

## GENETICS

## Deep Inside Champions, Just Genes?

Dov Greenbaum, Jieming Chen, Mark Gerstein

In *The Sports Gene*, David Epstein provides a well-researched collection of interesting anecdotal cases related to the long-standing nature-versus-nurture debate. These lean heavily toward track and field. In addition to Epstein's personal experiences as a former collegiate runner, track potentially provides a more direct connection between genetics and basic physical phenotypes (such as strength, speed, and endurance) than other sports in which confounding factors such as team play may make for a more complex relationship.

The book sets out to present a balanced account of the debate. Epstein (a science writer at *Sports Illustrated*) argues that genetic influences are not necessarily uniform across a sport. Rather, different influences come into play at different points over an athlete's career. Gender differences provide an example of his nuanced treatment. Notwithstanding seemingly hard-wired differences (due to, among other influences, variation in testosterone levels), some had predicted, given historical trajectories, that women's athletic abilities would eventually converge on men's (1, 2). Epstein counters that—because current data suggest that female athletic abilities are plateauing below those of men—earlier trends might have been confounded by doping during the Cold War.

Epstein also discusses a group of female athletes who have naturally higher levels of testosterone (possibly enhancing their performances): those who are genetically XY. However, the devil is in the biological details: some XY females are insensitive to the effects of increased testosterone, and, Epstein argues, these athletes are sometimes unfairly disqualified from competing against XX-female colleagues.

As evidenced throughout the book, athletes make excellent research subjects. Their physical traits are carefully measured and recorded. Moreover, society treats the pri-

vacancy of their personal data as less sacrosanct than legally protected medical records. This enables researchers to more readily link substantial amounts of quantifiable physiological measurements, physical abilities inherent from genetics, and the underlying mechanistic biology.

Also, with the near-universal pop-cultural qualities of sport, discussing genetics in athletics provides an optimal vehicle for teaching basic biological concepts to a broad audience. It's much more palatable to learn about Usain Bolt's genes than about the depressing phenotype of a rare genetic disorder.

### The Sports Gene

Inside the Science of Extraordinary Athletic Performance/What Makes the Perfect Athlete

by David Epstein

Current, New York, 2013.

352 pp. \$26.95. ISBN

9781591845119. Yellow

Jersey, London. £16.99.

ISBN 978-0224091619.

where, Epstein explains what supermuscular babies, beefy cows, and incredibly fast whippets have taught us about the function of the myostatin gene. He also describes loss-of-function variants in the *ACTN3* gene and how these are related to the contrast between top-notch endurance runners (particularly those from East Africa) and exceptionally fast sprinters (from West Africa).

The erythropoietin receptor, myostatin, and *ACTN3* provide classic single-gene stories. Despite the obvious constraints on books written for a broad audience, we would have appreciated more on how sports relates to current efforts in genome-wide studies. In particular, personal genomics provides a new way for athletes to fully grapple with the totality of their underlying nature. One can imagine athletes devising customized training regimens using these increasingly affordable



**Why so fast?** Usain Bolt on his way to a world's record in the 100-m finals at the 2008 Olympics in Beijing.

In addition, sports, like genetics, is concerned fundamentally with outliers. Rare mutations with strong effects are nature's gift to genetics in that they can sometimes illuminate the underlying biochemical mechanisms. In a similar vein, the never-ending search for exceptional talent that will lead to victory in highly competitive sports effectively picks out individuals in the upper end of the athletic bell curve (with those rare mutations that genetics seeks).

Epstein provides many instances of identifiable outliers (even within the rarified context of world-class performers) and describes their connections to genetics. For example, we learn about a Finnish skier whose rare mutation in the erythropoietin receptor gave him hemoglobin levels so high that he was suspected of doping. Else-

and available data. These data may prove especially useful for avoiding athletic injuries. Epstein, for instance, discusses a number of specific genes that can provide some light on injuries, including the *ApoE* gene (where the  $\epsilon 4$  allele worsens the prognosis after a concussion) and the genes associated with hypertrophic cardiomyopathy.

Akin to personal genomics in its emphasis on building databases of personal information, the quantified-self movement focuses on taking real-time measurements of individuals' physiology using smart sensors. Sports training will be revolutionized by these measurements, which offer a much more precise view of an athlete's dynamic physiology throughout the day and over the course of training. The merger of personal genomics and quantified-self measurement

The reviewers are at the Program in Computational Biology and Bioinformatics and the Department of Molecular Biophysics and Biochemistry, Yale University, New Haven, CT 06520, USA. E-mail: dov.greenbaum@yale.edu; mark@gersteinlab.org

could culminate in real-time “personal functional genomics” in which large-scale biochemical assays, such as RNA sequencing, help athletes prepare for competition. The Personal Genomes Project has already taken steps to make shareable collections of such data sets possible (3), and one researcher has already integrated his genome with his transcript, protein, and metabolite measurements to chart his physiological states over time (4).

Through Epstein’s easily accessible presentation, supported by comprehensive and copious notes, *The Sports Gene* provides a wonderful background to a future in which emerging technologies will bring the nature-versus-nurture debate into even sharper focus. Perhaps in this future, the author will write an equally compelling sequel: “Sports Genomics.”

#### References

1. B. J. Whipp, S. A. Ward, *Nature* **355**, 25 (1992).
2. A. J. Tatem, C. A. Guerra, P. M. Atkinson, S. I. Hay, *Nature* **431**, 525 (2004).
3. www.personalgenomes.org.
4. R. Chen et al., *Cell* **148**, 1293 (2012).

10.1126/science.1245795

## PUBLIC HEALTH

# An Ethnographic Check-Up

Nicole S. Berry

The field of global health has undergone a profound transformation over the past 15 years. The amount of money earmarked as “global assistance for health” has increased fourfold from 1990 levels (1). The majority of this money previously streamed from government treasuries to international organizations (e.g., the World Health Organization or the World Bank). Yet, the past decade has brought substantial increases in funding from private philanthropists (e.g., the Bill and Melinda Gates Foundation); a redirection of state monies toward new players, such as public-private partnerships (e.g., The Global Fund); and a superfluity of nongovernmental organizations. What was a relatively staid field at the end of the Cold War has now been reworked into a fresh field with new players, emphases, and modes of engagement (2).

The reviewer is in the Faculty of Health Sciences, Simon Fraser University, 8888 University Drive, Burnaby, BC V5A 1S6, Canada. E-mail: nicole\_berry@sfu.ca

Given these recent transformations, *When People Come First: Critical Studies in Global Health* is a welcome examination of “the actual impacts of [global health] initiatives on care, health systems, and governance.” For the authors, considering people first articulates with a theoretical commitment to analyze the lived experiences and knowledges of those whom the enterprise of global health seeks to help. Indeed, the authors argue that to meet our obligations to people, within this domain such knowledges are as crucial as “gold standard” evidence. On an applied level, the authors align themselves with recent work by economist Michael Porter (3), anthropologist (and World Bank president) Jim Yong Kim, and physician-anthropologist Paul Farmer (4) to emphasize that putting people first means shifting from a focus on program success (e.g., access, patient compliance, or deliverables) to “results obtained by the patients (measured in survival rates and in the degree and sustainability of recovery).”

The editors—anthropologists João Biehl (Princeton University) and Adriana Petryna (University of Pennsylvania)—showcase work by scholars who wade into the new territory of global health concerned that the programs, paradigms, and interventions that dot the world’s landscape frequently do little to alleviate the profound suffering and ill health of marginalized and vulnerable communities they are meant to help. The volume draws on ethnography-based case studies that intentionally pull in social, economic, and political context to push the reader to think critically about the assumptions that underlie practices that epitomize contemporary global health. These include evidence-based public health (Vincanne Adams), reliance on nongovernmental organizations to distribute money (James Pfeiffer), public-private mixes (Stefan Ecks and Ian Harper), and attention to compliance (Ian Whitmarsh).

The volume is divided into three main thematic sections. The essays about “evidence” point to a current propensity to seek improved health outcomes by streamlining approaches—e.g., only admitting certain types of evidence, searching for magic bullets, and favoring technological fixes. The authors argue that aspects that get bracketed out of these approaches—such as public health histories (Marcos Cueto) and the complexities of “clients’” experiences (Adams)—are central to actually trans-

forming health outcomes. The second section offers a more fine-grained attention to global health interventions. Following the impetus of the volume’s title, these chapters explore the complex lives of people and consider how global health interventions designed to help intersect these lives only in limited ways. These essays demonstrate the partiality of what we seek to achieve as well as what we have achieved. The last section,

“markets,” contextualizes people’s experiences at the intersection of the increasing importance of the private sector in global health, greater divides in wealth, and states’ commitments to provide health for their citizens. The authors consider the effects of new modes of organizing access to health [e.g., public-private partnerships (Ecks and Harper)] and approaches predicated on a right to treatment (chapters by Clara Han and by Biehl and Petryna). Although these have been championed as better for the disenfranchised, they have instead resulted in complicated and less than ideal forms of care.

The authors’ empirical accounts of the complexities of the global health landscape expose a litany of assumptions that drive global health and demonstrate why we must be suspicious of these. One such assumption is that many of the interests that underwrite contemporary global health activity (such as increasing private competition, creating a “science of global health,” and demonstrating programmatic achievements) are proxies for global health success. The authors argue that these competing interests not only subvert the moral necessity of putting people first, they frequently hinder the goal of improving people’s health. Another assumption is that medical technologies themselves save lives. These technologies must be put to work in a complex global environment marked not only by ethnic, linguistic, cultural, and political heterogeneity but also by increasing inequity and inequality. *When People Come First* shows that the issue of how people get access to the treatments they need—be those pain killers, tuberculosis drugs, or cutting-edge DNA therapies—is very much alive.

#### References

1. N. Ravishankar, *Lancet* **373**, 2113 (2009).
2. J. Cohen, *Science* **311**, 162 (2006).
3. M. E. Porter, *N. Engl. J. Med.* **361**, 109 (2009).
4. J. Y. Kim, P. Farmer, M. E. Porter, *Lancet* **382**, 1060 (2013).

10.1126/science.1243510

### When People Come First Critical Studies in Global Health

João Biehl and  
Adriana Petryna, Eds.  
Princeton University  
Press, Princeton, NJ, 2013.  
454 pp. \$75, £52. ISBN  
9780691157382. Paper, \$29.95,  
£19.95. ISBN 9780691157399.