

인간유전체 사업

권 오 주*†

Human Genome Project

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ABSTRACT

The completion of the rough draft of the human genome is a remarkable achievement. It provides the overall structures of huge DNA molecules that constitute the genome and an outline of the information needed to create a human being. This paper reviewed new ideas, projects, and scientific advances made by the Human Genome Project. We also discussed the future of medicine and biomedical research in postgenomic era.

KEY WORDS : Gene · Genome · Genomics · Proteome · Proteomics · Microarray · SNP.

136 1865 Mendel (“ Book of Life ”)
 , DNA (Avery 1944), 2 15 30
 DNA (Watson Crick 1953), DNA 1990 National Institutes of Health(NIH)
 (recombinant DNA technologies) U.S. Department of Energy가
 , 1993 Well-
 , 21 come Trust Sanger Center
 2001 2
 가 . 6 20 (Human Genome Organization,
 (International HUGO) , DNA
 Human Genome Sequence Consortium, “ Nobel James Watson
 ”) Celera Genomics 30 가
 Nature Science . 가
 DNA

인간유전체 사업의 배경

DNA
 (Human Genome Project;http://www.nh-
 gri.nih.gov/HGP) 30 , 가 가

*가
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, X Y
 (mutation) 가
 가 가 가 가
 (recombination)
 223
 가
 exon intron
 alternative splicing
 가 가
 (gene duplication) 99.9%가 0.1%
 (domain evolution) 가
 (bioinformatics) 10,000 8
 spliced variants 가
 (proteome)
 polymorphism(SNP) single nucleotide
 가 32
 (rearrangement) 250 SNP
 (domain motif) 140 SNP 2001 2
 7%
 1.1%(Celera Genomics) 1.5% 230 SNP가 (http://www.ncbi.nlm.nih.gov/SNP).
 () 가 SNP
 junk DNA 50% 가
 SNP가
 X 가 SNP 가
 junk 가 junk DNA가
 , junk DNA

유전체 정보의 이용

Celera Genomics
 300
 junk DNA가 800
 “Book of Life”
 . 1996
 mouse rat Bermuda
 1 2 kb
 10% Alu DNA 24 public
 junk DNA database GenBank “Bermuda Rule”
 가

GenBank
(http://www.ncbi.nlm.nih.gov).

가

Celera Genomics
(http://www.celera.com)

60 가

100 가 (2).

Celera Genomics

가

56

, mouse(mouse sequencing consortium,
Celera Genomics), rat(), (), (),
, chimpanzee(,) 가 .

database

2000 10 mouse sequencing consortium

2000

30

2001

5

30

500~1500

mouse

95%

7,500~15,000

2000

Celera Genomics

99%가

4,300

, 2001 6

가

NIH
20,000

Celera

parative genomics)

DNA

(com-

exon

앞으로의 연구 방향

가

2,000,

720

1 7,200

가

Applied Biosystems

(Perkin Elmer)

10

90%

15

가

DNA

Whitehead/MIT Genome Center

Sanger Center

가

50

, Watson Crick DNA
2003 4 25

0.01%

. 2001

1/3

10

가

, 2

20

Table 2. Sequenced organisms

Organism	Genome size (Mb)	Completion date	Estimated no. of genes
<i>H. influenzae</i>	1.8	1995	1,740
<i>S. cerevisiae</i>	12.1	1996	6,034
<i>C. elegans</i>	97	1998	19,099
<i>A. thaliana</i>	100	2000	25,000
<i>D. melanogaster</i>	180	2000	13,061
<i>M. musculus</i>	3,000	-	unknown
<i>H. sapiens</i> (Public)	3,200	-	31,780
<i>H. sapiens</i> (Celera)	3,200	-	39,114

chips software
 (, Applied Biosystems, Affymetrix, Infor-
 max) ; , service

(Incyte Genomics, Celera Geno-
 mics) ; ,

(Human Genome Sciences, Millennium)

가
 , 가
 (transcript)

가
 biotechnology

가
 가
 가
 가

“ ”

(proteome) ,

(proteomics) (Banks 2000 ; Naaby -
 Hansen 2001).

HUGO
 Human Proteome Organization

(HUPO) ,
 HUPO

가 (modification)

mass spectro-
 metry protein microarrays

가
 , Celera Incyte

가

가 -
 ,
 2001 600

인간유전체 사업이 의학에 미치는 영향

(3).

, DNA microarrays

가
 가
 microarrays
 (Aitman 2001 ; Zhu Snyder 2001).

1986 positional cloning

1,100
 가 (http://www.ncbi.nlm.nih.
 gov/omim).

(sequence - based)

가
 가 positional cloning

가
 가
 가
 가
 가 (Marcote
 2001).

Table 3. Paradigm shifts in biomedical research

Past and current	Current and future
Structural genomics	Functional genomics
Genomics	Proteomics
Map-based gene discovery	Sequence-based gene discovery
Monogenic disorders	Multifactorial disorders
Specific DNA diagnosis	Monitoring of susceptibility
Analysis of one gene	Analysis of multiple genes in gene families, pathways, or systems
Gene action	Gene regulation
Etiology (specific mutation)	Pathogenesis (mechanism)
One species	Several species

(prediction) (prevention)
 (Bailey 2001 ; Collins Mc-
 Kusick 2001). 가
 가
 (polymorphism) 가 , (personal pill) . 가 2010
 100 가 가 가
 가 가 가
 SNP 가 . 2020
 가 가 (Liggett 2001).
 SNP가 DNA “ (designed
 , SNP 1% drug) ”

결 론

linkage association
 (marker) SNP가 , “ postgenomic
 SNP가 가 . era ” . 1992
 , SNP Francis Collins 가
 (susceptibility) “ 가
 , SNP (pharmacogenomics) (not the end of genomics),
 가 (not even the beginning of the end)
 (It may be the end of the beginning) ”
 (tail- , 21 - ology - omics
 ored treatment) . ,
 가 .
 99.9% 가
 0.1% SNP가 . , 가 , 10 20
 , 가
 , 가 가
 , 가
 . 2000 “
 ”
 SNP
 SNP chip

중심 단어 :
 Microarray · SNP.

참고문헌

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(susceptibility)

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