



Publication Analysis 2000-2006

Human Genetics



England has clearly dominated European human genetics research at the start of the 21st century. The particular success of mathematical geneticists is also remarkable. And then there is the case of an Icelandic company...

Under the headline “The 10 Hottest Nerds”, the US magazine, *Newsweek*, recently presented 10 candidates who they think (in their wisdom) might become the new “Einsteins of Biology”. Among them were two researchers who also made it into the top 30 list of the most cited European human geneticists presented here: Kari Stefansson (13th), founder and CEO of the Icelandic company, deCODE genetics, and Swedish citizen, Svante Pääbo (19th), director at the Max-Planck-Institute of Evolutionary Anthropology in Leipzig, Germany.

In the *Newsweek* article they were flanked by other “heroes” of the recent genomics revolution, such as Eric Lander, Craig Venter, George Church and David Botstein. Whether any of them will one day become “Einsteins of biology” (or if not them, who else?) is open to bets. For the time being, however, the *Newsweek* selection does at least serve to demonstrate the high impact that the field of human genetics has had on the public consciousness since the advent of genomics and its new technologies.

A new “Einstein” from human genetics?

One can be sure that bibliometric factors hardly played any role in selecting *Newsweek*'s ten candidates. Which of course is in complete contrast to our own analysis of European human genetics research where numbers of publications and citations are the sole criteria for ranking nations, papers and researchers.

Before discussing the results, however, first a few lines on the limitations of our analysis. As for all biomedical disciplines so far examined, the publication performance of the individual European countries between 2000 and 2006 is restricted to the specialist journals in the field. In this case these were the 131 journals listed under the category “Genetics and Heredity” in Thomson Scientific's “Journal Citation Reports”, including, for example, *Nature Genetics*, *The American Journal of Human Genetics* or *Genome Research*. Regrettably, we had to exclude multidisciplinary journals such as *Nature* and *Science* because Thomson Scientific's databases provide no tools with sufficient reliability to exclusively extract the human genetics articles. Indeed, the most prominent papers in the field could thus have been omitted from this part of the analysis. However, this limitation only applied for the analysis of the individual countries' performances (see tables p. 35) for

which we believe a survey of the expert journals provides sufficiently valid indicators for their overall productivity in human genetics research over the period 2000 to 2006. Nevertheless, for the rankings of the most-cited researchers and papers (see tables p. 36), the human genetics research publications in all journals were included.

Now for the results. Regarding the countries' overall publication and citation rates the clear winner is England. By numbers of publications as well as the numbers of their citations the distance to its second-placed, constant European “rival”, Germany, is considerably greater than in most other biomedical disciplines. Researchers from England published some 40% more articles in the specialist journals between 2000 and 2006, altogether collecting almost 42% more citations than the corresponding German articles. The average ratio of citations per article is, however, nearly the same for both countries – 14.9 for England versus 14.8 for Germany.

Close behind the European “Big Two” lies France in third place, but then there is a broad gap to the fourth, Italy. By overall citation numbers, the Netherlands (5th) almost caught up with Italy despite publishing significantly fewer articles in the specialist journals between 2000 and 2006. The reason is clear: The Dutch articles were more frequently cited on average than the Italian ones.

Iceland in the driver's seat

The best average ratios of citations per article among the top 20 countries were achieved by Scotland (16.1%), Austria (16.0%) and Switzerland (15.7%), a level similar to the average rate for US human genetics article.

Astoundingly, all of these nations were left far behind by a small country whose overall numbers of publications and citations were too low to make it into the top 20, Iceland. Between 2000 and 2006, researchers from the nordic island published only 174 articles in the human genetics specialist journals, but these were cited nearly 6,000 times to-date, yielding an average rate of 34 citations per article!

Nevertheless, the reason for this “success” soon becomes clear. Primarily it is the result of the activities of one of *News-*

week's ten prospective "new Einsteins": Kari Stefansson and his Reykjavik-based company, deCODE genetics, which works on the genetics of common complex diseases and the development of diagnostic tests. In the *Newsweek* article, Stefansson explained the concept of deCODE, as follows: "It is not just the new technology for genotype sequencing, it is also the understanding that to make discoveries you need information about people, their diseases and their health. In Iceland, we have so much data on the health care of people, and that has put us in the driver's seat."

England still growing stronger

Apart from Kari Stefansson (13th) two more scientists from deCODE made it into the list of the 30 most-cited European human geneticists: co-founder Jeffrey Gulcher (16th) and statistical geneticist, Augustine Kong (29th). A particularly noteworthy result, since as a commercial company, deCODE surely hasn't published all of its results in academic journals.

Back to the other European winner of this analysis, England. Four of the five most-cited European human genetics articles had correspondence addresses in England, only the paper in fourth place came from ... – guess who – ... deCODE genetics. Of

Europe...

Country	Citations	Articles	Cit./Art.
1. England	222,553	14,899	14.9
2. Germany	156,809	10,594	14.8
3. France	133,516	9,482	14.1
4. Italy	71,816	6,208	11.6
5. Netherlands	69,999	4,985	14.0
6. Sweden	47,274	3,018	15.7
7. Spain	41,959	3,966	10.6
8. Scotland	40,737	2,537	16.1
9. Switzerland	39,382	2,498	15.8
10. Belgium	31,835	2,441	13.0
11. Finland	30,813	1,984	15.5
12. Israel	30,388	1,980	15.4
13. Denmark	23,293	1,554	15.0
14. Austria	17,675	1,106	16.0
15. Norway	12,322	870	14.2
16. Wales	12,245	1,217	10.1
17. Russia	11,909	2,387	5.0
18. Ireland	11,076	1,096	10.1
19. Poland	8,243	932	8.8
20. Portugal	7,741	838	9.2

Articles which appeared between 2000 and 2006 in human genetics journals as listed by Thomson Scientific. Their citation numbers were recorded up until December 2007. A country's figures are derived from articles where at least one author working in the respective European nation is included in the author's list. Israel is included because it is a member of many European research organisations (EMBO, FEBS etc.), as well as participating in the EU Research Framework Programmes.

... and the world

	Citations	Articles	Cit./Art.
Europe	736,468	61,289	12.0
USA	909,424	56,943	16.0
Japan	134,021	10,384	12.9
Canada	100,078	7,717	13.0
Australia	56,786	4,642	12.3
China	27,154	3,313	8.2

the five most-cited researchers, four were working in England or Wales during 2000-2006, including the "Number One", Lon Cardon from the Wellcome Trust Centre of Human Genetics at Oxford University. The second most-cited scientist, Leena Peltonen, left the National Public Health Institute in Helsinki and joined the Wellcome Trust Sanger Institute in Hinxton, UK, in September 2007 as Head of Human Genetics. Perhaps one reason why England's human genetics research might perform even better in future years.

Altogether 11 researchers from England or Wales achieved a place in the European top 30 list of the most-cited authors in human genetics. Five were working in Germany, another four in French research institutions.

However, the number of citations which an individual researcher attracts with his or her publications doesn't reflect upon their mere quality alone. It also depends to a great extent on the respective "hotness" of a certain research field. The "hotter" a given field, the more people usually work in it – meaning there are potentially more people out there to cite your own papers.

Statisticians highly regarded

Seen from this perspective, two facts are particularly remarkable when looking behind the names of the top 30 most-cited human geneticists. Firstly, a conspicuously high portion of them are working in the subfield of neuro-psychiatric genetics, notably trying to understand the genetic basis of neurological and psychiatric disorders. And secondly, among the most highly-cited articles and researchers there is a strong handful dealing with basic statistical or bioinformatical analyses of broad genomic or genotyping data. See, for example, the first three most-cited papers, they all have Peter Donnelly (4th) as senior author and they all describe the application of statistical methods to the analysis of huge amounts of genotyping data.

Finally, a last word on the growing number of mega-author-papers in genomics, many of which clearly belong to the field of human genetics. These cause a dilemma when assigning citations to individual co-authors. Take, for example, the 2001 *Nature* paper on the "Initial sequencing and analysis of the human genome" by the International Human Genome Sequencing Consortium. To-date this has been cited almost 6,000 times! Yet, would it be fair to assign the total of 6,000 citations to every single one of the several hundred co-authors? Surely not. That's why, for our analysis, the citations of a paper were only assigned to one single co-author when it had less than fifty authors in total.



Publication Analysis 2000-2006 – Human Genetics

Most Cited Authors...

	Cit-ations	Art-icles
1. Lon R. Cardon , Wellcome Trust Ctr. Human Genet. Univ. Oxford	5,579	87
2. Leena Peltonen , Natl. Publ. Hlth. Inst., Biomedicum, Helsinki	5,560	168
3. Michael J. Owen , Neuropsychiat. Genet. Unit Univ. of Wales, Cardiff	5,236	179
4. Peter Donnelly , Wellcome Trust Ctr. Human Genet. Univ. Oxford	5,115	27
5. Douglas F. Easton , Canc. Res. UK Gen. Epidemiol. Cambridge	4,880	133
6. Stefan Schreiber , Dep. Med. & Clin. Mol. Biol. Univ. Kiel	4,827	157
7. Alexis Brice , Hosp. Pitié-Salpêtrière, INSERM, Paris	4,815	159
8. Richard R. Copley , Wellcome Trust Ctr. Human Genet. Univ. Oxford	4,641	35
9. Chris P. Ponting , Human Anat. & Genet., MRC, Univ. Oxford	4,566	66
10. Hans Lehrach , Max Planck Inst. Mol. Genet. Berlin	4,523	151
11. John A. Todd , Med. Gen. Cambridge Inst. Med. Res. Univ. Cambridge	4,435	104
12. Cornelia M. van Duijn , Epidemiol & Biostatist. Univ. Rotterdam	4,211	183
13. Kari Stefansson , deCODE genetics Reykjavik	4,158	80
14. Pak C Sham , Statist. & Psychiat. Genet. Kings Coll. Univ. London	4,108	162
15. Jean Weissenbach , Genoscope – Ctr. Natl. Séquençage, CNRS, Evry	4,049	86
16. Jeffrey Gulcher , deCODE genetics Reykjavik	4,008	72
17. Arnold Munnich , Genet. Hôpital Necker – Enfants Malades Paris	3,933	230
18. Jaako Kaprio , Natl. Publ. Hlth. Inst. Helsinki	3,872	190
19. Svante Pääbo , Max Planck Inst. Evol. Anthropol. Leipzig	3,872	74
20. Christine van Broeckhoven , Mol. Genet. Univ. Antwerp	3,700	173
21. Eamonn R. Maher , Div. Med. Genet. Dept. Pediat. Univ. Birmingham	3,657	120
22. Stylianos E Antonarakis , Div. Med. Genet. Sch. Med. Univ. Geneva	3,527	112
23. William O. Cookson , Wellcome Trust Ctr. Human Genet. Univ. Oxford	3,459	54
24. Ronald J.A. Wanders , Lab. Genet. Metab. Dis. Univ. Amsterdam	3,442	252
25. Yosef Shiloh , Human Genet. & Mol. Med. Tel Aviv Univ.	3,363	47
26. Michael C. O'Donovan , Neuropsychiat. Genet. Unit Univ. of Wales	3,327	116
27. Peter Lichter , German Canc Res Ctr Heidelberg	3,311	126
28. Christine Petit , Genet. Deficits Sensor. Inst. Pasteur, INSERM, Paris	3,295	112
29. Augustine Kong , deCODE genetics Reykjavik	3,270	42
30. Wolfgang Maier , Psychiat. Univ. Bonn	3,268	194



Lon Cardon (1.)



Leena Peltonen (2.)



Peter Donnelly (4.)



Cornelia van Duijn (12.)



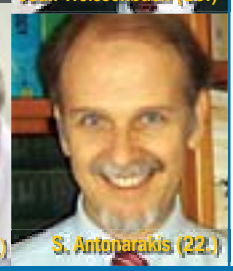
Kari Stefansson (13.)



Jean Weissenbach (15.)



C. van Broeckhoven (20.)



S. Antonarakis (22.)

Citations of articles published between 2000-6 were recorded until December 2007. The "most cited papers" had correspondence addresses in Europe or Israel.

... and Papers

	Citations
1. Pritchard JK, Stephens M, Donnelly P Inference of population structure using multilocus genotype data. <i>GENETICS</i> 155 (2): 945-959 JUN 2000	1,458
2. Stephens M, Smith NJ, Donnelly P A new statistical method for haplotype reconstruction from population data. <i>AMERICAN JOURNAL OF HUMAN GENETICS</i> 68 (4): 978-989 APR 2001	1,440
3. Stephens M, Donnelly P A comparison of Bayesian methods for haplotype reconstruction from population genotype data. <i>AMERICAN JOURNAL OF HUMAN GENETICS</i> 73 (5): 1162-1169 NOV 2003	653
4. Kong A, Gudbjartsson DF, Sainz J, Jonsson GM, Gudjonsson SA, ... , Gulcher JR, Stefansson K A high-resolution recombination map of the human genome. <i>NATURE GENETICS</i> 31 (3): 241-247 JUL 2002	624
5. Johnson GCL, Esposito L, Barratt BJ, Smith AN, Heward J, Di Genova G, ... , Gough SCL, Clayton DG, Todd JA Haplotype tagging for the identification of common disease genes. <i>NATURE GENETICS</i> 29 (2): 233-237 OCT 2001	617