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Three decades of the Human Genome Organization

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Abstract

The Human Genome Organization (HUGO) was initially established in 1988 to help integrate international scientific genomic activity and to accelerate the diffusion of knowledge from the efforts of the human genome project. Its founding President was Victor McKusick. During the late 1980s and 1990s, HUGO organized lively gene mapping meetings to accurately place genes on the genome as chromosomes were being sequenced. With the completion of the Human Genome Project, HUGO went through some transitions and self-reflection. In 2020, HUGO (which hosts a large annual scientific meeting and comprises the renowned HUGO Gene Nomenclature Committee [HGNC], responsible for naming genes, and an outstanding Ethics Committee) was merged with the Human Genome Variation Society (HGVS; which defines the correct nomenclature for variation description) and the Human Variome Project (HVP; championed by the late Richard Cotton) into a single organization that is committed to assembling human genomic variation from all over the world. This consolidated effort, under a new Executive Board and seven focused committees, will facilitate efficient and effective communication and action to bring the benefits of increasing knowledge of genome diversity and biology to people all over the world.

KEYWORDS
genome biology, Human Genome Organization, HGNC, HGVS, HVP, international

1 | IN THE BEGINNING

In 1988, as the Human Genome Project was getting under way, Nobel laureate Sydney Brenner proposed the creation of an international coordinating body to help integrate scientific genomic activity and proposed the name HUGO, the Human Genome Organization. On April 30, 1988, at a meeting at Cold Spring Harbor, the proposal was endorsed and Victor McKusick was invited to be the founding President. By early September, a Founding Council had been assembled comprising of 42 scientists from 17 countries, the majority of whom met in Montreaux, Switzerland and elected Walter Bodmer, Jean Dausset, and Kenichi Matsubara as HUGO Vice Presidents, John Tooze as Secretary, and Walter Gilbert as Treasurer. Charles Cantor, Malcolm Ferguson-Smith, Leroy Hood, Lennart Philipson and Frank Ruddle served as executive committee members. A further 178 members were also chosen, bringing the original membership of HUGO to 220 scientists, drawn from 23 countries. It was to be, in the words of Norton Zinder, "a U.N. for the human genome" (Figure 1). Stylianos Antonarakis (one of the co-authors of this article) was among the initial 220 scientists that comprised HUGO!

HUGO sought to be "international and interdisciplinary" with purposes "to assist with the coordination of research on the human genome, to foster collaboration, avoiding unnecessary duplication of effort"; and also coordinate parallel studies in model organisms. HUGO also aimed to facilitate the exchange of data and biomaterials and encourage the spreading of related technologies. Finally, HUGO's

FIGURE 1 HUGO membership in early 1989. *Those scientists whose names are in bold and marked with an * are members of the founding council.* HUGO Members included: Bruce M. Alberts, USA; Richard Gelinas, USA; **Peter L. Pearson*, The Netherlands**; Philip Avner, France; Walter Goad, USA; **Elizabeth B. Robson*, UK**; Bart G. Barrell, UK; Francois Gros, France; Leon E. Rosenberg, USA; Kare Berg, Norway; John L. Hamerton, Canada; Joseph Sambrook, USA; **Walter Bodmer*, UK**; **Tasuku Honjo*, Japan**; Peter Seeburg, West Germany; David Botstein, USA; Michael Hunkapiller, USA; Louis Siminovitch, Canada; William R.A. Brown, UK; **Nancy A. Jenkins*, USA**; Mark H. Skolnick, USA; **George F. Cahill, Jr.*, USA**; Y.W. Kan, USA; Ellen Solomon, UK; Mario Capecchi, USA; Lev L. Kisselev, USA; **Grant R. Sutherland*, Australia; Webster K. Cavenee*, Canada**; Louis M. Kunkel, USA; **Glauco Tocchini-Valentini*, Italy**; Walter Gehring, Switzerland; David Patterson, USA; Stylianos E. Antonarakis, Switzerland; Mark L. Pearson, USA; Norman Arnheim, USA; Georgy P. Georgiev, USSR; Michael Ashburner, UK; Raymond F. Gesteland, USA; **Ulf Pettersson*, Sweden; Walter Gilbert*, USA;** Lennart Philipson*, West Germany; Richard Axel, USA; Richard Roberts, USA; Francisco J. Ayala, USA; Joseph L. Goldstein, USA; David Baltimore, USA; Peter N. Goodfellow, UK; Thomas H. Roderick, USA; Yoram Groner, Israel; Giovanni Romeo, Italy; Alexander A. Bayev, USSR; Hans-Hilger Ropers, The Netherlands; Arthur L. Beaudet, USA; Frank Grosveld, UK; Paul Berg, USA; **Karl-Heinz Grzeschik*, West Germany**; Janet D. Rowley, USA; James F. Gusella, USA; **Frank H. Ruddle*, USA**; Georgio Bernardi, France; Yoshiyuki Sakaki, Japan; Adrian Bird, Austria; Nicholas Hastie, UK; Frecerick R. Blattner, USA; Michael Hayden, Canada; **Bernard Hirt*, Switzerland**; David Schlessinger, USA; Lars Bolund, Denmark; Charles R. Scriver, Canada; **Piet Borst*, The Netherlands; Leroy E. Hood*, USA**; Dirk Bootsma, The Netherlands; David E. Housman, USA; Susan W. Serjeantson, Australia; Peter Humphries, Ireland; **Nobuyoshi Shimizu*, Japan; Sydney Brenner*, UK; Thomas B. Shows*, USA**; Roy J. Britten, USA; Yoji Ikawa, Japan; Michael S. Brown, USA; **Francois Jacob*, France**; Maxine F. Singer, USA; Alec J. Jeffreys, UK; Marcello Siniscalco, USA; W. Ted Brown, USA; Robert L. Sinsheimer, USA; George Brownlee, UK; Trefor Jenkins, South Africa; Gail A.P. Bruns, USA; Bertrand Jordan, France; Cassandra Smith, USA; **Fotis C. Kafatos*, Greece**; Cedric A.B. Smith, UK; Graham Cameron, West Germany; Oliver Smithies, USA; Howard M. Cann, France; Minoru Kanehisa, Japan; **Charles R. Cantor*, USA;** Haig H. Kazazian, Jr., USA; **Edwin M. Southern*, UK**; Kenneth R. Kidd, USA; Michel Steinmetz, Switzerland; C. **Thomas Caskey*, USA**; John Silston, UK; Bruce Cattanach, UK; **George Klein*, Sweden**; Luca Cavalli-Sforza, USA; Yuji Kohara, UK; Eugene D. Sverdlov, USSR; Raju S. Kucherlapati, USA; Glenys Thomson, USA; Howard Cedar, Israel; Shirley Tilghman, USA; **Pierre Chambon*, France**; Peter A. Lalley, USA; Verne M. Chapman, USA; Jean-Marc Lalouel, USA; Susumu Tonegawa, USA; George Church, USA; Eric Lander, USA; **John Tooze*, West Germany**; Daniel Cohen, France; Mark Lathrop, France; Lap-Chee Tsui, Canada; **Francis S. Collins*, USA**; David H. Ledbetter, USA; Christoper Tyler-Smith, UK; John Collins, West Germany; Philip Leder, USA; Nguyen Van Cong, France; P. Michael Conneally, USA; Hans Lehrach, UK; Herman van den Berghe, Belgium; Howard J. Cooke, UK; Leonard S. Lerman, USA; Alex van der Eb, The Netherlands; Andrew Coulson, UK; Peter Little, UK; Marvin van Dilla, USA; Charles Coutelle, East Germany; **Mary Lyon*, UK**; Gert Jan van Ommen, The Netherlands; David R. Cox, USA; Jacob V. Maizel, USA; Akiyoshi Wada, Japan; Diane W. Cox, Canada; Jean-Louis Mandel, France; Douglas C. Wallace, USA; Ian Craig, UK; Tom Maniatis, USA; Dorothy Warburton, USA; **Jean Dausset*, France; Kenichi Matsubara*, Japan**; John J. Wasmuth, USA; Kay E. Davies, UK; Allan M. Maxam, USA; **James D. Watson*, USA**; Ronald W. Davies, USA; Phyllis J. McAlpine, Canada; **David Weatherall*, UK**; Muriel Davisson, USA; **Victor A. McKusick*, USA**; Robert A. Weinberg, USA; Larry L. Deaven, USA; P. Meera Kahn, The Netherlands; Jean Weissenbach, France; Albert de la Chapelle, Finland; O.J. Miller, USA; Sherman M. Weissman, USA; Helen Donis-Keller, USA; **Andrei D. Mirzabekov*, USSR**; Charles Weissmann, Switzerland; Ford Doolittle, USA; Jan Mohr, Denmark; Raymond L. White, USA; **Renato Dulbecco*, USA**; Newton Morton, UK; Michael Wigler, USA; John H. Edwards, UK; Robert Moyzis, USA; Huntington F. Willard, Canada; Argiris Efstratiadis, USA; Daniel Nathans, USA; Robert T. Williamson, UK; H. John Evans, UK; Susumu Nishimura, Japan; Allan C. Wilson, USA; Marc Fellous, France; S. Numan, Japan; Ernst L. Winnacker, West Germany; **Malcolm A. Ferguson-Smith*, UK**; Robert L. Nussbaum, USA; Savio L.C. Woo, USA; Walter Fiers, Belgium; Stephen J. O'Brien, USA; **Ronald G. Worton*, Canada**; Uta Francke, USA; Michio Oishi, Japan; Mitsuaki Yoshida, Japan; **Jean Frezal*, France**; Maynard Olson, USA; Hans G. Zachau, West Germany; Theodore Friedmann, USA; Stuart H. Orkin, USA; **Norton D. Zinder*, USA**; Anna-Marie Frischauf, UK; Jurg Ott, USA; **Harald zur Hausen*, West Germany**; Antonio Garcia-Bellido, Spain; David C. Page, USA; Tobias Gedde-Dahl, Jr., Norway; Mary Lou Pardue, USA

purpose was also to inspire public debate and provide information and advice on the scientific, ethical, social, legal, and commercial implications of the human genome projects.

Over the years, the HUGO offices have moved around from country to country, starting from Switzerland, and then being moved to the United States, United Kingdom, Japan, Singapore, and

FIGURE 2 The Presidents of HUGO and the years of their service as president

South Korea. The current HUGO headquarters office has now returned back to the United States, in Farmington, Connecticut with a satellite European office recently moving from London to Newcastle upon Tyne in the United Kingdom.

There have been 11 presidents of HUGO. Following Victor McKusick (USA), who was the founding president of HUGO from 1988 to 1991, Walter Bodmer (UK) became the second president of HUGO from 1991 to 1993. He was followed by Thomas Caskey (USA) from 1993 to 1995; Grant Sutherland (Australia) from 1996 to 1997; Gert-Jan van Ommen (Netherlands) from 1998 to 1999; Lap-Chee Tsui (Canada) from 2000 to 2002; Yoshiyuki Sakaki (Japan) from 2002 to 2005; and Leena Peltonen (Finland) from 2005 to 2007. From 2007 to 2012, Edison Liu from the Genome Institute of Singapore led HUGO, relocating the HUGO headquarters to Singapore. Thereafter, Stylianos Antonarakis (Switzerland) served as the 10th president of HUGO from 2012 to 2017. The current president, Charles Lee (South Korea and USA), started his term in 2017 and continues to lead HUGO after being re-elected by the executive board members of "the new HUGO" (Figure 2). Sadly, we recognize the passing of several of the HUGO past presidents including Victor McKusick in 2008 (Valle & McKusick, 1921–2008), Leena Peltonen in 2010 (van Ommen, 2010), and Gert-Jan van Ommen in 2020 (Knoppers & Bovenberg, 2021).

2 | THE ANNUAL HUMAN GENE MAPPING MEETINGS

Early on, the annual HUGO meetings (Figure 3) were dedicated to human gene mapping and were referred to as the annual Human Gene Mapping Meetings (HGMs). Through the 1990s, the HGMs became a focal point for debate and learning, as the field of Medical Genomics matured. With the completion of the Human Genome Project, the HGMs evolved from a small, targeted meeting into a broader scientific conference for all genetic and genomic researchers with an excellent platform for participation from industry partners, bio-technology companies, and pharmaceutical giants. Currently, the HGMs comprise a stimulating program of plenary lectures, symposia, workshops, poster presentations, satellite meetings, and social events (e.g., gala dinners, laboratory tours, etc.) and cover a wide range of topics including system biology, epigenomics, genomic technologies, drug discovery, gene therapy, pharmacogenomics, genomic medicine, computational genomics, and bioinformatics.

The HGM2020 meeting was scheduled to be in Perth, Australia from April 5 to 8, 2020; but due to the COVID-19 pandemic, it eventually became the first and only HGM meeting to be conducted fully virtually (Forrest et al., 2020). Interestingly, there was unprecedented higher levels of participation from countries such as India, Philippines, Malaysia, Nigeria, and Sri Lanka due, in large part, to the previous inaccessibility for scientists from those countries to attend the HGM meeting. We were reminded that there are many reasons why scientists have difficulties attending scientific meetings in person including family commitments, time commitments, or lack of funding. Therefore, to align with HUGO's mission of promoting and supporting fundamental genomic research throughout the world—including those scientists from low- and middle-income countries, HUGO will strive to engage scientists from every nation and provide ways for them to actively participate in HUGO committees and meetings.

Due to the ongoing pandemic, HGM2021 has now been postponed and will occur in person in Tel Aviv on May 23–25, 2022. Accommodations will be made for scientists to also participate in the meeting virtually, although challenges still remain with such participation (e.g., being able to engage in real time with the speakers and

FIGURE 3 Destinations of the annual Human Genome Meetings (HGMs).1996—Heidelberg, Germany; 1997—Toronto, Canada; 1998—Torino, Italy; 1999—Brisbane, Australia; 2000—Vancouver, Canada; 2001—Edinburgh, Scotland; 2002—Shanghai, China; 2003—Cancun, Mexico; 2004—Berlin, Germany; 2005—Kyoto, Japan; 2006—Helsinki, Finland; 2007—Montreal, Canada; 2008—Hyderabad, India; 2010—Montpellier, France; 2011—Dubai, UAE; 2012—Sydney, Australia; 2013—Singapore, Singapore; 2014—Geneva, Switzerland; 2015—Kuala Lumpur, Malaysia; 2016—Houston, USA; 2017—Barcelona, Spain; 2018—Yokohama, Japan; 2019—Seoul, South Korea; 2020—Perth, Australia; 2022—Tel Aviv, Israel (upcoming)

audience given the time zone differences of different participants). In addition, there will undoubtedly be some activities (e.g., social events, etc.) that will have limited effectiveness in a virtual setting, as compared to being in person. Nevertheless, we believe that hybrid models will be a permanent feature for all future HGM meetings, consistent with the desires of 85% of those responding to a survey after the HGM2020 meeting (Forrest et al., 2020).

3 | THE INTERNATIONAL NATURE OF HUGO

The strength of HUGO has been, and continues to be, its international focus, particularly for the parts of the world outside of North America (which is already well represented by the American Society of Human Genetics—https://www.ashg.org) and Europe (which is already well represented by the European Society of Human Genetics—https://www.eshg.org/). More recently, HUGO has particularly served as an important venue for the gathering of human geneticists from Asian, South American, and African countries.

To promote this international position of HUGO, two prestigious international awards were established: the first are the Chen Awards dedicated to Asian-Pacific scientists and are financially supported by the Chen Foundation. The Chen Awardees were *Li Wen-Hsiung* in 2008, Academia Sinica, Taiwan; *Yusuke Nakamura* in 2010, University of Tokyo, Japan; *Ng Huck Hui* in 2010, Genome Institute of Singapore, Singapore; *Yang Huanming* in 2011, Beijing Genomics Institute (BGI), China; *Liu Jianjun* in 2011, Genome Institute of Singapore, Singapore; *John Mattick* in 2012, Garvan Institute of Medical Research, Australia; *Charles Lee* in 2012, Bringham and Women's Hospital/Harvard Medical School, USA; *Yoshihide Hayashizaki* in 2013, RIKEN, Japan; *Patrick Tan* in 2013, Genome Institute of Singapore, Singapore; *Edison Liu* in 2014, The Jackson Laboratory, USA; *Piero Carninci* in 2014, RIKEN, Japan; *Jung Wang* in 2015, Beijing Genomics Institute (BGI), China; *Bing Ren* in 2016, University of California San Diego, USA; *Feng Zhang* in 2016, Massachusetts Institute of Technology, USA; *Narry Kim* in 2017, Seoul National University, South Korea; *Aravinda Chakravarti* in 2018, Johns Hopkins University, USA; *Ami Bhatt* in 2018, Stanford University, USA; *Felix Jin Li* in 2019, Fudan University, China; *Ramanuj Dasgupta* in 2019, Genome Institute of Singapore, Singapore; *Anshul Kundaje* in 2019, Stanford University, USA.

The second is the African Prize dedicated to African scientists working in Africa. This award was introduced in 2015 during the presidency of Stylianos Antonarakis and has been financially supported by Inqaba Biotechical Industries. The African Prize awardees were *Alan Christoffels* in 2015, South African National Bioinformatics Institute (SANBI), South Africa; *Raj Ramesar* in 2016, University of Cape Town, South Africa; *Samia Temtamy* in 2017, National Research Center Egypt, Egypt; *Collen Masimirembwa* in 2018, African Institute of Biomedical Science and Technology, Zimbabwe; *Christian Happi* in 2019, Redeemer's University and Director of the World Bank funded African Center of Excellence for Genomics of Infectious Diseases (ACEGID), Nigeria. Both of these awards have greatly helped in the recognition of the efforts of scientists working in these areas and provided role models for regional genomic research with international impact.

3.1 | A new HUGO

Over the last two decades, a variety of other human genome-related organizations have emerged. Two of these, the Human Genomic Variation Society (HGVS) and the Human Variome project (HVP) have

expanded their communal interests into correctly annotating variants and gathering clinically relevant genomic variant data at a global scale, respectively.

3.1.1 | The *Human Genome Variation Society*

Initially founded by Professor Richard Cotton (Melbourne, Australia) and a group of HUGO members in 2001, the HGVS has been focused on accurate naming of genomic variants based on location and effect in the context of the genome, transcript, and protein. With an expanding understanding of the complexity of genome biology and the plethora of mechanisms and consequences of variation, HGVS has kept busy. Their work can be considered in three sections: First, Professor Johan den Dunnen (Leiden) has been updating nomenclature to describe variation as our knowledge of the genome has expanded exponentially. Their work has succeeded in creating a sustainable system that establishes and supports nomenclature standards. Second,

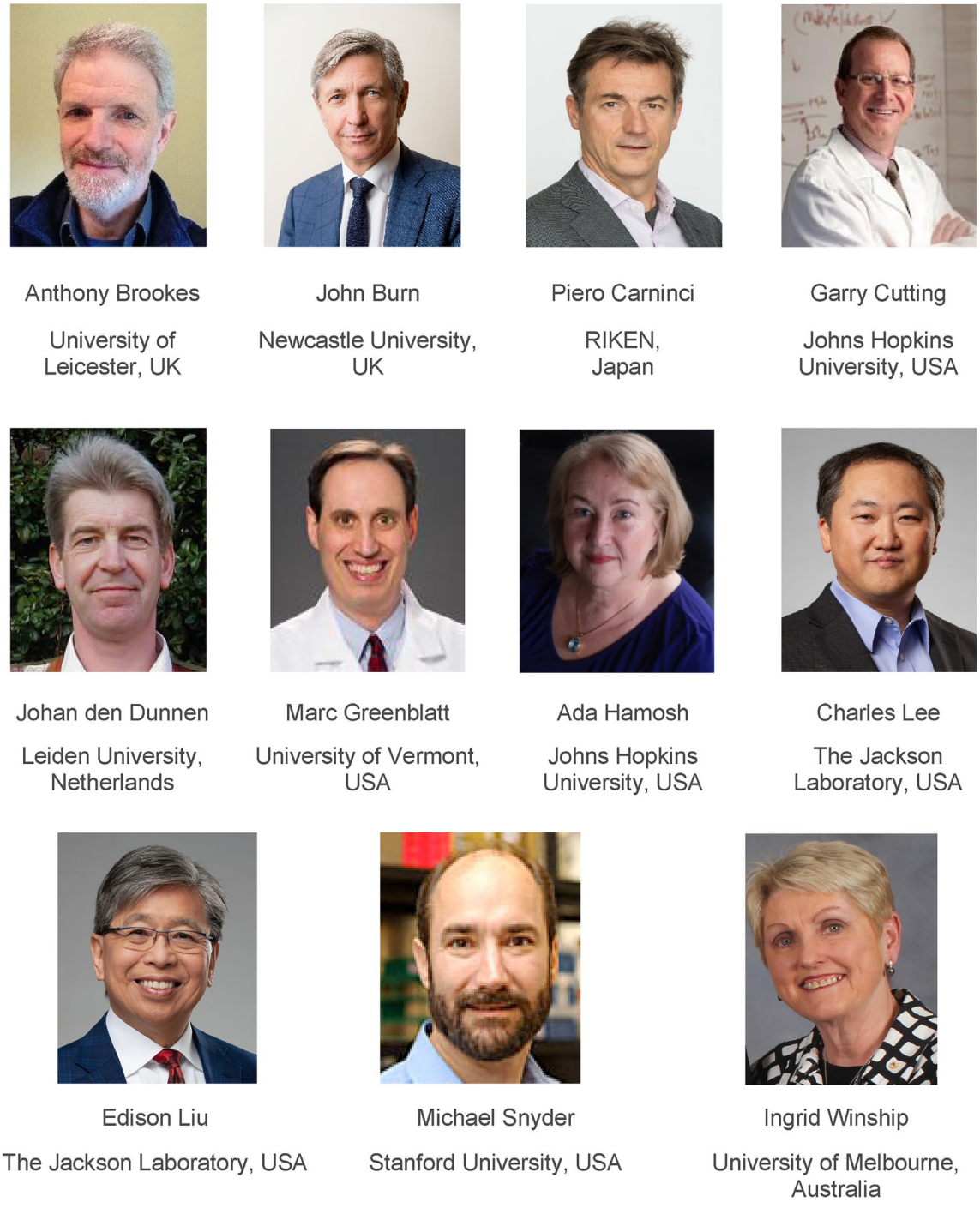

FIGURE 4 The members of the executive board of the new HUGO

HGVS has organized academic meetings addressing multiple academic aspects of variant science, especially in the areas of pathogenicity and interpretation of genetic variants. Third, HGVS has focused on promoting the sharing of genome variation data through the development of standards for databases, starting with locus specific mutation (variant) databases and expanding to national and ethnic variation databases, disease-centered databases, and others.

3.1.2 | The *Human Variome Project*

In 2006, the late Richard (Dick) Cotton, built on his experience in HUGO and HGVS to bring together leaders in genomics, from around the world, to launch the HVP which aimed to collect, curate, and distribute all human genetic variation affecting health. Working through several committees and establishing country nodes and disease- and gene- specific databases, the HVP became a UNESCO-recognized non-governmental organization (Burn & Watson, 2016; Kohonen-Corish et al., 2010). The Leiden Open Variation Database (LOVD), now in its third version, was developed by Johan den Dunnen and colleagues and has always been freely available. As an off-the-shelf database, it enables interested researchers to quickly stand up a database of gene-specific variants greatly facilitating data sharing (Fokkema et al., 2005). Following the death of Dick Cotton in 2015, the management of the HVP was transferred to the United Kingdom where it has been supported by an incorporated charity, Global Variome, overseen by Professors Ingrid Winship, Garry Cutting and John Burn as trustees. Following the emergence of the Global Alliance for Genomics and Health (GA4GH; https://www.ga4gh.org), the HVP has focused on maintaining the UNESCO link and supporting two large-scale international projects: the BRCA challenge—which was developed in partnership with the GA4GH to catalog and curate the variation in the BRCA1 and BRCA2 genes, which led to the BRCA Exchange (Cline et al., 2018) and the Global Globin Network to address worldwide variation in the alpha- (HBA) and especially beta globin (HBB) genes and modifiers and their roles in hemoglobinopathies. This effort is under the leadership of Prof. Raj Ramesar from South Africa and Prof. Zilfalil bin Alwi from Malaysia.

In 2020, it was agreed to bring HGVS and HVP together with HUGO to restore the broader base imagined by the HUGO founders. The members of the executive board of "the new HUGO" (Figure 4) have now begun to work together to bring these various activities under one umbrella.

The new HUGO currently comprises seven committees focusing on: Education, Ethics, Gene and Disease-specific Database Advisory, International Scientific Advisory, Nomenclature, Journals and Communication, and Council of Scholars. It is the hope that both *Human Mutation* and *Human Genomics* will become official journals of the merged organizations.

The ethics committee of HUGO has been well respected for many years. As advances in genome sciences will undoubtedly continue to change science and society throughout the world, an organization that can legitimately democratize the interests and access of genomic data and technologies throughout the world is of immense importance. HUGO needs to ensure true international access to genetic data as well as genomic education, irrespective of whether a country's limited resources has impeded its ability to contribute its own genomic data.

Another note is the establishment of the new Nomenclature committee which will report on the efforts of the HGVS nomenclature committee and the HUGO Gene Nomenclature Committee (HGNC). The HGVS nomenclature committee had its roots from a mutation/variant nomenclature committee established at Johns Hopkins and supported by Victor McKusick (Antonarakis, 1998; Antonarakis & McKusick, 1994). After the publication of some initial guidelines (Ad Hoc Committee on Mutation Nomenclature, 1996; Antonarakis, 1998), a formal nomenclature committee was established within HGVS under the leadership of Johan den Dunnen, and in 2000, a comprehensive set of recommendations (den Dunnen & Antonarakis, 2000) was published which is now known as the HGVS recommendations/nomenclature (http://www.HGVS.org/varnomen). These guidelines have gradually acquired worldwide acceptance and

FIGURE 5 Jennifer Lee, Operations Manager of HUGO (left panel) and Amy McAllister, Project Coordinator of HUGO (right panel)

are currently acknowledged as the standard nomenclature in molecular diagnostics (Gulley et al., 2007; Richards et al., 2015; den Dunnen et al., 2016). With respect to HGNC originally the Human Gene Nomenclature Committee (Shows et al., 1979), it was rebranded as the HUGO Gene Nomenclature Committee following HUGO's establishment in 1989. It was initially run by Dr. Phyllis J. McAlpine until 1997 and later chaired by Dr. Sue Povey (Povey et al., 2001) until 2007. Currently, HGNC (www.genenames.org) is under the leadership to Dr. Elspeth Bruford. Housed at the European Bioinformatics Institute, the work of the HGNC is essential in standardizing gene nomenclature for proper literature and database use. Updated HGNC guidelines (Bruford et al., 2020) and an accompanying editorial were recently published.

One of the new committees being established is the HUGO Council of Scholars. The HUGO Council of Scholars is intended to be comprised of distinguished investigators in genomic sciences as well as leaders in national/regional genetics and genomics efforts who are enthusiastic to actively engage in HUGO initiatives and functions. Scholars will be selected by the HUGO Executive Board and may be asked to speak at HUGO meetings, advise, or participate in specific committees, or lead in the writing of key opinion/policy papers representing HUGO.

4 | CONCLUSIONS

More than 30 years after its inception, to ensure that the benefits of the Human Genome Project accrue to all the inhabitants of our planet, HUGO is now reimagined and strengthened by bringing the HGVS and HVP back into the fold. We have no doubts that Dr. McKusick, HUGO's founding president, would have been pleased with these developments and the progress that these collaborative efforts will bring.

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DATA AVAILABILITY STATEMENT

n/a.

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