

Preface

We all know someone whose life is profoundly affected by the genes they inherited—perhaps an aunt with early-onset Alzheimer’s disease, or a cousin with cystic fibrosis, or a neighbor’s child with Down syndrome, or diabetes, or muscular dystrophy. Might Grandma’s failing eyesight have a genetic basis? Dad’s heart disease certainly seems to be genetic: Grandpa died of a heart attack in his late fifties. The consequences of altered genes can be seen all around us.

Progress in unraveling the genetic basis of disease and behavior is also evident all around us. Hardly a day goes by without a story in a newspaper or on a website proclaiming that some gene has been found to contribute to the risk for a serious disease such as diabetes, cancer, or colitis, or a condition such as depression, alcoholism or autism, or a trait such as fearlessness, aggressiveness, or anxiety.

How, we wondered, does anyone make sense of all of this? We quickly realized that almost no one does make sense of it. It’s not that the research is so complex or the public too dense to understand it. No, it’s simply that most people probably haven’t thought much about the workings of genetics since their high school biology course, and long ago forgot the key principles. As a consequence, the revelation that a newly identified gene is linked to an increased risk for cancer has about as much context for us as when we learn that a new political party has formed in Uzbekistan. As genetics researchers, we set out to make human genetics fathomable to all those whose tax dollars generously support this research—including, for the past two decades, the research we do in our own labs.

This book seeks to answer clearly and simply the key questions nearly everyone has about genes and genetics. Why do we resemble our parents more than any other set of parents? Does cancer run in families? Why do

some genetic diseases haunt families only when both parents carry the defective gene, whereas other genetic diseases are passed on to children from only one parent? What is our personal DNA code? How much of our behavior and our risk for diseases are influenced by this DNA code? Do the genes we get from Mom and Dad influence our moods? How does the single cell that is the fertilized egg become a baby made up of trillions of cells? What is evolution by natural selection (and was Darwin really the guy who came up with the idea)? This book is not a genetics text; these chapters and the stories they tell are meant to be quickly assimilated and easily digested.

To nonscientists, science can seem an endless stream of dry technical details. Yet all of us are tantalized by stories about real people—about the trials of the rich and the famous, as well as about the tribulations of the average Joe and Jane. Many of the most bizarre aspects of the tales we tell you here hinge on the inheritance of one tiny piece of DNA.

We'll consider the fates of some celebrated people: literary luminaries, including a Nobel Prize-winning novelist driven by a heavy genetic burden, a prolific writer of science fiction whose overeating led to more than obesity, and an immigrant magazine editor who confronted a U.S. president as well as a relentless disease; media and movie celebrities, like the seductive actress who slowly lost her mind, and the widow who rose to the highest ranks of network news and used her fame to promote cancer tests; and a star athlete who faced the dual challenges of a terminal illness as well as the world's prejudice.

We'll also recount a few of the most notable cases in the annals of medicine: the story of the so-called "Bubble Boy," who waited in vain for a cure that came too late; a confused and forgetful woman whose doctor gave his name to a disease that afflicts millions of us; and a young diabetic who nearly died waiting for the first batches of insulin to become available.

And we'll describe ordinary citizens facing extraordinary challenges: a mother wrongly accused of poisoning her infant son; mountain-climbing brothers at risk for a neurological disease; a man who made sure that all his relatives gave blood in a quest to uncover the cause of their mysterious affliction.

What all these tales have in common is a twist of genetic fate: the inheritance of one minuscule change rather than another in our vast per-

sonal DNA code, a change that made the difference between health and disease, between happiness and heartbreak, between life and death.

In the course of telling these people's tales we can't help but introduce a few of our fellow biologists who have contributed to the remarkable advances in genetics. Long before "intelligent design" was in the news, two men raced each other to explain evolution. A scientist who thought outside the box tackled a disease of cattle and ended up discovering a potent rat poison that became the most common treatment for heart attacks. A physicist split the gene, and a German woman confronted scientific orthodoxy and laid bare how a baby can develop from a single cell.

We are confident that after reading these accounts, you will understand what genes are and how scientists track them down. You will be able to appreciate why particular versions of genes in a personal DNA code put someone at risk for diabetes, or dementia, or depression. And we hope that the principles of genetics presented in this book help you understand the results of a genetic test that you or a loved one may someday undergo.

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