

LITERATÚRA A POZNÁMKY

Literatúra dotýkajúca sa genetiky a molekulárnej biológie je nesmierne rozsiahla a rýchlo zastaráva. Každá kniha, článok, či vedecký príspevok vyžaduje pri publikovaní zaradenie najnovších informácií alebo revíziu. Nové poznatky pribúdajú závratnou rýchlosťou (platí to aj pre moju knihu). Mnohí vedci sa zaoberajú takou problematikou, že je pre nich obťažné držať krok s ostatnými prácami. Pri písaní tejto knihy som zistil, že časté návštevy knižnice a konverzácia s vedcami sú nedostatočné. Nový spôsob ako držať krok s dobou znamená surfovať na internete.

Najbohatším prameňom genetických poznatkov je jedinečná webová stránka Victora McKusicka známa ako OMIM (Online Mendelian Inheritance in Man). Jej adresa je <http://www.ncbi.nlm.nih.gov/omim/>. Obsahuje zvláštnu stať s prameňmi o každom ľudskom géne, ktorý bol zmapovaný alebo sekvencovaný a je pravidelne aktualizovaná – čo je takmer nepredstaviteľný výkon. Weizmannov inštitút v Izraeli má tiež výbornú webovú stránku s „génovými kartami“, ktoré sumarizujú všetko známe o každom géne a spojenia k iným relevantným webovým stránkam: bioinformatics.weizmann.ac.il/cards.

Tieto webové stránky však poskytujú iba súhrny poznatkov a nehodia sa pre menej odvážnych, pretože je v nich veľa

odbornej terminológii a predpokladaných poznatkov, čo mnohých nadšencov odrádza. Koncentrujú sa tiež na význam každého génu pre zdedené poruchy. Preto vlastne zdôrazňujú názor, s ktorým som sa v tejto knihe snažil bojovať: že hlavnou funkciou génov je spôsobovať choroby.

Pri dopĺňaní a vysvetľovaní posledných poznatkov som sa spoliehal predovšetkým na učebnice. K najlepším patria: *Human molecular genetics* (Bios Scientific Publishers, 1996) od Toma Strachana a Andrewa Reada, *Basic genetics* (William C. Brown, 1995) od Roberta Weavera a Philipa Hedricka, *DNA science* (Cold Spring Harbor Laboratory Press, 1990) od Davida Micklosa a Grega Freyera a *Genes VI* (Oxford University Press, 1997) od Benjamin Lewina.

K populárnejším knihám o genóme ako celku, ktoré môžem odporúčať, patria: Christopher Will: *Exons, introns and talking genes* (Oxford University Press, 1991), Walter Bodmer a Robin McKie: *The book of man* (Little, Brown, 1994) a Steve Jones: *The language of the genes* (Harper Collins, 1993), ako aj Tom Strachan: *The human genome* (Bios, 1992). Na všetkých sa však nevyhnutne prejavuje ich vek.

V každej kapitole tejto knihy mi bol oporou zväčša jeden alebo dva hlavné zdroje a mnoho jednotlivých vedeckých príspevkov. Účelom poznámok je usmerniť čitateľa, ktorý si praje sledovať opisované predmety až k týmto prameňom.

CHROMOZÓM 1

Myšlienka o tom, že gén, rovnako ako život, pozostávajú z digitálnej informácie, sa nachádza v knihe Richarda Dawkinsa *River out of Eden* (Weidenfeld and Nicolson, 1995) a v Jeremy Campbellom *Grammatical man* (Allen Lane, 1983). Vynikajúci príspevok k debatám o pôvode života, ktoré prebiehajú doposiaľ, sa nachádza v diele Paula Daviesa *The fifth miracle* (Penguin, 1998). Podrobnejšie informácie o svete RNA poskytujú Gesteland, R. F. a Atkins, J. F. (editori) *The RNA world* (Cold Spring Harbor Laboratory Press, New York, 1993)

- ¹ Darwin, E.: *Zoonomia: or the laws of organic life*, II. diel, s. 244, 3. vydanie, J. Johnson, London, 1801
- ² Campbell, J.: *Grammatical man: information, entropy, language and life*. Allen Lane, London, 1983
- ³ Schrödinger, E.: *What is life? Mind and matter*, Cambridge University Press, Cambridge, 1967
- ⁴ Citované v Judson, H. F.: *The eighth day of creation*, Jonathan Cape, London, 1979
- ⁵ Hodges, A.: *Turing*. Phoenix, London, 1997
- ⁶ Campbell, J.: *Grammatical man: information, entropy, language and life*, Allen Lane, London, 1983
- ⁷ Joyce, G. F.: RNA evolution and the origins of life, *Nature* 338, 1989, s. 217 – 24; Unrau, P. J. a Bartel, D. P.: RNA-catalysed nucleotide synthesis, *Nature* 395, 1998, s. 260 – 63
- ⁸ Gesteland, R. F. a Atkins, J. F. (editori): *The RNA world*, Cold Spring Harbor Laboratory Press, Cold Spring Harbor, New York, 1993
- ⁹ Gold, T. (1992). The deep, hot biosphere. *Proceedings of the National Academy of Sciences of the USA* 89, s. 6045 – 49; Gold, T.: An unexplored habitat for life in the universe? *American Scientist* 85, 1997, s. 408 – 11
- ¹⁰ Woese, C.: The universal ancestor, *Proceedings of the National Academy of Sciences of the USA* 95, 1998, s. 6854 – 9
- ¹¹ Poole, A. M., Jeffares, D. C. a Penny, D.: The path from the RNA world, *Journal of Molecular Evolution* 46, 1998, s. 1 – 17; Jeffares, D. C., Poole, A. M. a Penny, D.: Relics from the RNA world, *Journal of Molecular Evolution* 46, 1998, s. 18 – 36.

CHROMOZÓM 2

Príbeh o ľudskej evolúcii z opičieho predka bol prerozprávaný, či zopakovaný mnohokrát. Dobré príspevky na túto tému sú v knihách: N. T. Boaz *Eco homo* (Basic Books, 1997), Alan Walker and Pat Shipman *The wisdom of bones* (Phoenix, 1996), Richard Leakey a Roger Lewin *Origins*

reconsidered (Little, Brown, 1992) a Don Johansonovej a Blake Edgarovej veľkolepo ilustrovanej *From Lucy to language* (Weidenfeld and Nicolson, 1996).

- ¹ Kottler, M. J.: From 48 to 46: cytological technique, pre-conception, and the counting of human chromosomes, *Bulletin of the History of Medicine* 48, 1974, s. 465 – 502.
- ² Young, J. Z.: *The life of vertebrates*, Oxford University Press, Oxford, 1950
- ³ Arnason, U., Gullberg, A a Janke, A.: Molecular timing of primate divergences as estimated by two non-primate calibration points. *Journal of Molecular Evolution* 47, 1998, s. 718 – 27.
- ⁴ Huxley, T. H.: *Man's place in nature and other anthropological essays*, s. 153, Macmillan, London, 1863/1901
- ⁵ Rogers, A. a Jorde, R. B.: Genetic evidence and modern human origins. *Human Biology* 67, 1995 s. 1 – 36.
- ⁶ Boaz, N. T.: *Eco homo*. Basic Books, New York, 1997
- ⁷ Walker, A. and Shipman, P.: *The wisdom of bones*, Phoenix, London, 1996
- ⁸ Ridley, M.: *The origins of virtue*, Viking, London, 1996

CHROMOZÓM 3

Spomedzi mnohých príspevkov k histórii genetiky je najlepším kniha Horace Judsona *The eighth day of creation* (Jonathan Cape, London, 1979; reprint v Penguin, 1995).

Pozoruhodným príspevkom o Mendelovom živote je román Simona Mawera *Mendel's dwarf* (Doubleday, 1997).

- ¹ Bearn, A. G. a Miller, E. D.: Archibald Garrod and the development and the concept of inborn errors of metabolism. *Bulletin of the History of Medicine* 53, 1979 s. 315 – 28; Childs, B.: Sir Archibald Garrod's conception of chemical individuality: a modern appreciation, *New England Journal of Medicine* 282, 1970, s. 71 – 7; Garrod, A.: *Inborn errors of metabolism*. Oxford University Press, Oxford, 1909

- ² Mendel, G.: Versuche über Pflanzen-Hybriden. *Verhandlungen des naturforschenden Vereines in Brünn* 4, 1865, s. 3 – 47
Anglický preklad publikovaný v *Journal of the Royal Horticultural Society*, Vol. 26, 1901
- ³ Citované vo Fisher, R. A.: *The genetical theory of natural selection*, Oxford University Press, Oxford, 1930
- ⁴ Bateson, W.: *Mendel's principles of heredity*. Cambridge University Press, Cambridge, 1909
- ⁵ Miescher, citovaný v publikácii Bodmer, W. a McKie, R.: *The book of man*. Little, Brown, London, 1994
- ⁶ Dawkins, R.: *River out of Eden*. Weidenfeld and Nicolson, London, 1995
- ⁷ Hayes, B.: The invention of the genetic code. *American Scientist* 86, 1998, s. 8 – 14.
- ⁸ Scazzocchio, C.: Alkaptonuria: from humans to moulds and back. *Trends in Genetics* 13, 1997, s. 125 – 7; Fernandez-Cannon, J. M. a Penalva, M. A.: Homogentisate dioxygenase gene cloned in *Aspergillus*. *Proceedings of the National Academy of Sciences of the USA* 92, 1995, s. 9132 – 6.

CHROMOZÓM 4

Pre záujemcov o dedičné poruchy, akou je Huntingtonova choroba, sú podstatnou informáciou publikácie Nancy a Alice Wexlerových, podrobnejšie uvedené v poznámkach. Veľmi prístupným sprievodcom je aj kniha Stephena Thomasa *Genetic risk* (Pelican, 1986).

- ¹ Thomas, S.: *Genetic risk*. Pelican, London, 1986
- ² Gusella, J. F., McNeil, S., Persichetti, F., Srinidhi, J., Novelletto, A., Bird, E., Faber, P., Vonsattel, J.-P., Myers, R. H. a MacDonald, M. E.: Huntington's disease, *Cold Spring Harbor Symposia on Quantitative Biology* 61, 1996, s. 615 – 26.
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- ⁴ Wexler, N.: Clairvoyance and caution: repercussions from the Human Genome Project, In: *The code of codes* (editori

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- ⁵ Huntington's Disease Collaborative Research Group: A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes, *Cell* 72, 1993, s. 971 – 83
 - ⁶ Goldberg, Y. P. *et al.*: Cleavage of huntingtin by apopain, a proapoptotic cysteine protease, is modulated by the polyglutamine tract. *Nature Genetics* 13, 1996 s. 442 – 9; DiFiglia, M., Sapp, E., Chase, K. O., Davies, S. W., Bates, G. P., Vonsattel, J. P. a Aronin, N.: Aggregation of huntingtin in neuronal intranuclear inclusions and dystrophic neurites in brain. *Science* 277, 1997, s. 1990 – 93
 - ⁷ Kakiuza, A.: Protein precipitation: a common etiology in neurodegenerative disorders? *Trends in genetics* 14, 1998, s. 398 – 402
 - ⁸ Bat, O., Kimmel, M. a Axelrod, D. E.: Computer simulation of expansions of DNA triplet repeats in the fragile-X syndrome and Huntington's disease, *Journal of Theoretical Biology* 188, 1997 s. 53 – 67
 - ⁹ Schweitzer, J. K. and Livingston, D. M.: Destabilisation of CAG trinucleotide repeat tracts by mismatch repair mutations in yeast, *Human Molecular Genetics* 6, 1997, s. 349 – 55
 - ¹⁰ Mangiarini, L.: Instability of highly expanded CAG repeats in mice transgenic for Huntington's disease mutation, *Nature Genetics* 15, 1997 s. 197 – 200; Bates, G. P., Mangiarini, L., Mahal, A. a Davies, S. W.: Transgenic models of Huntington's disease, *Human Molecular Genetics* 6, 1997, s. 1633 – 7
 - ¹¹ Chong, S. S. *et al.*: Contribution of DNA sequence and CAG size to mutation frequencies of intermediate alleles for Huntington's disease: evidence from single sperm analyses, *Human Molecular Genetics* 6, 1997, s. 301 – 10
 - ¹² Wexler, N. S.: The Tirasias complex: Huntington's disease as a paradigm of testing for late-onset disorders, *FASEB Journal* 6, 1992, s. 2820 – 25

- ¹³ Wexler, A.: *Mapping fate*. University of California Press, Los Angeles, 1995

CHROMOZÓM 5

Jednou z najlepších publikácií o love na gény je kniha Williama Cooksona *The gene hunters: adventures in the genome jungle* (Aurum Press, 1994). Cookson je jedným z mojich najdôležitejších zdrojov pri informovaní o génoch astmy.

- ¹ Hamilton, G.: Let them eat dirt, *New Scientist*, 18. júla 1998, s. 26 – 31; Rook, G. A. W. a Stanford, J. L.: Give us this day our daily germs, *Immunology Today* 19, 1998, s. 113 – 16
- ² Cookson, W.: *The gene hunters: adventures in the genome jungle*, Aurum Press, London, 1994
- ³ Marsh, D. G. *et al.*: Linkage analysis of IL₄ and other chromosome 5q31.1 markers and total serum immunoglobulin-E concentrations, *Science* 264, 1994, s. 1152 – 6
- ⁴ Martinez, E. D. *et al.*: Association between genetic polymorphism of the beta-2-adrenoceptor and response to albuterol in children with or without a history of wheezing, *Journal of Clinical Investigation* 100, 1997, s. 3184 – 8.

CHROMOZÓM 6

Príbeh o tom, ako Robert Plomin hľadal gény, ktoré ovplyvňujú inteligenciu sa objaví v knihe, ktorú chystá Rosalind Ardenová. Plominova učebnica *Behavioral genetics* je mimoriadne dobre zrozumiteľný úvod do tejto problematiky (tretie vydanie, W. H. Freeman, 1997). Kniha Stephena Jay Goulda *Mismeasure of man* (Norton, 1981) je vhodným príspevkom k ranej histórii eugeniky a IQ. Nádherne sa číta kniha Lawrence Wrighta *Twins: genes, environment and the mystery of identity* (Weidenfeld and Nicholson, 1997).

- ¹ Chorney, M. J., Chorney, K., Seese, N., Owen, M. J., Daniels, J., McGuffin, P., Thompson, L. A., Detterman, D. K., Benbow, C., Lubinski, D., Eley, T. a Plomin, R.: A quanti-

- tative trait locus associated with cognitive ability in children, *Psychological Science* 9, 1998, s. 1 – 8
- ² Galton, F.: *Inquiries into human faculty*, Macmillan, London, 1883
- ³ Goddard, H. H. (1920), citované v Gould, S. J.: *The mismeasure of man*, Norton, New York, 1981
- ⁴ Neisser, U. et al.: Intelligence: known and unknown, *American Psychologist* 51, 1996, s. 77 – 101
- ⁵ Philpott, M.: Genetic determinism, v Tam, H. (ed.): *Punishment, excuses and moral development*, Avebury, Aldershot, 1996
- ⁶ Wright, L.: *Twins: genes, environment and mystery of identity*, Weidenfeld and Nicolson, London, 1997
- ⁷ Scarr, S.: Developmental theories for the 1990s: development and individual differences, *Child Development* 63, 1992, s. 1 – 19
- ⁸ Daniels, M., Devlin, B. a Roeder, K.: Of genes and IQ. V: Devlin, B., Fienberg, S. E., Resnick, D. P. a Roeder, K. (eds), *Intelligence, genes and success*. Copernicus, New York, 1997
- ⁹ Herrnstein, R. J. a Murray, C.: *The bell curve*, The Free Press, New York, 1994
- ¹⁰ Haier, R. et al.: Intelligence and changes in regional cerebral glucose metabolic rate following learning, *Intelligence* 16, 1992, s. 415 – 26.
- ¹¹ Gould, S. J.: *The mismeasure of man*, Norton, New York, 1984
- ¹² Furlow, F. B., Armijo-Prewitt, T., Gangestad, S. W. a Thornhill, R.: Fluctuating asymmetry and psychometric intelligence, *Proceedings of the Royal Society of London, Series B* 264, 1997, s. 823 – 9
- ¹³ Neisser, U.: Rising scores on intelligence tests, *American Scientist* 85, 1997, s. 440 – 47

CHROMOZÓM 7

Evolučnej psychológii, ktorá je predmetom tejto kapitoly, sa venuje veľa kníh, vrátane diel Jeroma Barkowa, Ledy Cosmidesovej a Johna Toobyho *The adapted mind* (Oxford University Press, 1992), Roberta Wrighta *The moral animal*

(Pantheon, 1994), Stevena Pinkera *How the mind works* (Penguin, 1998) a mojej knihy *The red queen* (Viking, 1993). Pôvod ľudských rečí skúmajú Steven Pinker *The language instinct* (Penguin, 1994) a Terence Deacon *The symbolic species* (Penguin, 1997).

- ¹ O smrti freudianizmu: Wolf, T.: Sorry but your soul just died. *The Independent on Sunday*, 2. februára 1997. O smrti meadizmu: Freeman, D.: Margaret Mead and Samoa: the making and unmaking of an anthropological myth, Harvard University Press, Cambridge, MA, 1983; Freeman, D.: *Frans Boas and „The flower of heaven“*. Penguin, London, 1997. O smrti behaviorizmu: Harlow, H. F., Harlow, M. K. a Suomi, S. J.: From thought to therapy: lessons from a primate laboratory, *American Scientist* 59, 1971, s. 538 – 49
- ² Pinker, S.: *The language instinct: the new science of language and mind*. Penguin, London, 1994
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- ⁴ Carter, R.: *Mapping the mind*. Weidenfeld and Nicolson, London, 1998
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- ⁶ Fisher, S. E., Vargha-Khadem, F., Watkins, K. E., Monaco, A. P. a Pembrey, M. E.: Localisation of a gene implicated in a severe speech and language disorder, *Nature Genetics* 18, 1998, s. 168 – 70
- ⁷ Gopnik, M.: Feature-blind grammar and dysphasia, *Nature* 344, 1990, s. 715
- ⁸ Fletcher, P.: Speech and language deficits, *Nature* 346, 1990, s. 226; Vargha-Khadem, F. a Paasingham, R. E.: Speech and language deficits, *Nature* 346, 1990, s. 226

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- ¹⁰ Gopnik, M. a Goad, H.: What underlies inflectional error patterns in genetic dysphasia? *Journal of Neurolinguistics* 10, 1997, s. 109 – 38; Gopnik, M.: Familial language impairment: more English evidence, *Folia Phonetica et Logopaedia* 51, 1999 (Autor získal text ešte pred uverejnením e-mailovou korešpondenciou s autorkou v roku 1998)
- ¹¹ Associated Press, 8. mája 1997; Pinker, S.: *The language instinct: the new science of language and mind*, Penguin, London, 1994
- ¹² Mineka, S. a Cook, M.: Mechanisms involved in the observational conditioning of fear, *Journal of Experimental Psychology, General* 122, 1993, s. 23 – 38
- ¹³ Dawkins, R.: *The blind watchmaker*. Longman, Essex, 1986

CHROMOZÓMY X A Y

Najlepším miestom, kde sa dá viac dozvedieť o intragenomickom konflikte, je učebnica autorov Michaela Majerusa, Billa Amosa a Gregoryho Hursta *Evolution: the four billion year war* (Longman, 1996) a kniha W. D. Hamiltona *Narrow roads of gene land* (W. H. Freeman, 1995). K výskumom, ktoré viedli k záveru o čiastočnom genetickom pôvode homosexuality, patria práce Deana Hamera a Petera Copelanda *The science of desire* (Simon and Schuster, 1995) a Chandlera Burra *A separate creation: how biology makes us gay* (Bantam Press, 1996).

- ¹ Amos, W. a Harwood, J.: Factors affecting levels of genetic diversity in natural populations. *Philosophical Transactions of the Royal Society of London, Series B* 353, 1998, s. 177 – 86

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- ³ Majerus, M., Amos, W. a Hurst, G.: *Evolution: the four billion year war*, Longman, Essex, 1996
- ⁴ Swain, A., Narvaez, V., Burgoyne, P., Camerino, G. a Lovell-Badge, R.: DAX1 antagonises sry action in mammalian sex determination, *Nature* 391, 1998, s. 761 – 7
- ⁵ Hamilton, W. D.: Extraordinary sex ratios, *Science* 156, 1967, s. 477 – 88
- ⁶ Amos, W. and Harwood, J.: Factors affecting levels of genetic diversity in natural populations, *Philosophical Transactions of the Royal Society of London, Series B* 353, 1998, s. 177 – 86
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- ⁸ Haig, D.: Genetic conflicts in human pregnancy, *Quarterly Review of Biology* 68, 1993, s. 495 – 531
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- ¹² Bailey, J. M. a Pillard, R. C.: A genetic study of male sexual orientation, *Archives of General Psychiatry* 48, 1991, s. 1089 – 96; Bailey, J. M. a Pillard, R. C.: Genetics of human sexual orientation, *Annual Review of Sex Research* 6, 1995, s. 126 – 50

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- ¹⁵ Blanchard, R.: Birth order and sibling sex ratio in homosexual versus heterosexual males and females, *Annual Review of Sex Research* 8, 1997, s. 27 – 67
- ¹⁶ Blanchard, R. a Klassen, P.: H-Y antigen and homosexuality in men. *Journal of Theoretical Biology* 185, 1997, s. 373 – 8; Arthur, B. I., Jallon, J.-M., Caflisch, B., Choffat, Y. a Nothiger, R.: Sexual behaviour in *Drosophila* is irreversibly programmed during a critical period. *Current Biology* 8, 1998, s. 1187 – 90
- ¹⁷ Hamilton, W. D.: *Narrow roads of gene land*, I. zv., W. H. Freeman, Basingstoke, 1995

CHROMOZÓM 8

Jedným z najlepších zdrojov mobilných genetických elementov je opäť učebnica autorov Michaela Majerusa, Billa Amosa a Gregoryho Hursta: *Evolution: the four billion year war* (Longman, 1996). Dobrým príspevkom k vynálezu genetických „odtlačkov prstov“, či genetickej identifikácie je práca Waltera Bodmera a Robina McKieho *The book of man* (Little, Brown, 1994). Teóriu súťaže spermíí skúma kniha Tima Birkheada a Andersa Mollera *Sperm competition in birds* (Academic Press, 1992).

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- ⁶ Yoder, J. A., Walsh, C. P. a Bestor, T. H.: Cytosine methylation and the ecology of intragenomic parasites, *Trends in Genetics* 13, 1997, s. 335 – 40; Garrick, D., Fiering, S., Martin, D. I. K. a Whitelaw, E.: Repeat-induced gene silencing in mammals, *Nature Genetics* 18, 1998, s. 56 – 9
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CHROMOZÓM 9

Najlepším úvodom do darwinovskej medicíny a do vzájomného spolupôsobenia génov a patogénov je dielo Randy Nessa a Georga Williamsa *Evolution and healing* (Weidenfeld and Nicolson, 1995).

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- ² Yamamoto, F., Clausen, H., White, T., Marken, S. a Hakomori, S.: Molecular genetic basis of the histo-blood group ABO system, *Nature* 345, s. 229 – 33

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- ⁴ Gilbert, S. C., Plebanski, N., Gupta, S., Morris, J., Cox, M., Aidoo, M., Kwiatowski, D., Greenwood, B. M., Whittle, H. C. a Hill, A. V. S.: Association of malaria parasite population structure, HLA and immunological antagonism, *Science* 279, 1998, s. 1173 – 7; tiež A. Hill, ústne oznámenie.
- ⁵ Pier, G. B. *et al.*: *Salmonella typhi* uses CFTR to enter intestinal epithelial cells, *Nature* 393, 1998, s. 79 – 82
- ⁶ Hill, A. V. S.: Genetics of infectious disease resistance, *Current Opinion i Genetics and Development* 6, 1996, s. 348 – 53
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Očakávania a riziká genetického skríningu boli podrobne diskutované v mnohých knihách, článkoch a správach, ale iba niektoré sa ukázali byť podstatnými zdrojmi poznatkov. Jedným z nich je kniha Chandlera Burra *A separate creation: how biology makes us gay* (Bantam Press, 1996).

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