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# Genetic Twists of Fate

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## Notes and Further Reading

### Chapter 1

The history of Google is told by David Vise and Mark Malseed in *The Google Story* (New York: Delacorte Press, 2005). Jim Watson has written several memoirs of his life in science, including *The Double Helix: A Personal Account of the Discovery of the Structure of DNA* (New York: Atheneum, 1968).

### Chapter 2

Isaac Asimov told his story in *It's Been a Good Life* (edited by J. J. Asimov); Amherst, NY: Prometheus Books, 2002; we also used details from *Asimov: The Unauthorised Life* by Michael White (London: Millenium, an imprint of Orion Books, 1994). The structure of DNA and how it codes information, as well as the principles of basic genetic inheritance described in other chapters, is available in books online at the PubMed Web site <<http://www.ncbi.nlm.nih.gov/sites/entrez>>, especially *Molecular Biology of the Cell* by B. Alberts, A. Johnson, J. Lewis, M. Raff, K. Roberts, and P. Walter (New York: Garland Science, 2002); *Genomes* by T.A. Brown (New York: Garland Science, 2002); and *Molecular Cell Biology* by H. Lodish, A. Berk, S. L. Zipursky, P. Matsudaira, D. Baltimore, and J. E. Darnell, (New York: W. H. Freeman & Co., 1999).

### Chapter 3

The story of how Patricia Stallings was falsely convicted of murdering her infant son has been told in several newspaper and magazine articles. We drew on articles by St. Louis Post-Dispatch reporters Bill McClellan ("Refusal To Accept Odd Coincidence Saved Stallings," St. Louis Post-Dispatch

September 25, 1991), Bill Smith ("Not Guilty! How the system failed Patricia Stallings," *St. Louis Post-Dispatch*, October 20, 1991), and Tom Uhlenbrock (*St. Louis Post-Dispatch* "Painfully true," March 18, 1993), by Rhonda Riglesberger ("How the Legal and Medical Systems failed Patricia and Ryan Stallings" in *Justice:Denied*, the magazine for the wrongly convicted, Volume 1, issue 8), and by Tim Graham ("When Good Science Goes Bad" in the October 2004 issue of *ChemMatters*). The case was described in the medical literature by James D. Shoemaker, Robert E. Lynch, Joseph W. Hoffman, and William S. Sly ("Misidentification of propionic acid as ethylene glycol in a patient with methylmalonic academia." *Journal of Pediatrics* 120:417–21). We were also informed by a report about the case that aired on December 9, 1998 on TLC Discovery Channel's Innocence Files show "Deadly Formula," and by conversations with Dr. William Sly of St. Louis University. Reliable information on genetic diseases can be found at many sites on the Internet, including the Mayo Clinic <<http://www.mayoclinic.com>>, the National Institutes of Health <<http://health.nih.gov/category/GeneticsBirthDefects>>, <<http://www.genome.gov/27527652>>, <<http://www.genome.gov/11008303>>, and their Genetics Home Reference Web site <<http://ghr.nlm.nih.gov/>>, and the National Organization for Rare Disorders <<http://www.rarediseases.org/>>.

## Chapter 4

The history of the use of *Drosophila* in the study of developmental biology is told in *Fly: The Unsung Hero of 20th Century Science* by Brookes, M., New York: HarperCollins Publishers Inc., 2001; *Lords of the Fly* by Kohler, R.E., Chicago: The University of Chicago Press, 1994; and *Coming to Life: How Genes Drive Development* by Nüsslein-Volhard, C., Carlsbad, CA: Kales Press, Inc., 2006. E.F. Keller discussed the career of Christiane Nüsslein-Volhard in "Drosophila embryos as transitional objects: The work of Donald Poulson and Christiane Nüsslein-Volhard," *Historical Studies in the Physical and Biological Sciences*. Volume 26, Part 2, pp. 313–346, 1996.

## Chapter 5

The story of the "Bubble Boy" David Phillip Vetter has been told many times; we relied mostly on an excellent article by Steve McVicker published in the

April 10, 1997 issue of the *Houston Press News* and *The Boy in the Bubble*, a documentary film produced and directed by Barak Goodman and John Maggio that aired on Public Television April 10, 2006. The use of gene therapy to correct SCID is described by Andrea Kon in the January 27, 2007, issue of *The Daily Telegraph's Telegraph magazine* ("A chance for life"), by Gaspar et al. in *The Lancet* 364:2181–2187 ("Gene therapy of X-linked severe combined immunodeficiency by use of a pseudotyped gammaretroviral vector"), and by Hacein-Bey-Abina et al. in the *New England Journal of Medicine* 346:1185–1193 ("Sustained correction of x-linked Severe Combined Immunodeficiency by *ex vivo* gene therapy"). How gene therapy caused leukemia in some patients is explained by Hacein-Bey-Abina et al. in the *Journal of Clinical Investigation* 118:3132–3142 ("Insertional oncogenesis in 4 patients after retrovirus-mediated gene therapy of SCID-X1"). The story of Jesse Gelsinger's untimely death was told by Sheryl Gay Stolberg in the November 29, 1999, issue of the *New York Times* ("The Biotech Death of Jesse Gelsinger"). More information on gene therapy can be obtained from the American Society of Gene & Cell Therapy <<http://www.asgt.org/>>.

## Chapter 6

The history of Elizabeth Hughes and the early days of insulin research is recounted in *The Discovery of Insulin* by M. Bliss (Chicago: University of Chicago Press, 1984). Information on the genetics of diabetes is available from the American Diabetes Association <<http://www.diabetes.org/>>. Information about stem cells and links to related resources can be found at the National Institutes of Health Stem Cell Information Web site (<http://stemcells.nih.gov/>).

## Chapter 7

Pearl S. Buck provides an account of her daughter's life in *The Child Who Never Grew* (New York: The John Day Company, 1950). The medical history of phenylketonuria is told by S.E. Christ, ("Asbjørn Følling and the discovery of phenylketonuria," *Journal of the History of the Neurosciences* 12 [2003]: 44–54), and by S.A. Centerwall and W.E. Centerwall ("The discovery of phenylketonuria: the story of a young couple, two retarded children, and a scientist," *Pediatrics* 105 [2000]: 89–103).

## Chapter 8

The story of the O'Brien's ill-fated Everest expedition is available at <http://www.everestnews.com/> and news articles in the *Chicago Sun-Times*, May 4, 2005, by Lori Rackl; the *Oswego Post Standard*, May 2, 2005, by Mike McAndrew; and the *Seattle Times*, May 3, 2005, by Sara Jean Green. Alice Wexler describes her family's history with Huntington's disease in *Mapping Fate: A Memoir of Family, Risk, and Genetic Research* (Berkeley: University of California Press, 1995). The Hereditary Disease Foundation <<http://www.hdfoundation.org/home.php>> has extensive information about the current state of Huntington's disease research.

## Chapter 9

Rita Hayworth's life is described in *If This Was Happiness: A Biography of Rita Hayworth*, by Barbara Leaming (New York: Viking, 1989) and *Rita Hayworth: The Time, the Place, the Woman* by John Kobal (New York: W.W. Norton and Co., 1977). Current research on Alzheimer's disease is available from the Alzheimer's Association <<http://www.alz.org/index.asp>>.

## Chapter 10

The three examples of behavioral traits we used to illustrate the concept of heritability were drawn from the medical and scientific literature: K.L. Toh et al., "An hPer2 phosphorylation site mutation in familial advanced sleep phase syndrome," *Science*, volume 291, pp 1040–3; C.R. Jones et al., "Familial advanced sleep-phase syndrome: A short-period circadian rhythm variant in humans," *Nature Medicine* 5:1062–1065; Y. Xu et al., "Functional consequences of a CKIdelta mutation causing familial advanced sleep phase syndrome," *Nature* 434:640–644; P. McGuffin and D. Mawson, "Obsessive-compulsive neurosis: two identical twin pairs," *British Journal of Psychiatry* 137:285–287; R. Muhle, S.V. Trentacoste, and I. Rapin, "The genetics of autism," *Pediatrics* 113: 472–486.

## Chapter 11

We learned about Aldred Scott Warthin and Pauline Gross from articles by Henry T. Lynch ("Aldred Scott Warthin, M.D., Ph.D. (1866–1931)" in *CA*:

*A Cancer Journal for Clinicians* 35:345–347), by Claudia Kalb (“Peering into the Future: Genetic Testing” in *Newsweek*, December 11, 2006), an unattributed article in the August 10, 1936 issue of *Time* (“G’s Family”), and a radio show, “Daughter of Family G,” written and produced by Ami McKay for the Canadian Broadcasting Corporation (available at [www.soundprint.org](http://www.soundprint.org)). Much has been written about Katie Couric and Jay Monahan; two articles that we read are by Jacquelyn Mitchard (August 13, 2006, issue of *Parade* magazine), and Joanna Powell’s interview of Katie Couric published in the October 1998 issue of *Good Housekeeping*. More information on colon cancer can be obtained from the Jay Monahan Center for Gastrointestinal Health <<http://monahancenter.org/>>. Information on all kinds of cancers can be obtained from the American Cancer Society <<http://www.cancer.org/docroot/home/index.aspx>> and from the National Cancer Institute <<http://www.cancer.gov/>>.

## Chapter 12

Seymour Benzer reviews his life in interviews by Heidi Aspaturian, September 11, 1990–February 1991 as part of the Oral History Project, California Institute of Technology Archives. Pasadena, CA, <[http://resolver.caltech.edu/CaltechOH\\_Benzer\\_S](http://resolver.caltech.edu/CaltechOH_Benzer_S)>. He is also the subject of a biography, *Time, Love, Memory* by J. Weiner (New York: Alfred A. Knopf, 1999). Benzer describes his early phage work in “The fine structure of the gene,” *Scientific American*, January 1962.

## Chapter 13

We learned much about all forms of Amyotrophic Lateral Sclerosis from the ALS Division of the Muscular Dystrophy Association <<http://www.als-mda.org>>. Especially helpful for our story were articles in their newsletter (“Mattingly Family Featured in Life Magazine,” *ALS Newsletter* 3, no. 4 (October 1998); “Gene Mapped for Early-Onset, Slowly Progressive Form of ALS,” *ALS Newsletter* 3, no. 2 (April 1998); “Gene Found for Early-Onset ALS” by Margaret Wahl, *MDA/ALS Newsmagazine*, 9, no. 5 (May 2004). The mapping and identification of the gene responsible for the Mattingly clan’s ALS was described in articles by Phillip Chance et al. published in the *American Journal of Human Genetics* 62:633–640, “Linkage of the Gene for an Autosomal Dominant Form of Juvenile Amyotrophic Lateral Sclerosis to Chromo-

some 9q34"; "DNA/RNA Helicase Gene Mutations in a Form of Juvenile Amyotrophic Lateral Sclerosis (ALS4)" volume 74, pp 1128–1135).

## Chapter 14

Henry Grunwald recounted his life in *One Man's America: A Journalist's Search for the Heart of His Country* (New York: Doubleday, 1997), and his battle with macular degeneration in *Twilight: Losing Sight, Gaining Insight*; New York: Alfred A. Knopf, 1999. The NIH <[http://www.nei.nih.gov/health/maculardegen/armd\\_facts.asp](http://www.nei.nih.gov/health/maculardegen/armd_facts.asp)> and the American Macular Degeneration Foundation <<http://www.macular.org/>> provide information about macular degeneration.

## Chapter 15

The story of warfarin's development is told by K.P. Link ("The Discovery of Dicumarol and Its Sequels," *Circulation* 19 [1959]:97–107), and by R.L. Mueller and S. Scheidt ("History of drugs for thrombotic disease. Discovery, development, and directions for the future," *Circulation* 89 [1994]:432–449).

## Chapter 16

The story of Alfred Russel Wallace and his contributions to evolutionary theory is well-told by Arnold C. Brackman in *A delicate arrangement: The strange case of Charles Darwin and Alfred Russel Wallace* (New York: Times Books, 1980), and by Wallace himself in *My Life*, volumes 1 and 2 (Chapman & Hall, LD, London, available at Google books). The full text of Darwin's and Wallace's first presentation of the theory of evolution by natural selection can be found at <<http://linnean.org/index.php?id=380>>. Much useful information about evolution can be found at The National Center for Science Education <<http://ncse.com/>>.

## Chapter 17

Arthur Ashe's life is told in *Charging the Net: A History of Blacks in Tennis from Althea Gibson and Arthur Ashe to the Williams Sisters*, by C. Harris and

L. Kyle-DeBose (Chicago: Ivan R. Dee, 2007). A discussion of race and genetics is provided by R.A. Kittles and K.M. Weiss in "Race, ancestry, and genes: implications for defining disease risk," *Annual Review of Genomics and Human Genetics* volume 4, pp 33–67, 2003. The BiDiL story is recounted in "The use of race and ethnicity in medicine: lessons from the African-American Heart Failure Trial," by J. N. Cohn, *Journal of Law, Medicine and Ethics* 34 (2006): 552–554.





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