Hum Genet (2014) 133:381–382
DOI 10.1007/s00439-014-1433-5

EDITORIAL

Human Genetics' 50th Anniversary Issue

Thomas J. Hudson

Published online: 5 March 2014

The journal *Human Genetics* celebrates its 50th anniversary in 2014. Two giants in the field of human genetics, Arno Motulsky (1923–), of the University of Washington in Seattle, and Friedrich Vogel (1925–2006), of the University of Heidelberg, co-founded the journal *Humangenetik* in 1964, which was renamed *Human Genetics* in 1976. At its launch, human genetics as a science already encompassed many of the fundamental concepts of inheritance, inborn errors of metabolism, and cytogenetics (including chromosomal abnormalities causing diseases such as Down syndrome) and population genetics (Motulsky 2010). Advances in molecular genetics resulting in part from the elucidation of the structure of DNA (Watson and Crick 1953) and the development of DNA technologies propelled the growth of medical genetics to emphasize gene discovery, the understanding of gene function, and expansion of human genetics from rare monogenetic disorders to more common and complex traits and diseases.

In the decades that followed, many major milestones were achieved as a result of international collaborative projects that generated and made available massive genome datasets (International Human Genome Sequencing Consortium 2001; International HapMap Consortium 2005); 1000 Genomes Project Consortium 2010). The development of the internet and the data sharing approaches adopted by such consortia accelerated discoveries by human geneticists worldwide (Toronto International Data Release Workshop Authors 2009). Human genetics has branched out into the clinic through formal specialities such as clinical genetics and genetic counseling. Knowledge gained from understanding the underlying causes and mechanisms of disease have permeated many other clinical disciplines. Policymakers and health-care providers are challenged by the arrival of new generations of DNA-based technologies in the clinical arena (Collins and Hamburg 2013) and medicine at large is grappling with new concepts of personalized medicine (Biankin and Chanock 2011; Dancey et al. 2012; Tran et al. 2013).

Over the past half-century, *Human Genetics* has been a pillar of the human genetics community. The scope of the journal has remained broad and includes articles on gene structure and function, genome-wide linkage and association studies in families and populations. Articles published by the journal have always included the latest conceptual and technological advances. The journal editors have shaped the scope of the journal to enhance submissions of articles that address clinically relevant questions or which provide new insights into human biology, including developmental genetics and complex diseases. In recent years, the journal has deliberately sought out new communities including bioinformatics and humanities such as ethics, law and sociology; the latter examples reflecting the growing requirement that human genetics be applied responsibly. An ever-growing rate of submissions to the journal, the evolution from a more European-centric author base to a worldwide community of scholars (for example, papers from Asia accounted for 24 % of all papers accepted in 2012) and a desire to maintain a high standard of excellence has led to a decrease in the acceptance rate (21 % in 2012). In recent years, the journal has created a venue for comprehensive *Special Issues* containing state-of-the-art

T. J. Hudson (✉)
Ontario Institute for Cancer Research, MaRS Centre, 661 University Avenue, Suite 510, 101, College Street, Toronto, ON M5G 0A3, Canada
e-mail: tom.hudson@oicr.on.ca

T. J. Hudson
Departments of Molecular Genetics and Medical Biophysics, University of Toronto, Toronto, ON, Canada

reviews and perspectives in rapidly evolving areas such as the genetics of neurological and psychiatric diseases and traits (2009), personalized medicine (2011), X-inactivation (2011), biobanking (2011), genetics of substance abuse disorders and addiction (2012), genetic epidemiology (2012), and functional characterization of regulatory elements in the human genome (expected in 2014).

On this occasion, Springer has invited authors of well-cited articles published in *Human Genetics* to contribute to this Anniversary Issue. The topics cover areas of human genetics that were important both when the original articles were published and now. These include mechanisms of mutagenesis (Mussotter et al. 2014), cytogenetics (Cremer et al. 2014; Hook and Warburton 2014), statistical genetics (Knecht and Krawczak 2014), population genetics (Kidd et al. 2014), aging (Horan and Cooper 2014), psychiatric traits (Gelernter 2014), and cancer (Wojcicka et al. 2014).

There is little doubt that human genetics will continue to flourish as a field for decades to come. With this in mind, the editors and publishers of *Human Genetics* offer their best wishes to past, present and future authors, reviewers and readers of the journal. Happy 50th Anniversary!

Acknowledgments T.J. H. is the co-editor-in-chief (Americas) of *Human Genetics*. T.J.H. is the recipient of a Senior Investigator Award from the Ontario Institute for Cancer Research, through generous support from the Ontario Ministry of Research and Innovation.

References

1000 Genomes Project Consortium (2010) A map of human genome variation from population-scale sequencing. Nature 2010(467):1061–1073. doi:10.1038/nature09534 (PMID: 20981092)

Biankin AV, Chanock SJ (2011) The road ahead: less travelled and more arduous than initially envisioned. Hum Genet 130:1–2. doi:10.1007/s00439-011-1046-1 (PMID: 21713432)

Collins FS, Hamburg MA (2013) First FDA authorization for next-generation sequencer. N Engl J Med 369:2369–2371. doi:10.1056/NEJMp1314561 (PMID: 24251383)

Cremer T, Cremer C, Lichter P (2014) Recollections of a scientific journey published in human genetics: from chromosome territories to interphase cytogenetics and comparative genome hybridization. Hum Genet. doi:10.1007/s00439-014-1425-5 (PMID: 24504674)

Dancey JE, Bedard PL, Onetto N, Hudson TJ (2012) The genetic basis for cancer treatment decisions. Cell 148:409–420. doi:10.1016/j.cell.2012.01.014 (PMID: 22304912)

Gelernter J (2014) *SLC6A4* polymorphism, population genetics, and psychiatric traits. Hum Genet. doi:10.1007/s00439-013-1412-2 (PMID: 24385047)

Hook EB, Warburton D (2014) Turner syndrome revisited: review of new data supports the hypothesis that all viable 45, X cases are cryptic mosaics with a rescue cell line, implying an origin by mitotic loss. Hum Genet. doi:10.1007/s00439-014-1420-x (PMID: 24477775)

Horan MP, Cooper DN (2014) The emergence of the mitochondrial genome as a partial regulator of nuclear function is providing new insights into the genetic mechanisms underlying age-related complex disease. Hum Genet. doi:10.1007/s00439-013-1402-4 (PMID: 24305784)

International HapMap Consortium (2005) A haplotype map of the human genome. Nature 437:1299–1320 (PMID: 16255080)

International Human Genome Sequencing Consortium (2001) Initial sequencing and analysis of the human genome. Nature 409:860–921 (PMID: 11237011)

Kidd KK, Pakstis AJ, Yun L (2014) An historical perspective on "The world-wide distribution of allele frequencies at the human dopamine D4 receptor locus". Hum Genet. doi:10.1007/s00439-013-1386-0 (PMID: 24162668)

Knecht C, Krawczak M (2014) Molecular genetic epidemiology of human diseases: from patterns to predictions. Hum Genet. doi:10.1007/s00439-013-1396-y (PMID: 24241280)

Motulsky AG (2010) History of human genetics. In: Speicher MR et al (eds) Vogel and Motulsky's human genetics: problems and approaches. Springer, Berlin. doi:10.1007/978-3-540-37654-5_2

Mussotter T, Bengesser K, Högel J, Cooper DN, Kehrer-Sawatzki H (2014) Population-specific differences in gene conversion patterns between human *SUZ12* and *SUZ12P* are indicative of the dynamic nature of interparalog gene conversion. Hum Genet. doi:10.1007/s00439-013-1410-4 (PMID: 24385046)

Toronto International Data Release Workshop Authors (2009) Prepublication data sharing. Nature 461:168–170. doi:10.1038/461168a (PMID: 19741685)

Tran B, Brown AM, Bedard PL, Winquist E, Goss GD, Hotte SJ, Welch SA, Hirte HW, Zhang T, Stein LD, Ferretti V, Watt S, Jiao W, Ng K, Ghai S, Shaw P, Petrocelli T, Hudson TJ, Neel BG, Onetto N, Siu LL, McPherson JD, Kamel-Reid S, Dancey JE (2013) Feasibility of real time next generation sequencing of cancer genes linked to drug response: results from a clinical trial. Int J Cancer 132:1547–1555. doi:10.1002/ijc.27817

Watson JD, Crick FH (1953) The structure of DNA. Cold Spring Harb Symp Quant Biol 18:123–131

Wojcicka A, de la Chapelle A, Jazdzewski K (2014) MicroRNA-related sequence variations in human cancers. Hum Genet. doi:10.1007/s00439-013-1397-x (PMID: 24241281)