tools have the option to output their results in HGVS format.

As well as stimulating the development of the Leiden Open Variation Database (LOVD) by Johan den Dunnen and colleagues, he motivated the establishment of the InSiGHT Database for Hereditary Colon Cancer, which is an exemplar for both the BRCA Challenge and the Global Globin 2020 international data-sharing and curation initiatives being developed by the Human Variome Project (HVP) and the Global Alliance for Genomics and Health.

Michael Katz of the March of Dimes Foundation observed that Dick recognized early on that the Human Genome Project was not the end but a beginning. He also realized that, as genetics becomes part of mainstream medical care, it will be diagnostic activities that generate the vast majority of genetic variant information. So he worked hard to build ethical and practical frameworks for the capture and sharing of diagnostic genetic output as well as the output from large-scale genomics projects.

He spent the last 30 years of his life persuading people to share his vision and what they know about genes, variants and phenotypes, culminating in the foundation of the Human Variome Project (HVP) in 2006. The HVP set out, and continues, to reach out to the developing world through the involvement of UNESCO and the World Health Organization, which have become fundamental partners in the HVP’s collaborative goals. A truly international effort to share and learn from our collective knowledge of human genetic variation will perhaps be Dick’s greatest legacy.