WHO HAS THE SPEED GENE, AND WHO DOESN'T?

HOW MUCH OF PERFORMANCE IS GENETIC?

HOW DID EARLY HUMANS BECOME ATHLETES?

AND CAN THE PERFECT ATHLETE BE GENETICALLY ENGINEERED?

SPORTS GENES

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ILLUSTRATIONS BY BRYAN CHRISTIE
Remember the guy or girl in high school who just had it? He was all-everything at quarterback and high jump; she led the pitching rotation and was also the starting point guard. Naturals. Or were they?

Did Ken Griffey Jr. inherit good genes, or did he become a superstar because he grew up in a clubhouse? Or both? For the price of a family outing at the ballpark, some companies will tell you if you have a certain sports gene. I have the sprinter gene, for instance, and you probably have it too.

Already, though, scientific research gives us a fuller picture of how we evolved into athletes, and it suggests that some things appear to be largely genetic (such as East African dominance of distance running) might not be, and that other things that seem entirely voluntary (such as an athlete’s will to train) might in fact have an important genetic component.

Scientific studies that associate particular genes with athleticism are published literally every month. These genes exist in differing versions, in all of us, from All-Pro to average Joe. As the study of performance genes accelerates, more assumptions about sports and genetics will no doubt prove false, and new answers will reshape our view of why and how some people become NFL running backs or Olympic swimmers, while others struggle to pass phy ed.

There is, for instance, the rubbery neck ligament that acts like a shock absorber for the head during running; the glint of sweat glands to help keep the body cool while running; the lack of body fur for the same reason; shoulders that move, unlike in apes, independently from the neck so that the arm can swing while the head remains still; long legs and narrow waists; larger surface area in hip, knee, and ankle joints, again for improved shock absorption; short toes, which are better for pushing off than for grasping tree branches; a chiseled foot, which acts as a spring, and big butt muscles to keep us upright. “Have you ever looked at an ape? They have no buns,” Bramble says. “We think running is one of the most transforming events in human history.”

No longer content merely to scavenge, our ancestors, despite having no greater weapons than sticks and stones, became deadly hunters. They overwhelmed their peripherally challenged quarry with a methodical chase that lasted until the...
beasts, unable to parasthesia literally while liv- ing, simply gave up from heat exhaustion.

Descants say we are because we think, but consider that we thought only after we ran. Even our large brain developed because we ran, growing only once our endurance enabled us to gorge on animal fat and protein enough to spare only sweating, largely hairless bipedal mammals—because we ran. As Lieberman puts it: “Endurance running is hardwired into our anatomy and physiology.”

For decades running was considered an unimportant part of human evolution because we humans are such pathetically winged at sprinting. In his world-record 200-meter dash, Usain Bolt averaged a little more than 3.2 mph for nearly 20 seconds. That would make him an absolute failure as a sprinter because an antelope can double that clip for minutes at a time. But with the help of our upright stance (which exposes less of our bodies to the sun) and our prodigious sweating, we can outrun just about any other animal on the planet if the race extends over hours in searing midday heat.

Sound familiar? Consider that hu-

ma n evolution has been the story of man nearly ever since the Neander-
ver-Horse-Race in Pescara, Italy. And that in one of two...
**The Uncommon Thread**

**In a Few Cases a Single Genetic Mutation has been Directly Linked to Heightened Athleticism**

Stephen M. Roth, an associate professor of kinesiology at Maryland, is among a group of scientists who track all the published work on potential performance genes and compiled a list known as the Human Gene Map for Performance. Roth notes that even seemingly straightforward athletic traits, such as strength or size, are highly complex, influenced as they are by multiple genetic and environmental factors. “With any single gene that seems to have a positive effect on performance, the effect is very subtle,” Roth says. There are, however, a few documented cases in which a single, rare genetic mutation has hugely increased an individual’s athleticism.

- Cerebromegalacia is a non-essential cross-country skier who was thin and average, but who, in his late teens, began to show signs of accelerated growth and a dramatic increase in strength.
- A young man with a rare form of endocrine disorder was found to have a mutation in a gene that affects the production of growth hormone.
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The mutation in each of these cases is rare, and it is not clear how widespread these effects might be. However, the cases do suggest that genetic factors can play a significant role in athletic performance. Further research is needed to understand the full extent of these effects and how they might be exploited in the development of new athletic training programs.
Part of the concern over the insurance issue was allayed last year when the federal Genetic Information Nondiscrimination Act took effect, barring employers and insurance companies from discriminating on the basis of genetic information. And research at Boston University has found that people who volunteer for an ApoE screen do not feel undue dread if they find out they have the deleterious variant. In fact, they usually embrace lifestyle habits, such as exercise, that doctors tell them might decrease their Alzheimer’s risk. “This is a very controversial area,” says Robert C. Green, a BU neurologist who helped conduct the work.

“The world of genetics for decades has suggested that there’s no reason to give people genetic-risk information unless there’s something proven you can do about it.”

For athletes facing head trauma, perhaps there are some relatively painless actions they could take. “If this gene is how you’re describing it,” says Duguay, whose test showed that he has one ApoE4 copy, “and I knew I had it when I was playing, I would’ve seriously considered wearing a helmet.”

Glen Johnson, a 41-year-old boxer with a 50-13-2 record, including wins over Roy Jones Jr. and Antonio Tarver, says he was considering retiring after his November loss to Chad Dawson. The older Johnson gets, the more he wants every bit of information that can tell him about what his life might be like after he quits fighting. Johnson—who understands that it isn’t a particular gene variant, but rather getting hit in the head, that is the key factor in brain injury—has already put his relatives and friends on alert to tell him if they notice any differences in his speech or memory. “I’d have to get a better understanding of [ApoE4], and

THE SINS OF THE FATHER...

YOU had the talent to be a sports star, but you lacked the discipline. You couldn’t bring yourself to go to bed early or pick up a barbell instead of an Oreo. On the bright side, your kid gets to start from square one: He or she inherits your talent but not the repercussions of your lackluster approach.

Or maybe not. Research in the growing science of epigenetics—which, among other things, looks at how genes can be switched on and off—suggests that your actions could have genetics-related consequences for your child. Better think twice before pounding down that next cookie.

At the turn of the 19th century, French naturalist Jean-Baptiste Lamarck proposed a theory of evolution that said animals quickly adapt to their environment and pass the adaptations along to their progeny, for example, giraffes acquired long necks because their ancestors stretched to reach leaves high on trees. But Lamarckian evolution was eclipsed in 1859 when Charles Darwin published his theory of evolution by natural selection, which says traits are acquired over millions of years as random genetic changes that happen to be beneficial are passed on. Today, however, the idea at the heart of Lamarck’s theory—that our choices can affect our children’s genes—is making a comeback.

Your behavior will not change the sequence of your child’s DNA, but it might change the action of certain molecules—called epigenetic marks—that attach to DNA and signal genes to turn on and off. Consider a study, published in 2008, of adults in the Netherlands whose mothers had suffered through a German-imposed food embargo in the winter of 1944–45. Six decades later, those whose mothers were in the early stages of pregnancy during the Dutch Hunger Winter had fewer “turn off” signaling methyl molecules attached to their IGF2 genes than did their siblings. The IGF2 gene is a key component in growth and development, and the study may help explain why children of mothers who experience famine during pregnancy have a higher risk of developing obesity, schizophrenia and diabetes later in life.

Studies now under way should further illuminate the epigenetic link to sports prowess. In one study published last year, rats that exercised regularly and rats that didn’t were twice thrown into beakers of water. On the first go-round, all the rats struggled vigorously to swim and attempt to escape the beaker. A day later, when they were again put into the water, the regularly exercised rats displayed better stress-coping mechanisms: Instead of clawing at a glass wall they could not climb, these rats, having learned from the previous day’s experience, conserved energy by floating. When their brains were examined afterward, researchers found that the exercised rats had altered epigenetic marks that in turn affected gene expression in a part of the brain that helps form memories. “Exercise has a great impact on the brain, not just the muscles,” says Johannes Reul, one of the researchers and a neuroscientist at the University of Bristol, England.

Perhaps one day a similar epigenetic explanation of memory will help us understand why great cornerbacks don’t get beat the same way twice.

—D.E.
I’d take a lot of other things and tests into consideration when I think about fighting again,” Johnson says, “but I’d never hide from extra information.”

**FINDING THE PERFECT ATHLETE**

If Pitsiladis is to pinpoint the athletically perfect genetic specimen, he or she must first exist. Just how many of these folks might have stepped right off Mount Olympus is a question that kept Alun Williams awake two years ago. Williams, a geneticist at Manchester Metropolitan University in England, and a colleague pored over the scientific literature and chose the 23 genetic variants that have been most strongly associated with talent in endurance sports. The scientists gathered information about the variants’ prevalence—some are found in more than 80% of people and others in fewer than 5%—and made statistical projections of how many “perfect” endurance athletes (people with two “correct” variants of each of the 23 genes) stride the earth.

Williams figured that perfection would be rare. After all, a Lance Armstrong comes around only once in a lifetime. But Williams was shocked when he ran the algorithm on his computer and saw that the odds of any person having all the right gene variants for endurance were less than one in a quadrillion. That’s a one followed by 15 zeroes. Think of it this way: If you pony up for 20 tickets each week, you’d have a better chance of winning the Mega Millions twice in a row than of hitting this genetic jackpot.

The bottom line is that even Lance isn’t a perfect specimen. Based on only the 23 chosen genes, there’s almost certainly no genetically perfect athlete alive. In fact, given that a meager 6.8 billion people live on our planet, chances are that nobody has the ideal endurance profile for more than 16 of the 23 genes. An individual is also unlikely to have only a few of them. Essentially everybody falls in the muddled middle, differing by only a handful of genes. It’s as if we’ve all played genetic roulette over and over, moving our chips around, winning sometimes and losing sometimes and gravitating toward mediocrity. “We’re all relatively similar because we’re relying on chance,” Williams says.

But if anyone is the beneficiary of a long genetic winning streak, it should be a world-record holder, shouldn’t it? Pitsiladis selected 24 gene variants most often associated with sprinting or endurance prowess and looked for them in the genomes of four men who have held the world record in the 100-meter dash and five who have held the world record in the marathon. What he saw was that based on those genes, the world-beaters are not genetic outliers at all. Pitsiladis analyzed the DNA of some of his graduate students for comparison and found that “a student of mine has a better rating for sprinting than the likes of an Asafa Powell or Usain Bolt.” (Pitsiladis is legally prohibited from identifying specific athletes with their genetic material, so he used Powell and Bolt as rhetorical examples.)

That rather startling result leaves two broad possibilities: First, there is a tremendous amount of work left to be done to find all the remaining genes that contribute to athletic success; second, something other than genetics is at work. Both may well be true, but only time and more research will rule on the former, while Pitsiladis has compiled considerable data on the latter.

Some of the most intriguing work comes from his study of the demographics of elite East African distance runners. When Pitsiladis analyzed Kenyan runners, he found that three quarters of all elite international competitors were from a single tribe, the Kalenjin, who make up a mere 10% of Kenya’s population. At first blush that would seem to indicate a genetic advantage in the Kalenjin, but Pitsiladis also found that they were likely to be living and training at altitude in the Rift Valley. When Pitsiladis compared 400 elite Kenyan athletes with a group of randomly selected Kenyans, he found that as children, the athletes were more likely to have lived at least several miles from school, and much more likely to have had to run there and back. Eighty-one percent of the elite Kenyan runners he studied had to rely on their feet to get to and from school, compared with only 22% of the control group. One 10-year-old boy whom Pitsiladis tested last year was already such an experienced runner that he clipped off six-minute miles when Pitsiladis tested him on a dirt track.

Haile Gebrselassie, the world-record holder in the marathon and perhaps the greatest distance runner ever, began running to school when he was five, covering more than six miles each way. For Ethiopians like him, Gebrselassie says, “every day is running. Every job is running: working in the fields or just getting somewhere. Life is running.” (The statistics bring to mind a mock charity drive announced in the late 1990s on a now defunct online track and field message board: Help Americans compete in distance running by donating school buses to Kenyan children.)

Pitsiladis and his colleagues found a similar pattern in Ethiopia. What was not shared between the Ethiopians and the Kenyans, however, was a particularly large proportion of their genes.

The maternal line of DNA has been found to be more influential in endurance than the paternal, so Pitsiladis’s group analyzed that genetic material and found that Ethiopian and Kenyan athletes “could not be more different genetically,” Pitsiladis says. The Ethiopian athletes, for example, were much more likely to have blocks of gene variants common in Europe and Asia than were the Kenyan athletes. Pitsiladis’s conclusion is that whatever specific genes are necessary for endurance, they aren’t exclusive to either Ethiopians or Kenyans.

His work suggests that some sports phenomena that seem on the surface to be entirely based in genetics might not be. (Or at least not in the way we’re used to thinking about genetics. A newer science called epigenetics is unraveling how environment and behavior, such as exercise, can actually turn particular genes on and off in patterns that might be passed on through generations.)

Similarly counterintuitive conclusions about the interplay between nature and nurture have come from outside sports: African-Americans are more prone than white Americans to hypertension, but the trend is not found among black people in some of the countries from which black Americans came, such as Jamaica and Nigeria. That points to the U.S., not to genes associated with blackness, as a culprit.

This is not to say that all ethnicities are the same. Nigerians are known for sprint-
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ING, and all Nigerians have at least one copy of the sprin gene variant. But so do nearly all Kenyans, as well as 80% of Europeans, two groups not renowned for sprinting. By the 1980s, when the first studies emerged, the pattern was clear: an athlete’s medicine, his medical students and for athletes, where is there evidence that the genetic variant is in their genes. But after 10 years of work, "I think we’ve never seen anything like this in a 2012, the LOZAZA study of about 1,000 people with spine injury. The study showed that 46% of the subjects had mutations in the APOL1 gene, which has been linked to a higher risk of kidney disease and HIV. The study also suggested that the mutations may play a role in the pathogenesis of chronic back pain. However, the study was limited in size and further research is needed to confirm its findings.

TREATMENT WITH LOZAZA may not have been shown to prevent heart attacks or strokes. So, what should you do if you have a sprin gene variant? The answer depends on your personal risk factors and the specific mutations you carry. In general, people with the sprin gene variant should be aware of their risk and take steps to manage it. This may include regular exercise, a healthy diet, and taking medications as prescribed.

In the face of evidence that the sprin gene variant is associated with a higher risk of heart attacks and strokes, it is important to stay informed and make lifestyle changes to reduce your risk. By taking steps to manage your health, you can help reduce your risk of heart disease and other health problems.