

ASHG: Continually advancing genetic discoveries

Joseph D. McInerney is the current Executive Vice President of the American Society of Human Genetics – a long established organisation which provides expertise and a base for scientists in the field of genetic research. He recently sat down with us at *Research Features* to discuss the work of the organisation and to outline the hopes and concerns he has for future genetic research.

From Watson and Crick's uncovering of DNA's double helix structure in 1953, to the initial use of gene editing in 1987, to the completion of the human genome project 16 years later in 2003 – genetics is an area of continual scientific excitement and discovery.

Its research has helped to determine cures and treatments for a number of different genetic disorders and its application is now utilised throughout the world of scientific research. As such, it is vital for scientists to work collaboratively – sharing ideas, data and their research findings.

With this in mind, the American Society of Human Genetics was first established back in 1948 – five years prior to Watson and Crick's ground-breaking discovery. *Research Features* spoke to Joe McInerney, their current Executive Vice President, to discuss the organisation's heritage and ascertain how the landscape of genetics research has changed since its inception.

What does your role as Executive Vice President of the American Society of Human Genetics (ASHG) involve?

I became a member of the society in 1980 and it has been my major professional home ever since. In 2013, I took on the role of ASHG Executive Vice President, and now I get to see the organisation operate from the inside, which I was not able to do as a member.

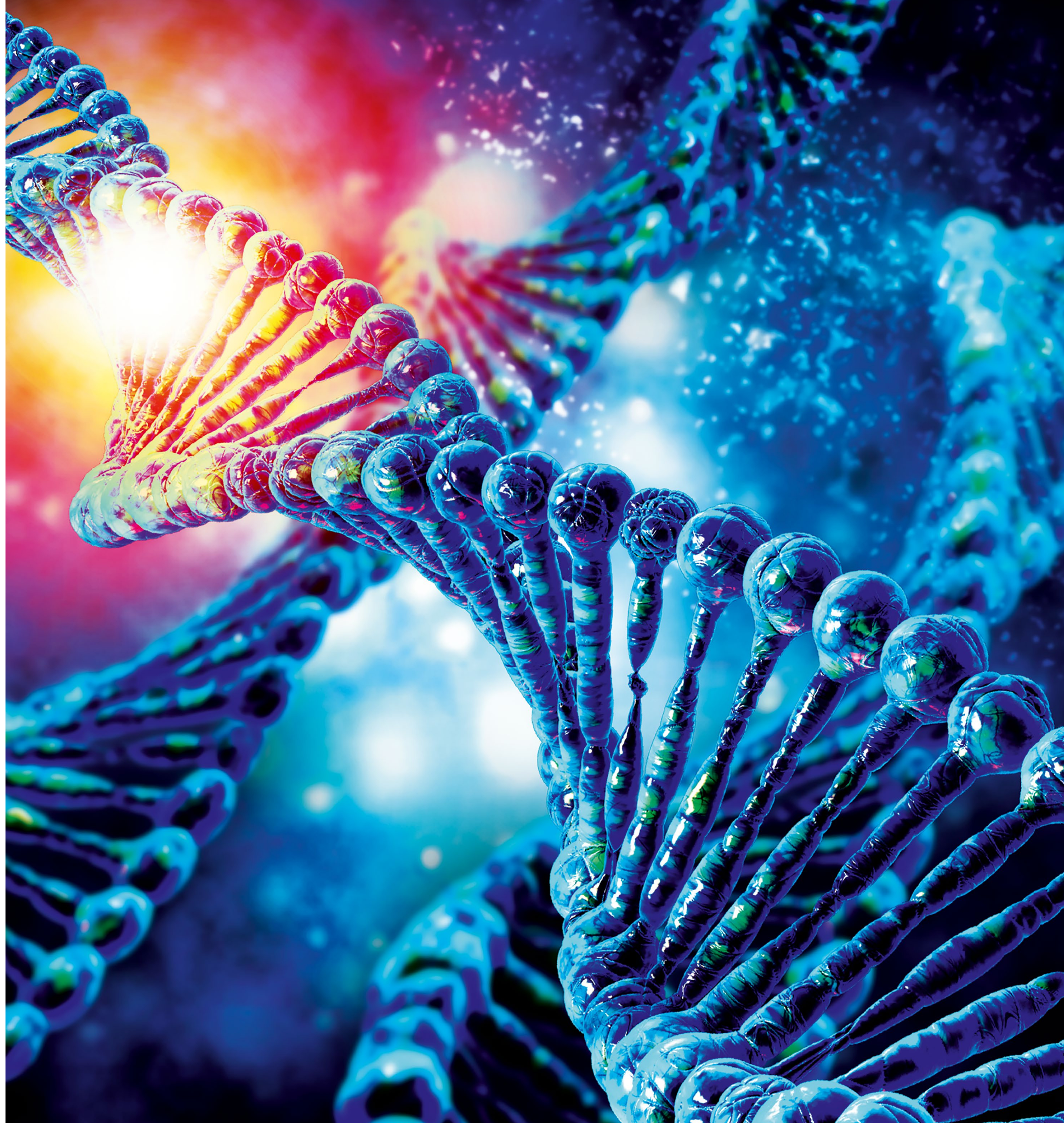
Could you tell us about the ASHG's background and the aims of the society?

The ASHG was established in 1948 and our first president was Nobel Laureate, Hermann Muller. The intent at the time was to create a forum where like-minded people could communicate with one another and support one another, and to help direct the growth of the field.

The formal mission of the society is to advance human genetics in science, health and society through excellence in research, education and advocacy, and our tagline derives from that. The tagline is 'Discover, Educate, Advocate.' Our membership is international. We have around 8,000 members and we have an increasing number of collaborations with other societies, including international societies such as the European Society of Human Genetics. ASHG is the largest professional genetics society in the world.

What impact do you think the ASHG has had on human genetics research since it was first established in 1948?

It is hard to quantify our contribution, but I think it is very safe to say that our members have been involved in the creation of much of the new knowledge in the field and have been responsible for many of the central advances, including much of the science and technology related to genome science and to investigations of the relationship between genetics and health and disease. ►





Senator James Cowan of Canada, recipient of ASHG's 2016 Advocacy Award, chats with meeting attendees about his efforts to prevent genetic discrimination

Our members have been working for a very long time on population genetics and human evolution, human history, the architecture of the human population. Our members helped to establish the first three departments in medical genetics in the United States in the 1950s, one at Johns Hopkins University, one at the University of Michigan and one at the University of Washington.

Are there any of the organisation's accomplishments that you are particularly proud of?

There are many. One is our high quality journal and the role it plays in the dissemination of cutting edge research around the world. There is our meeting too, which is another forum for the dissemination of research and the exchange of ideas, and for networking and building collaborations. The meeting is really quite impressive, it is the largest genetics

meeting in the world, each year attracting between 7,000 and 8,000 people.

I think the emphasis we have placed on our trainees, our young people, our doctoral students, our postdocs, and graduate students is another important accomplishment. As the career landscape changes significantly, many of our young people indicate that they do not see themselves moving into academic positions and are looking for opportunities elsewhere, including increasingly in industry, and we have done our best to support our trainees when they are facing those kinds of decisions. I am really proud of our role in policy and advocacy, and in education. We have very strong education programmes here at a variety of levels. We also have a very good policy and advocacy history. I am proud of the role we played in the case of *Myriad vs the*

Association for Molecular Pathology. The case, addressed by the Supreme Court in 2013, resulted in the barring of patents on human genes and it helped to make BRCA testing, a test for susceptibility to breast cancer, more widely available.

Very significant too was our support for the *Genetic Information Nondiscrimination Act*, which prevents the use of genetic information in decisions related to health insurance and employment.

You mentioned that a lot of young people would rather go into industry than academia. Is that a problem for academia?

I do not think it is a problem for academia, except in the sense that I think people in academia have to begin to think a little bit differently about the kind of training and mentorship they are providing to young scientists. If more and more young scientists are not going to move into the academic setting, then there are a number of reasons for that which need to be considered. I think much of it has to do with funding opportunities and the fact that young people see themselves doing a five-year PhD and then two three-year postdocs, and they are facing the prospect of an ever more competitive funding climate

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ASHG is one of 30 scientific societies which belong to FASEB and is headquartered at the FASEB campus in Bethesda, Maryland

and they might not end up getting their first R01 grants, here in the US, from NIH (Research Project Grants for health-related research and development) until their late 30s. I think more and more young people are saying that they are not willing to sign up for that path, and they are looking for opportunities elsewhere. Not everyone, but certainly an increasingly large number of trainees do not see themselves going into the academic setting and I do not think that is necessarily a bad thing, but it certainly presents a new set of challenges for those of us involved in training and mentoring young people.

What are your goals for the ASHG while you serve as the society's Executive Vice President?

My first overarching goal is to make sure that we as a society remain relevant to all segments of the field as it undergoes rapid growth and specialisation. I want to make sure that we remain the 'big tent' for all genetics individuals, including those who are going into highly specialised areas of genetics and also those who may not consider themselves geneticists, but who certainly are using genetic and genomic tools as they investigate important questions in basic biology, and in health and disease.

The National Institutes of Health (NIH) here has more than 30 institutes and centres, and there is only one that is formally called the National Human Genome Research Institute and has genetics in the title, but virtually every one of those institutes is involved in genetics research in one way or another. The people in those institutes might not see themselves

as geneticists. They may be developmental biologists, cardiologists, specialists in addiction, specialists in ageing, for example, but they are all using genetic tools and I want to make sure that those individuals see the American Society of Human Genetics as a potential home for them to collaborate with individuals who are formally trained in research.

I am trying to find ways to keep our young people engaged as well, to ensure that they remain members of the society once they have completed their training. We have a lot of programmes underway for trainees and we recently instituted a new membership category for early career investigators, because we know that as people make the transition from formal training into their first professional positions their finances are constrained.

Finally, trying to make genetics attractive to under-represented minorities is an ongoing, major challenge, not only for us, but for all of science.

You have already mentioned the ASHG's Annual Meeting. What else does the ASHG do to encourage interaction among human genetics professionals?

We continue to work on building collaborations with other groups. For example, we have joined symposia with other societies. We began this with a session that we call *Building Bridges*, with the European Society. We also have joint memberships with the European Society of Human Genetics. Increasingly, we are initiating collaborations

with organisations that are not formally genetics groups. For example, we are now in the middle of a collaboration with the American Physiological Society, one of the oldest professional societies in the United States, founded in the late 1800s. We have just been working with them on a collaborative symposium on cardiovascular genetics.

We also are working on collaborations with some of the Asian genetics societies and we are active in the International Federation of Human Genetics Societies as well. We are also increasingly encouraging collaborations between genetics professionals and those from non-genetics groups.

What are key considerations in the formation of science policy, and what involvement does the ASHG have in this field?

My colleagues who are heavily involved in science policy have said to me over the years that there are a few questions to ask upfront if you are thinking about the determination of new policy. Firstly, you need to consider whether the situation is truly in need of a policy solution. Secondly, you need to ask if there is an appropriate policy solution that one could construct that would be acceptable to the various stakeholders. There is the classic example here in the United States with prohibition - the policy was an abject failure because it was not acceptable to the various stakeholders, that is, those of us who like to drink Irish whisky. I am being a little glib here, but you can come up with examples in biology and medicine as well.

Another important issue in thinking about policy is, is the policy enforceable? Do you actually have the mechanisms to ensure that the policy works and that the people it is intended to influence abide by it and obey it? When policy-related issues come to our attention, we apply those criteria and others and we then ask ourselves whether we can make a helpful and credible contribution given the expertise available to us. We also look at what is important to our members. Are there issues that are important to the members where we can help promote their interests from an advocacy stand point? Things like making policy statements about the importance of research funding, access to reproductive services, those kinds of things.

Our policy staff have developed a new policy platform that is now available on our website and it lays out the broad areas where we will be attending to policy-related issues. These

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are the conduct of genetic research, the use of genetics in health care, the use of genetics in society, and then issues related to genetic literacy across the board.

Research on the human genome generates some important ethical questions. How does the ASHG go about identifying and addressing these issues?

These kinds of issues come to us in a variety of ways, but generally they emerge from the advances in science and technology that often become evident to our members first, usually before they get to the general public. Sometimes we can see them coming. For example, if you were paying attention to what was happening in genetics in the 1970s and 80s, you could see that the growth of prenatal diagnosis at that time was going to raise some important ethical issues. Things like prenatal diagnosis for sex election and therapeutic abortion in the case of genetic diagnosis.

A while back, people who were working in ethics came up with a term that I really like, 'anticipatory ethics'. It is about trying to stay ahead of the curve and anticipate what is coming down the pipe, anticipate what the issues are, anticipate the groups that might be affected, who the stakeholders are, who has the requisite expertise to help us through these issues.

Sometimes you can see what is coming and hope to prepare for it, but sometimes we are taken a little bit by surprise, as with the rapid expansion of gene-editing technology over the last couple of years, and its potential application to germ line genome editing (a technique to modify reproductive cells). We are currently working on a policy statement about that.

Another example was genetic testing in children, in the wake of whole genome sequencing and whole exome sequencing. These types of sequencing will reveal genetic variants related to disorders that clinicians would not generally test for in children and

adolescents. That landscape is changing considerably because of the availability of this type of testing, and so we were obliged to go back and revisit a statement that we had developed a number of years ago, to update our own thinking about genetic testing in children and adolescents in the wake of new technology. We try to make informed judgements about these ethical questions and consider where the expertise of our members can be most helpful. There are a lot of issues out there and we cannot take all of them on, so it is also important to identify those that are most in need of our attention.

Stories about designer babies and other headline-grabbing portrayals of the impact of genetic engineering appear from time to time in the popular press. What steps does the ASHG take to reframe public debate on genetic advances?

We see our role as helping to ensure that the treatment of genetics is reality-based. A lot of the things we hear about designer babies are simply not possible. So we try to work with the press to say what is possible and what is not possible. The notion at the moment, that you can make a series of decisions and apply a series of genetic tools that will ensure you have a child with any particular trait, like enhanced intelligence – that is simply not reality. So we work with the press to reframe what the public debate should really be about, and to keep it reality-based.

One of the aims of the ASHG in promoting the science of human genetics is to encourage its application for the common good, how does it go about this?

The intent to increase our store of basic knowledge – I think that is an essential benefit to humanity. Even just knowing more is good, and then we promote the responsible application of human genetics in personal and public health through our policy and advocacy efforts. We also have many different educational activities to help with the appropriate integration of genetics into the public sphere, into public thinking, and also



Joseph McInerney (right) discusses ASHG programmes with Dr Hudson Freeze, ASHG member and FASEB president, at ASHG's exhibit booth at the Society's 2016 annual meeting, in Vancouver

into application. We have educational programmes ranging from high school up to education for practising health care providers, to help ensure the appropriate integration of genetics into mainstream health care.

What developments do you see happening in the field of genetic research over the next ten years?

I have to be careful here because we in the genetics community have been criticised for trying to over sell the fruits of genetic research, especially where personal and public health are concerned, even though we have made good progress in that field. But I will talk a little bit about what might be coming down the road.

Certainly our Precision Medicine Initiative here in the United States, which was an initiative of our former president Barack Obama, will continue. It is a research project, created in 2015, with the aim of advancing tailored medical care for the individual. As part of the project, genetic and health data is being collected from one million people. Genetic and genomic tools will be employed to investigate the distribution of disease in society and the association of identified genetic variance with predisposition to disease, and to provide insights into treatment and prevention.

Certainly there will be an ongoing consideration in the future of the relationship between one's genes and one's environment. I also think we will see a greater involvement of global populations in genetics research to ensure that our research truly reflects the variation that is out there around the world. I think you will see insight into common complex diseases that are major causes of mortality and morbidity around the world – cancer, heart disease, diabetes, psychiatric illness – I expect there to be continued insights into those maladies.

I am also sure you will see a continued focus on how we manage big data. The Human Genome Project – a research programme which aimed to map and understand all the genes of human beings – has received a lot of attention. However, the scientific community has sequenced the genomes of thousands of other species now, and so there is an enormous avalanche of genomic data that we have to figure out how to manage appropriately. Not only how to share the data effectively, but how to mine it in the most appropriate way to answer the questions we want to ask. So you will see lots of focus on computational biology and the management of big data. We never know where the next big technological breakthroughs will occur, but we will certainly see continued investigation into gene regulation and expression.

A further major area of activity is the continued exploration of the genetic architecture of our species, its population history. What does the species look like from a population genetics standpoint? And of course that provides insights into our own history, into human evolution. How we came to be what we are. Those are some of the things I see happening in the next ten years.

Before taking up your current post at the ASHG you were the recipient of the society's Excellence in Education Award in 2005. This was in partial recognition of the key role you played in keeping evolution in high school biology texts, which you carried out in the face of strong opposition from those trying to introduce creationism into high school science teaching. Why was it so important to you personally to stand up for science generally, and science education in particular?

I am very passionate about this. I have been involved in the evolution–creation issue for more than 30 years. For me the heart of this issue is the recognition that science is really a unique and powerful way of asking questions and seeking answers, and we do significant damage to science when individuals or groups try to impose non-scientific ways of thinking about the process for reasons of ideology, whether the ideology is religious or political.

There is a philosopher and neuroscientist, Sam Harris, who wrote not long ago that the core of science is not controlled experiments or mathematical modelling, it is intellectual honesty. Creationism and other pseudosciences are simply intellectually dishonest, and they confuse people and politicians, often with false equivalents. For example, there are not two co-equal sides in the evolution–creation debate. One side has rigorously vetted evidence, derived from the well established methods of science. The other has revealed knowledge, often from scripture, that is completely bereft of scientific evidence and requires faith-based acceptance. These are not equal ways of looking at the world. They do not produce knowledge of equal value when one is thinking about the natural world.

Science education in schools is the only place where most of the public encounters the methods and habits of mind of scientific enquiry, and gets an exposure to a thoroughgoing scepticism, the notion that we should question everything, and I think we need to protect that educational process from ideologically-driven challenges.

• *If you would like to find any more information about the American Society of Human Genetics, please visit their website at <http://www.ashg.org/>.*



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