

인간 유전체 정보 연구 및 활용에 있어서의 법적·윤리적 쟁점\*

구 영 모\*\*†

Legal & Ethical Issues in Human Genomics and Its Application\*

Young-Mo Koo, Ph.D.\*\*†

인간 게놈 프로젝트와 ELSI

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- 2) Limits of prenatal diagnostic services
- 3) Scope of newborn screening programs
- 4) Goals of population screening programs
- 5) Validity of predictive genetic tests
- 6) Predictive genetic testing of minors
- 7) Exclusionary uses of genetic test results
- 8) Workplace genetic screening and monitoring
- 9) Forensic and identification uses of genetics
- 10) Special issues of population genetic studies
- 11) Special issues of genetic family studies
- 12) Implication of behavioral and psychiatric genetics
- 13) Special issues of human gene transfer research
- 14) Argument for and against germ - line genetic engin-

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15) Challenges of regulating enhancement of genetics

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최근 미국 주의회에서 발의된 유전 정보에 관한 입법\*

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\*Murray & Mehlman eds. Encyclopedia of Ethical, Legal, and Policy Issues in Biotechnology vol. 1, John Wiley & Sons, Inc. 2000 pp428 - 435 HIPAA : Health Insurance Portability and Accountability Act of 1996 ERISA : Employee Retirement Income Security Act of 1974

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ELSI 관련 문헌

1) “Non-directive genetic counseling” dilemmas

- Clarke A(1998) : “Genetic screening and counselling” Kuhse H, Singer PA Companion to Bioethics , Blackwell publisher
- Gert B(1996) : “Applying Morality to the Nine Huntington Disease Cases : An Alternative Model for Genetic Counseling” *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Gert B(1996) : “Clarifying the Duties and Goals of Genetic Counselors : Implications for Nondirectiveness” *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Kuhse H, Singer P(1998) : “Genetic screening and counselling” *A Companion to Bioethics* , Oxford Univ Press
- Michie S, Marteau T(2000) : “Genetic counselling : some issues of theory and practice” Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge Univ Press
- Moreno JD(1998) : “Ethics committees and ethics consultants” Kuhse H, Singer PA Companion to Bioethics, Blackwell publisher
- Murray TH(1996) : “Educating Clinicians about Genetics” *The human Genome Project and the Future of Health Care*, Indiana Univ Press
- Seller MJ(1994) : “Genetic counseling” Gillon, R. *Principles of Health Care Ethics*, John Wiley & sons
- Walters L(2000) : “Screening and Counseling for Genetic Conditions : President’s Commission” Jonsen AR (eds) *Source Book in Bioethics*, Georgetown Univ Press

2) Limits of prenatal diagnostic services

- Green J, Anionwu E(2000) : “Psychosocial aspects of prenatal screening and diagnosis” Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge Univ Press
- Holm S(1998) : “Ethical issues in Pre-implantation diagnosis” Harris J, Holm S. *The Future of Human Reproduction : Ethics, Choice, and Regulation*, Clarendon press
- Hull R(1993) : “Fetal Monitoring, Sex Preselection, Nonreproductive Use of Fetuses, and Forced Medical Treatment of Pregnant Women” *Ethical Issues in the New Reproductive Technologies*, Wadsworth Publishing Co
- Kuhse H, Singer P(1998) : “Prenatal screening, sex selection and cloning” *A Companion to Bioethics*, Oxford Univ Press
- Robinson P(1998) : “Prenatal screening, sex selection and Cloning” Kuhse H, Singer PA Companion to Bioethics, Blackwell publisher

3) Scope of newborn screening programs

- Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers Ch. 3*, Beacon Press
- Walters L(2000) : “Genetic Screening : Programs, Principles, and Research” Jonsen AR (eds) *Source Book in Bioethics*, Georgetown Univ Press

4) Goals of population screening programs

- Harris J(1998) : “Screening and Discriminating” *Clones, Genes, and Immortality : Ethics and the Genetic Revolution*, Oxford Univ Press
- Penslar RL(1995) : “The Ethics of Genetic Screening and Testing” *Research Ethics : Case and Materials*, Indiana Univ Press

5) Validity of predictive genetic tests

- Davison C(2000) : “Predictive genetics : the cultural implication of supplying probable futures” Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge Univ Press
- Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers Ch. 3*, Beacon Press
- Gert B(1996) : “Ethical and Legal Dilemmas Arising During Predictive Testing for Adult-Onset Disease : The Experience of Huntington Disease” *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Gert B(1996) : “Morally Relevant Features of Genetic Maladies and Genetic Testing” *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Gert B(1996) : “The Concept of Genetic Malady” *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Marteau T, Anionwu E(2000) : “Evaluating carrier testing : objectives and outcomes” Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge Univ Press
- Wexler N(2000) : “Presymptomatic Testing for Genetic Disease” Pence GE *Classic Cases in Medical Ethics : Accounts of Cases*

## 6) Predictive genetic testing of minors

BMJ's Ethics, Science and Information Division "Children and Young People" Medical Ethics Today : Its Practice and Philosophy, BMJ

Clarke A, Flintner H(2000) : "The genetic testing of children : a clinical perspective" Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge University Press

Michie S(2000) : "Predictive genetic testing in children : paternalism or empiricism?" Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge university press

Murray TH(1996) : "The Genome Project and Health Services for Minority Populations" *The human Genome Project and the Future of Health Care*, Indiana Univ Press

Peters T(1997) : "The so called 'Gay gene' and Scientized Morality" *Playing God? : Genetic Determinism and Human Freedom*, Routledge

## 7) Exclusionary uses of genetic test results

### 8) Workplace genetic screening and monitoring

European Group on Ethics in Science and New Technologies to the European Commission(2000) : *Genetic Testing in the Workplace : Proceedings of the Round Table, Secretariat-General of the Commission*

Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers Ch. 10*, Beacon Press

Murray TH(1996) : "Genetics and Employment : More Disability Discrimination" *The human Genome Project and the Future of Health Care*, Indiana Univ Press

### 9) Forensic and identification uses of genetics

Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers Ch. 11*, Beacon Press

McEwen, Jean E(1998) : "DNA Banks" Rothstein, Mark A (Editor) *Genetic Secrets : Protecting Privacy and Confidentiality in the Genetic Era*, Yale University Press.

Murch Randall S, Budowle Bruce(1998) : "Are Developments in Forensic Applications of DNA Technology consistent with Privacy Protections?" Rothstein, Mark A (Editor) *Genetic Secrets : Protecting Privacy and Confidentiality in the Genetic Era*, Yale University Press

CODIS Brief History of Forensic DNA Typing

US Department of Justice(1999) : *Postconviction DNA Testing : Recommendations for Handling Requests*

US Department of Justice(2000) : *The Future of Forensic DNA Testing*

US Department of Justice(2000) : *What Every Law Enforcement Officer Should Know About DNA Evidence*

Zweig, Franklin M(eds)(1998) : "Courts and the Challenges of Adjudicating Genetic Testing's Secrets" Rothstein, Mark A(Editor) *Genetic Secrets : Protecting Privacy and Confidentiality in the Genetic Era*, Yale University Press

김낙권(1992) : 「군에서 유전자 감식과 유전자 은행 필요성」. 『국방저널』 302 : 110-115

김병수(2001) : 「국내 유전정보의 수집·이용 실태와 문제점」. 《시민배심원회의 전문가 패널발표 자료집》, 녹색연합·시민과학센터·환경운동연합

신경진 외(1999) : 「개인식별에 있어서 법의치과학적 방법과 유전자 검사법 활용」. 『대한구강내과학회지』 24(4) : 479-487

원혜욱(2001) : 「유전자감정결과의 증거사용에 관하여」. 『형사정책연구』 45 : 6-22

유영찬 외(1998) : 「경찰과학수사의 발전방안에 관한 연구」. 『치안논총』 14호

이승환(1996) : 「강력범죄의 해결사 DNA typing」. 『한국분자생물학회지』 8(2) : 28-37

한면수(1994) : 「최근 유전자 감정사례-유전자 자료은행 설립이 시급하다-」

한면수(1997) : 「유전자감식법에 의한 신원 확인」. 『항공우주의학』 7(4) : 16-27

한면수 DNA : 『프로필에 의한 개인식별과 FTA에 의한 검체의 관리』

### 10) Special issues of population genetic studies

Benatar SR. "A Perspective from Africa on Human Rights and Genetic Engineering"(1999) : Burley J, Dawkins R. *The Genetic Revolution and Human Rights : The Oxford Amnesty Lectures 1998 Oxford Univ Press*

### 11) Special issues of genetic family studies

Richard M(2000) : "Families, kinship and genetics" Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge University Press.

Thom D, Jennings M(2000) : "Human pedigree and the 'best stock' : from eugenics to genetics?" Marteau T, Richard M. *The Troubled Helix : social and psychological implications of the new human genetics*, Cambridge university press

## 12) Implication of behavioral and psychiatric genetics

- Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers* Ch. 7, Beacon Press
- Penslar RL(1995) : "Behavior Control" *Research Ethics : Case and Materials*, Indiana Univ Press
- Penslar RL(1995) : "Research Involving Human Subject : The Administration of Alcohol" *Research Ethics : Case and Materials*, Indiana Univ Press
- Tobin AJ(1999) : "Amazing Grace : Sources of Phenotype Variation in Genetic Boosterism" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Rowe DC, Jacobson KC(1999) : "In the Mainstream : Research in Behavioral Genetics" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Sherman SL, Waldman ID(1999) : "Identifying the Molecular Genetic Basis of Behavioral Traits" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Rothstein MA(1999) : "Behavioral Genetic Determinism : Its Effects on Culture and Law" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Andrews LB(1999) : "Predicting and Punishing Antisocial Acts : How the Criminal Justice System Might Use Behavioral Genetics" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Nelkin D(1999) : "Behavioral Genetics and Dismantling the Welfare State" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Duster T(1999) : "The Social Consequence of Genetic Disclosure" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press
- Carson RA(1999) : "The Fate of the Responsible Self in a Genetic Age" Carson RA, Rothstein MA. *Behavioral Genetics : The Clash of Culture and Biology*, Johns Hopkins Univ Press

## 13) Special issues of human gene transfer research

- Chadwick R(1998) : "Gene therapy" Kuhse H, Singer PA *Companion to Bioethics*, Blackwell publisher
- Gert B(1996) : "Ethics of Gene Therapy" *Morality and the new genetics : a guide for students and health care providers*, Jones and Bartlett Pub
- Kuhse H, Singer P(1998) : "Gene Therapy" *A Companion to Bioethics*, Oxford Univ Press
- Murray TH(1996) : "Medicine, Gene Therapy, and Society" *The human Genome Project and the Future of Health Care*, Indiana Univ Press
- Walters L(2000) : "Gene Therapy for Human Patients : Information for the Genral Public" Jonsen AR (eds) *Source Book in Bioethics*, Georgetown Univ Press
- Walters L(2000) : "Gene Therapy in Man : Recommendations of European Medical Research Council" Jonsen AR (eds) *Source Book in Bioethics*, Georgetown Univ Press
- Walters L(2000) : "NIH 'Point to Consider' for Gene Therapy Researchers" Jonsen AR (eds) *Source Book in Bioethics*, Georgetown Univ Press
- Walters L, Palmer JG(1996) : "Genes : Function and Heredity" *The Ethics of Human Gene Therapy*, Oxford Univ Press
- Walters L, Palmer JG(1996) : "Somatic Cell Gene Therapy" *The Ethics of Human Gene Therapy*, Oxford Univ Press

## 14) Argument for and against germ-line genetic engineering

- Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers* Ch. 8, Beacon Press
- Peters T(1997) : "The Question of Germline Intervention" *Playing God? : Genetic Determinism and Human Freedom*, Routledge
- Stock G, Campbell J(2000) : *Engineering the Human germline*, Oxford Univ Press
- Walters L, Palmer JG(1996) : "Germ line Cell Gene Therapy" *The Ethics of Human Gene Therapy*, Oxford Univ Press

## 15) Challenges of regulating enhancement of genetics

- Beauchamp TL, Walters L(1999) : "Eugenic and Human Genetics" *Contemporary Issues in Bioethics*, Wadsworth Pub Co
- Wade N(2001) : *Life Script : How the Human Genome Discoveries Will Transform Medicine and Enhance Your Health*, Simon & Schuster
- Walters L, Palmer JG(1996) : "Enhancement Genetic Engineering" *The Ethics of Human Gene Therapy*, Oxford Univ Press
- Weatherall D(1994) : "Human Genetic Manipulation" Gillon R. *Principles of Health Care Ethics*, John Wiley & sons

## 16) Issues in intellectual property and commercialization

- Harris J(1998) : *“Commercial Exploitation” Clones, Genes, and Immortality : Ethics and the Genetic Revolution*, Oxford Univ Press
- Harris J, Holm S(1998) : *“Sperm as Property” The Future of Human Reproduction : Ethics, Choice, and Regulation*, Clarendon press
- Holtug N(1998) : *“Creating and Patenting New Life Forms”* Kuhse H, Singer PA *Companion to Bioethics*, Blackwell publisher
- Hubbard R, Wald E(1999) : *Exploding the Gene Myth ; How Genetic Information is Produced and Manipulated by Scientists, Physicians, Employers, Insurance Companies, Educators, and Law Enforcers Ch. 9*, Beacon Press
- OTA(1989) : *New Developments in Biotechnology : Patenting Life - Special Report*
- Penslar RL(1995) : *“Intellectual Property” Research Ethics : Case and Materials*, Indiana Univ Press
- Peters T(1997) : *“Should We Patent God’s Creation?” Playing God? : Genetic Determinism and Human Freedom*, Routledge
- RAFI(2000) : *Phase II for Human Genome Research - Human Genetic Diversity Enters the Commercial Mainstream*
- Steiner H. *“Silver Spoons and Golden Genes”(1999) : Burley J, Dawkins R. The Genetic Revolution and Human Rights : The Oxford Amnesty Lectures 1998* Oxford Univ Press
- The Nordic Committee On Bioethics(1999) : *“Who Owns Our Genes?” Gary Taubes “Your Genetic Destiny for Sale” Technology Review, April 2001*
- Yoxen E(1998) : *The Gene Business : Who Should Control Biotechnology*, Oxford Univ Press