한반도선진화재단 공동체자유주의 세미나

정밀의료와 디지털 헬스케어, 의료서비스의 혁신 패러다임

2016.11.3

김종원 M.D. Ph.D.

성균관의대 삼성서울병원
진단검사의학과/유전 클리닉

연구에서 매출까지

보건의료 R&D 연구
기초연구
중개연구
비임상
임상
인허가
시장진입 및 환자적용
교육부
미래창조부
산업통상자원부
복지부
복지부
식약처
복지부
심사평가원
의료기관
의료기관
보건의료기술연구개발
기술개발
실용화
임상연구인프라조성
임상시험
임상연구
보험급여평가
의료서비스
연구중심병원
기술개발&실용화
의약품,의료기기... 개발
실용화&임상적용

오바마 대통령의 의회 연두 국정연설
정밀의료발의 (Precision Medicine Initiative) (2015.1)

"Tonight I'm launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes.

And to give us all access to the personalized information we need to keep ourselves and our families healthier."

President Barack Obama
2015 State of the Union Address | January 20, 2015

정밀의료발의 (Precision Medicine Initiative)

- 정밀의료
  - 개인의 유전자, 환경, 생활유형 차이에 따라 질환의 예방과 치료를 달리 하는 것

- 정밀의료 발의
  - 정밀의료의 개념을 실제 진료에 적용할 수 있도록 과학적인 증거들을 생산하는 것임.

왜 지금인가?

- 인간유전체 염기서열 분석 (sequencing of the human genome)
- 향상된 의생명 분석 기술
- 빅데이터를 이용한 새로운 분석 도구

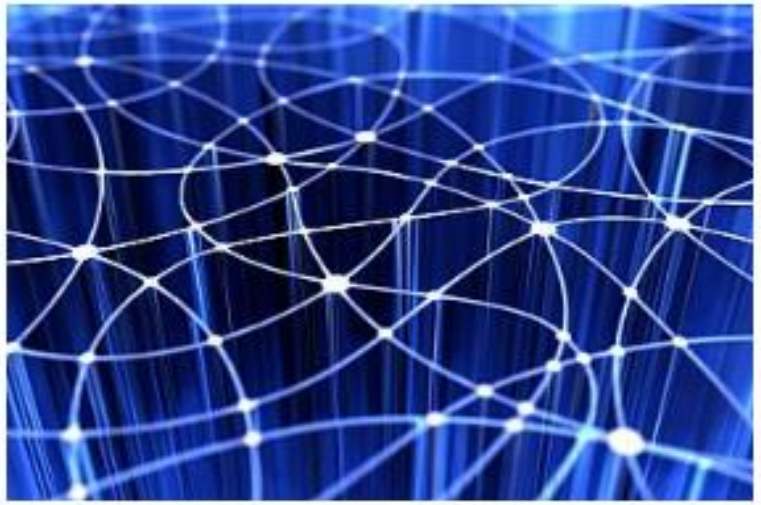
단기 목표: 암을 목표로 함.

- 새로운 암의 아형(subtype)과 치료 표적의 발굴
- 민간 부문과 공도으로 정밀 치료의 시도, 표적치료제(targeted drugs)
- 치료반응의 이해 확대
  - 약물내성을 극복하기 위한 지식 축적
  - 복합 치료(combination therapy)의 사용
  - 암의 재발을 예측하고, 모니터링

장기 목표

- 백만명 이상 자발 참여자를 대상으로 그들이 지정하는 전자 의무기록과 유전 자료, 검체, 식사/생활습관 정보를 공유하여 연구 코호트 (cohort)를 생성한다.
- 코호트 자료는
  - 약물유전체 연구에 기여 (the right drug for the right patients at the right dose)
  - 치료와 예방을 위한 새로운 표적 발굴
  - 이동기기가 건강 행동에 영향을 끼치는 지 검증
  - 많은 질환의 정밀의료를 위한 과학적 토대 구축

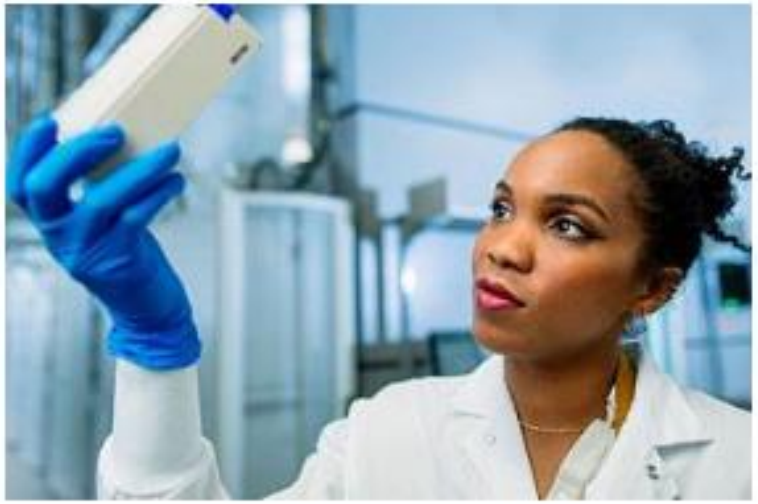
프로그램의 구성



Data and Research Center

Participant Technologies Center

Healthcare Provider Organizations



Biobank

연구 코호트(National Research Cohort)

과학 연구의 새로운 모델
자발적 참여
책임있는 자료공유
개인 비밀 보호

October 12, 2016

PMI ounces new
ch Program

called the *All*
rogram and will be the
gest health and medical research
program on precision medicine. A set of
core values is guiding its development and
implementation:

All of Us℠ | The Precision Medicine Initiative®
THE FUTURE OF HEALTH BEGINS WITH YOU

정밀의료가 실현되면 어떻게 되는가?

- 50세 여자가 제2형 당뇨로 외래를 방문했다.
- 현재
- 환자의 혈당이 잘 조절되지 않지만, 보통 2형 당뇨에 맞는 약처방을 한다.
- 손가락에서 채혈침으로 혈액을 채취하여 자가 혈당 측정을 한다.

정밀의료가 실현되면 어떻게 되는가?

- 50세 여자가 제2형 당뇨로 외래를 방문했다.
- 미래: 2년후
- 자원자들의 연구 네트웍
  - 자원자들이 자신의 DNA를 건강정보와 함께 연구자에게 보낸다.
  - Smartphone으로 자신의 건강 및 연구 결과를 볼 수 있다.
- 자원자의 혈당수준을 체내 매립형 칩으로 측정한 것을 스마트 손목시계를 통해 연구자의 컴퓨터에 전달하는 것을 자원자는 동의한다.
  - 이 자료를 통해, 환자는 식이와 약물 복용 일정을 조정한다.

정밀의료가 실현되면 어떻게 되는가?

- 50세 여자가 제2형 당뇨로 외래를 방문했다.
- 미래: 5년후
- 임상의사는 2형 당뇨에 진전된 연구 성과를 기반으로 한 신약에 대한 정보를 환자에게 준다.
- 환자가 신약의 이름을 스마트폰의 Rx 앱에 입력할 때, 환자의 유전체 정보는 해당 환자가 그 약물을 천천히 대사할 것으로 예상한다.
  - 따라서 의사는 약물 복용 일정을 변경한다.

정밀의료가 실현되면 어떻게 되는가?

- 50세 여자가 제2형 당뇨로 외래를 방문했다.
- 미래: 10년후
- 그녀의 60세 생일을 자축하고, 그녀가 코호트의 한 부분임을 자랑스러워 한다.
- 혈당 수준은 잘 유지되어, 당뇨 합병증을 격지 않는다.
- 환자의 자녀들은 코호트 연구에 자원자가 되기로 결정한다.

THE WALL STREET JOURNAL.

Biden's Cancer Research 'Moonshot' Needs Funding, White House Says

Obama administration wants Congress to fund $1 billion effort and provide other money

Oct. 17, 2016 5:32 p.m. ET

WASHINGTON—The White House on Monday said its "cancer moonshot" initiative is paying off in such areas as the digital storage of large groups of genetic data and research into environmental causes, but it urged Congress to approve funding for the venture.

The drive, headed up by Vice President Joe Biden, is aimed at attaining a decade's worth of new medical therapies and scientific knowledge about cancers within five years. The administration has been trying for months to get Congress to approve money for the $1 billion effort, along with funding for a precision-medicine initiative and an infusion of money for the National Institutes of Health.

Mr. Biden gave more details of the campaign's medical and technical accomplishments on Monday. He announced, for instance, a joint effort by the National Cancer Institute, Amazon Web Services and Microsoft Corp. to build a model for keeping the genetic data of large populations stored in the computer cloud.

Another project involves linking potentially 250,000 cancer and genetic cell samples from the Department of Defense to information stored at the Environmental Protection Agency to research environmental factors that cause or promote cancer growth.

THE WALL STREET JOURNAL.

IBM Offering U.S. Employees Watson Technology to Identify Cancer Treatments

IBM uses software in several oncology treatment centers globally but says it's the first time it will be available to U.S. employees

Oct. 11, 2016 8:00 a.m. ET

The computing giant today says it will offer its Watson artificial intelligence software to its U.S. employees to help them identify appropriate treatments and options for clinical trials. The benefit will be available beginning early next year to employees and their families …

The cancer-fighting service uses a mixture of artificial intelligence and human doctors. The program will use Watson for several cancer-related services, including one that matches eligible patients with relevant clinical trials for lung, breast, colorectal and gastric cancers…. Another service analyzes the genomic profile of a patient's tumor and provides doctors with information about potential cancer-causing mutations, which may benefit from targeted gene therapies, a promising area of cancer treatment.

Most of the Watson services will be free for patients, except for the genomic service, which involves lab fees;

……..

IBM has 377,000 workers world-wide……..

The company is also making Watson available to help workers navigate the thicket of health insurance options during its annual benefits enrollment period, Ms. Brickmeier said. Watson analyzes a worker's claims history and other factors to help guide employees toward the appropriate level of coverage, she said.

Parkinson's App Celebrates Milestone, Featured by Apple

Monday, March 21, 2016

Share 766 Tweet

A Parkinson's iPhone app developed by Sage Bionetworks and University of Rochester Medical Center (URMC) neurologists marks the first anniversary of its release. The app was also highlighted by Apple today during its semi-annual product launch event.

Sage Bionetworks, a Seattle-based nonprofit biomedical research organization, today released an updated version of its mPower (Mobile Parkinson's Observatory for Worldwide, Evidence-based Research) app that includes an improved user interface and functionality developed in response to feedback by users. Sage also announced that mPower would be the first app incorporated into a new Apple platform called CareKit, which will turn the app into a valuable tool to help better inform patients about their symptoms and care.

The mPower app – which was created by Sage in collaboration with URMC neurologists Ray Dorsey, M.D., M.B.A., and Karl Kieburtz, M.D., M.P.H., and with the support of the Robert Wood Johnson Foundation – was first unveiled in March 2015 during Apple's "Spring Forward" product launch event.

Parkinson's App Celebrates Milestone, Featured by Apple

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The mPower app gathers real time data from Parkinson's patients in an effort to more fully understand the disease and how it impacts daily life. The app also allows patients to track symptoms and how treatments are impacting the progression of the disease. In addition, the app uses other iPhone functions – such as the touch screen, motion sensors, and GPS – to measure dexterity, balance and gait, and memory multiple times per day.

"mPower allows researchers to follow day-to-day fluctuations in Parkinson's disease symptoms and allows for insights that would be impossible to achieve when a patient is only being examined every six months," said Stephen Friend, M.D., Ph.D., president of Sage Bionetworks. "This kind of data has never been tracked and captured before, and now with the help of CareKit, we can provide quantitative insights to inform the dialog a person has with a health professional about his or her own disease."

Since its launch, the app has been downloaded more than 60,000 times from Apple's App Store, and with more 12,000 registered users it represents one of the largest studies ever conducted in the disease…

Sage released a huge tranche of data from the more than 9,500 mPower users……..

한국에서
'혁신(Innovation)'
은 어떻게 일어나는가?

생명윤리 및 안전에 관한 법률

제25조 (유전자검사의 제한) ①유전자검사기관은 과학적 입증이 불확실하여 검사대상자를 오도(오도)할 우려가 있는 신체외관이나 성격에 관한 유전자검사 그 밖에 심의위원회의 심의를 거쳐 대통령령이 정하는 유전자검사를 하여서는 아니된다.

②유전자검사기관은 근이영양증 그 밖에 대통령령이 정하는 유전질환을 진단하기 위한 목적외에는 배아 또는 태아를 대상으로 유전자검사를 하여서는 아니된다.

③의료기관이 아닌 유전자검사기관에서는 질병의 진단과 관련한 유전자검사를 할 수 없다. 다만, 의료기관의 의뢰를 받아 유전자검사를 하는 경우에는 그러하지 아니하다.

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- Prostate Cancer
- Celiac Disease
- Crohn's Disease
- Hemochromatosis
- Restless Legs Syndrome
- Age-related Macular Degeneration
- Parkinson's Disease
- Coumadin® / Warfarin Sensitivity
- Plavix® Efficacy

Browse all 119 health and traits topics »

TIME's Best Inventions of 2008

From a genetic testing service to an invisibility cloak to an ingenious public bike system to the world's first moving skyscraper — here are TIME's picks for the top innovations of 2008

NEXT
View All

1. The Retail DNA Test

By ANITA HAMILTON Wednesday, Oct. 29, 2008

MORE

Before meeting with Anne Wojcicki, co-founder of a consumer gene-testing service called 23andMe, I know just three things about her: she's pregnant, she's married to Google's Sergey Brin, and she went to Yale. But after an hour chatting with her in the small office she shares with co-founder Linda Avey at 23andMe's headquarters in Mountain View, Calif., I know some things no Internet search could reveal: coffee makes her giddy, she has a fondness for sequined shoes and fresh-baked bread, and her unborn son has a 50% chance of inheriting a high risk for Parkinson's disease.

Learning and sharing your genetic secrets are at the heart of 23andMe's controversial new service — a $399 saliva test that estimates your predisposition for more than 90 traits and conditions ranging from baldness to blindness. Although 23andMe isn't the only company selling DNA tests to the public, it does the best job of making them accessible and affordable. The 600,000 genetic markers that 23andMe identifies and interprets for each customer are "the digital manifestation of you," says Wojcicki (pronounced Wo-jis-key), 35, who majored in biology and was previously a health-care investor. "It's all this information beyond what you can see in the mirror."

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United States Government Accountability Office

GAO

Testimony

Before the Subcommittee on Oversight and Investigations, Committee on Energy and Commerce, House of Representatives

For Release on Delivery
Expected at 9:30 a.m. EDT
Thursday, July 22, 2010

DIRECT-TO-CONSUMER GENETIC TESTS

Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices

Statement of Gregory Kutz, Managing Director
Forensic Audits and Special Investigations

GAO
Accountability * Integrity * Reliability

GAO-10-847T

질병 예측 검사의 평가

- 2010년 7월 질병 예측성 유전자를 시행하는 DTC genetic test에 관해 보고를 하였는데, 서비스를 제공하는 4개의 회사들 간의 검사 결과가 표준화가 되지 않았음을 지적함.

동일인을 4개 회사에 의뢰한 결과

Figure 1: Selected Contradictory Risk Predictions for Donor 1

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Female	37	Leukemia	Above average	Below average	Average	Not tested
		Breast cancer	Average	Above average	Average	Above average

Figure 2: Selected Contradictory Risk Predictions for Donor 2

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Female	41	Type 1 diabetes	Above average	Above average	Below average	Not tested
		Restless leg syndrome	Below average	Above average	Not tested	Average

Figure 3: Selected Contradictory Risk Predictions for Donor 3

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Male	48	Prostate cancer	Average	Average	Below average	Above average
		Hypertension	Average	Below average	Above average	Not tested

Figure 4: Selected Contradictory Risk Predictions for Donor 4

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Male	61	Celiac disease	Above average	Average	Not tested	Above average
		Multiple sclerosis	Below average	Average	Average	Below average

Figure 5: Selected Contradictory Risk Predictions for Donor 5

Gender	Age	Condition	Company 1	Company 2	Company 3	Company 4
Male	63	Heart attack	Average	Above average	Above average	Average
		Atrial fibrillation	Average	Below average	Average	Average

Testimony

- Congressional Testimony 2011
- Congressional Testimony 2010
- Congressional Testimony 2009
- Congressional Testimony 2008
- Congressional Testimony 2007
- Congressional Testimony 2006
- Congressional Testimony 2005
- Congressional Testimony 2004
- Congressional Testimony 2003
- Congressional Testimony 2002
- Congressional Testimony 2001
- Congressional Testimony 2000
- Congressional Testimony 1999
- Congressional Testimony 1998
- Congressional Testimony 1997
- Congressional Testimony 1996

Direct-to-Consumer Genetic Testing and the Consequences to the Public

Statement of

Jeffrey Shuren, M.D.
Director, Center for Devices and Radiological Health
Food and Drug Administration
Department of Health and Human Services

Before

the Subcommittee on Oversight and Investigations
Committee on Energy and Commerce
U.S. House of Representatives

July 22, 2010

INTRODUCTION

Good morning, I am Dr. Jeff Shuren, Director of the Center for Devices and Radiological Health (CDRH or the Center) at the Food and Drug Administration (FDA or the Agency). I am pleased to be here this morning to explain FDA's recent activities related to direct-to-consumer (DTC) genetic tests and our future plans for the regulation of laboratory-developed tests.

Scientific advances resulting from the Human Genome Project completed in 2003 have expanded our understanding of the genetic contribution to health and disease. These advances have also resulted in the development of new tests that can better identify individuals at risk for particular medical conditions and target medical treatments based on the likelihood that a patient will respond or experience an adverse event based on their individual genetic profile. FDA supports the promise and development of innovative genetic tests.

As Margaret A. Hamburg, M.D., Commissioner of Food and Drugs, and Francis S. Collins, M.D., Ph.D., Director of the National Institutes of Health, note in their jointly authored article entitled "The Path to Personalized Medicine," published in the June 15, 2010, *New England Journal of Medicine*, "Major investments in basic science have created

청문회후 Direct-To-Consumer 업체 진행

- 23andMe 는 지속적인 검사 활동및 TV 광고 선전
- Navigenics: Life Technologies에 인수 합병 (2012.7)
- DecodeMe & Decode Genetics: Amgen $415M 에 인수 합병됨. (2012.12)

23andMe, Inc.

Department

Nov 22, 2013

Ann Wojcicki
CEO
23andMe, Inc.
1390 Shoreline Way
Mountain View, CA 94

Document Number: G
Re: Personal Genome

Dear Ms. Wojcicki,

The Food and Drug A
Saliva Collection Kit a
of the Federal Food, D

This product is a devi
intended for use in the
prevention of disease
company's website at
PGS for providing "he
status," "health risks,"
to "take steps toward
cancer. Most of the int
device uses under se
require premarket app

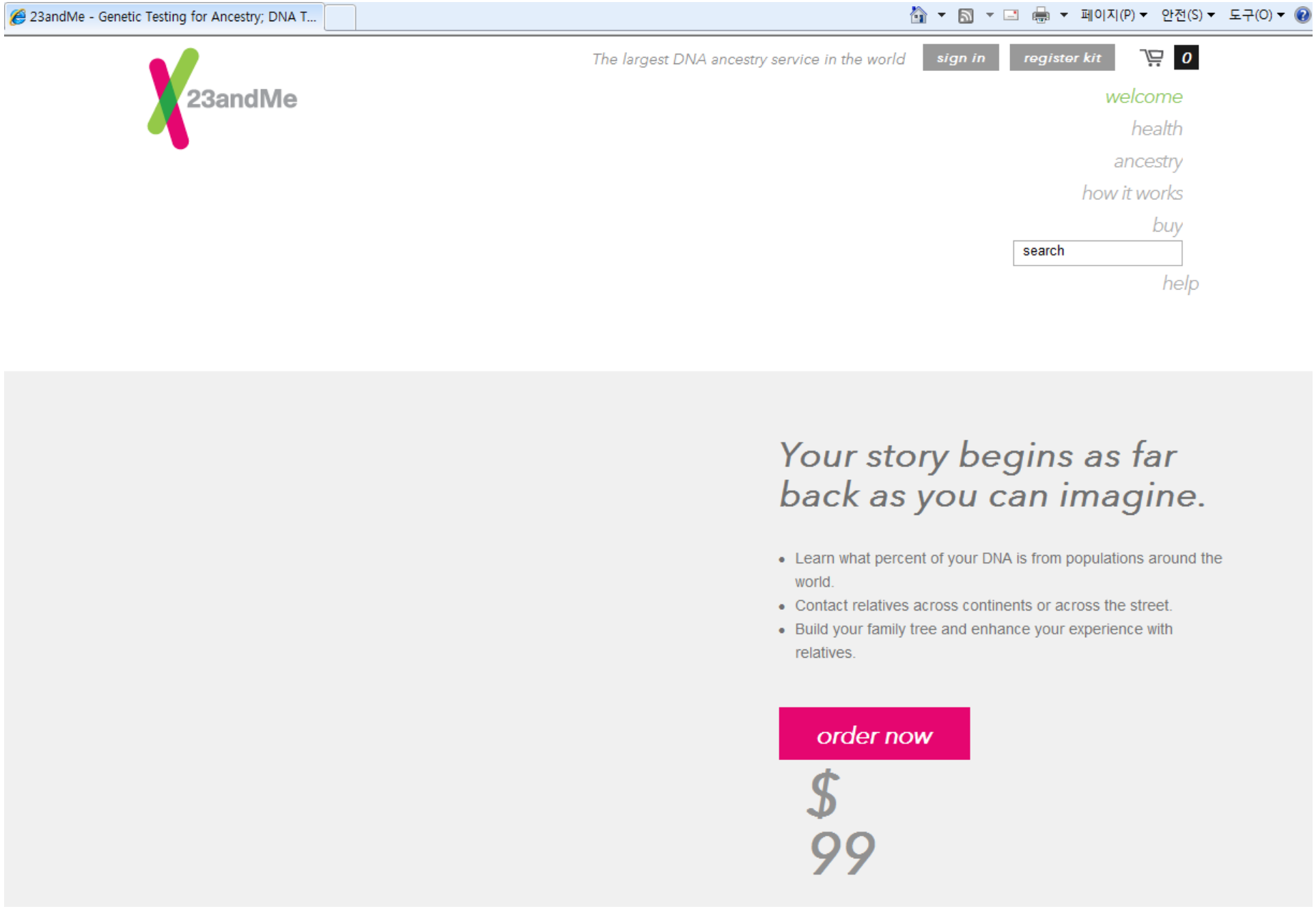
However, even after these many interactions with 23andMe, we still do not have any assurance that the firm has analytically or clinically validated the PGS for its intended uses, which have expanded from the uses that the firm identified in its submissions. In your letter dated January 9, 2013, you stated that the firm is "completing the additional analytical and clinical validations for the tests that have been submitted" and is "planning extensive labeling studies that will take several months to complete." Thus, months after you submitted your 510(k)s and more than 5 years after you began marketing, you still had not completed some of the studies and had not even started other studies necessary to support a marketing submission for the PGS. It is now eleven months later, and you have yet to provide FDA with any new information about these tests. You have not worked with us toward de novo classification, did not provide the additional information we requested necessary to complete review of your 510(k)s, and FDA has not received any communication from 23andMe since May. Instead, we have become aware that you have initiated new marketing campaigns, including television commercials that, together with an increasing list of indications, show that you plan to expand the PGS's uses and consumer base without obtaining marketing authorization from FDA.

Therefore, 23andMe must immediately discontinue marketing the PGS until such time as it receives FDA marketing authorization for the device. The PGS is in class III under section 513(f) of the FD&C Act, 21 U.S.C. 360c(f). Because there is no approved application for premarket approval in effect pursuant to section 515(a) of the FD&C Act, 21 U.S.C. 360e(a), or an approved application for an investigational device exemption (IDE) under section 520(g) of the FD&C Act, 21 U.S.C. 360j(g), the PGS is adulterated under section 501(f)(1)(B) of the FD&C Act, 21 U.S.C. 351(f)(1)(B). Additionally, the PGS is misbranded under section 502(o) of the Act, 21 U.S.C. § 352(o), because notice or other information respecting the device was not provided to FDA as required by section 510(k) of the Act, 21 U.S.C. § 360(k).

Please notify this office in writing within fifteen (15) working days from the date you receive this letter of the specific actions you have taken to address all issues noted above. Include documentation of the corrective actions you have taken. If your actions will occur over time, please include a timetable for implementation of those actions. If corrective actions cannot be completed within 15 working days, state the reason for the delay and the time within which the actions will be completed. Failure to take adequate corrective action may result in regulatory action being initiated by the Food and Drug Administration without further notice. These actions include, but are not limited to, seizure, injunction, and civil money penalties.

We have assigned a unique document number that is cited above. The requested information should reference this document number and should be submitted to:

James L. Woods, WO66-5688
Deputy Director
Patient Safety and Product Quality


23andMe - Genetic Testing for Ancestry; DNA T...
페이지(P)
안전(S)
도구(O)
23andMe
The largest DNA ancestry service in the world
sign in
register kit
0
welcome
health
ancestry
how it works
buy
search
help
Your story begins as far back as you can imagine.
Learn what percent of your DNA is from populations around the world.
Contact relatives across continents or across the street.
Build your family tree and enhance your experience with relatives.
order now
$
99

해외 사례에 대한 검토 (미국) (1)

23andMe

Box for 23andMe's direct-to-consumer genetic test.

After the Food and Drug Administration (FDA) forced 23andMe to stop marketing the health reports in its spit kits in 2013, the personal genomics company has now been approved to offer its tests directly to consumers, according to a press release posted today on the company's website. It is the first company to have FDA approval to market genetic tests directly to consumers, without a doctor's approval or intervention.

Not all the results offered by 23andMe are the same now as they were in 2013, as they can only run the tests that have been approved by the FDA. The service will continue to provide customers with, "the chance to find and connect with DNA relatives in a database of more than 1 million customers," as noted in the press release.

But it's offering some new services, too. In addition to its sleek new user interface, the company offers a genetic carrier screening, a test that can pick up on mutations that prospective parents may pass on to their offspring. The test will provide information on 36 inherited disorders, according to the New York Times. The new test will also reportedly cost $199, $100 more than it did in 2013.

Although the tests are now designated as diagnostic tools by the FDA, some experts are still concerned that the direct-to-consumer model still doesn't provide users with enough information to truly understand their results. "While [the] FDA has approved carrier tests there still isn't infrastructure to support the consumption of that information by consumers, and that's where I have concerns," Dietrich Stephan, a genetics professor at the University of Pittsburgh Medical Center told the Genetic Experts News Service.

해외 사례에 대한 검토 (미국) (2)

FDA Warns Three Companies Over DTC Genetic Tests

Posted 09 November 2015

By Michael Mezher

Last week, the US Food and Drug Administration (FDA) sent letters to three genetic testing companies for marketing unapproved direct-to-consumer (DTC) tests.

The letters are addressed to three companies: DNA4Life, DNA-CardioCheck, Inc. and Interleukin Genetics, Inc., which FDA says are marketing genetic tests to consumers without being cleared.

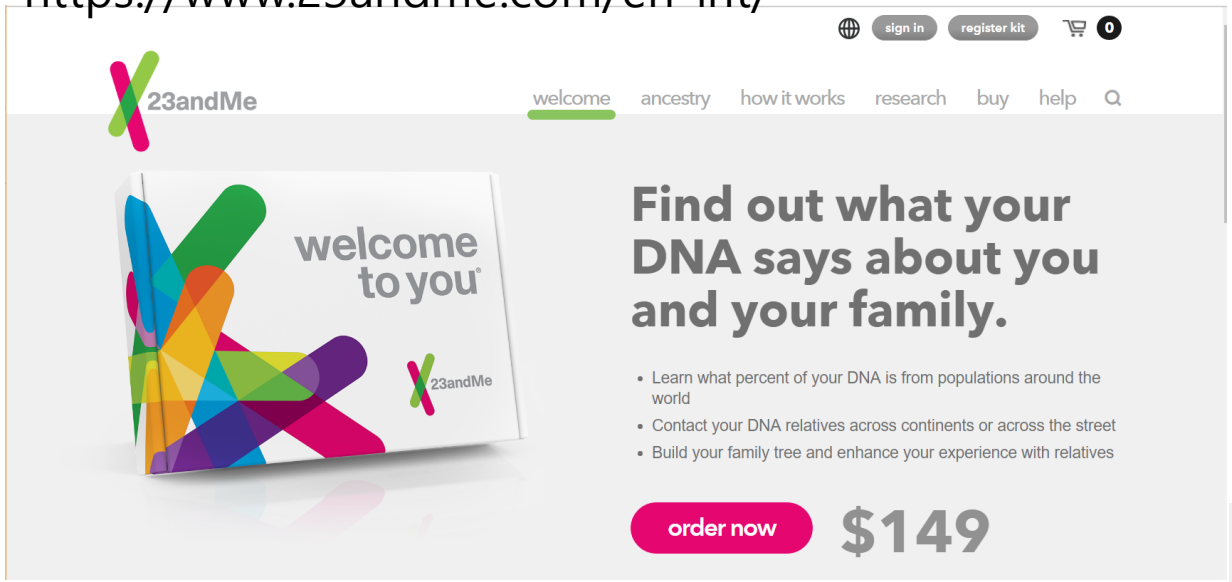
In each of the letters, James Woods, FDA Deputy Director for the Patient Safety and Product Quality Office of In Vitro Diagnostics, says the agency is "unable to identify any … clearance number" for the tests cited, and instructs the companies to either show that the tests have been cleared, or provide their rationale for why the tests do not need to be cleared.

FDA Press Officer Eric Pahon told *Focus* that "FDA appreciates that many consumers would like to be informed about their genomes, and their genetic risk for development of future disease. We agree that access to tests through a DTC model can allow consumers to take responsibility for certain aspects of their health, and to learn more about genetics and its contributions to risk, among other probable benefits. We believe that certain types of tests are being appropriately offered through the DTC model, but others may need to demonstrate that they are safe and effective and that appropriate controls are in place to mitigate risks."

FDA Letters to Genetic Testing Companies

Company	Test Name	Indication or Claim
DNA4Life	Pharmacogenetic Report	"Intended to predict how patients will respond to more than 120 of the most commonly prescribed medications."
DNA-CardioCheck, Inc.	DNA-CardioCheck	"Intended to test for DNA genetic markers linked to thrombophia, deep vein thrombosis, cardiovascular disease and stroke."
Interleukin Genetics, Inc.	PerioPredict Genetic Test, Osteoarthritis Genetic Test, Weight Management Genetic Test	"Intended to identify individuals with genetic predisposition for increased risk to diabetes and heart attack, osteoarthritis associated conditions, and obesity-related genotype for weight loss."

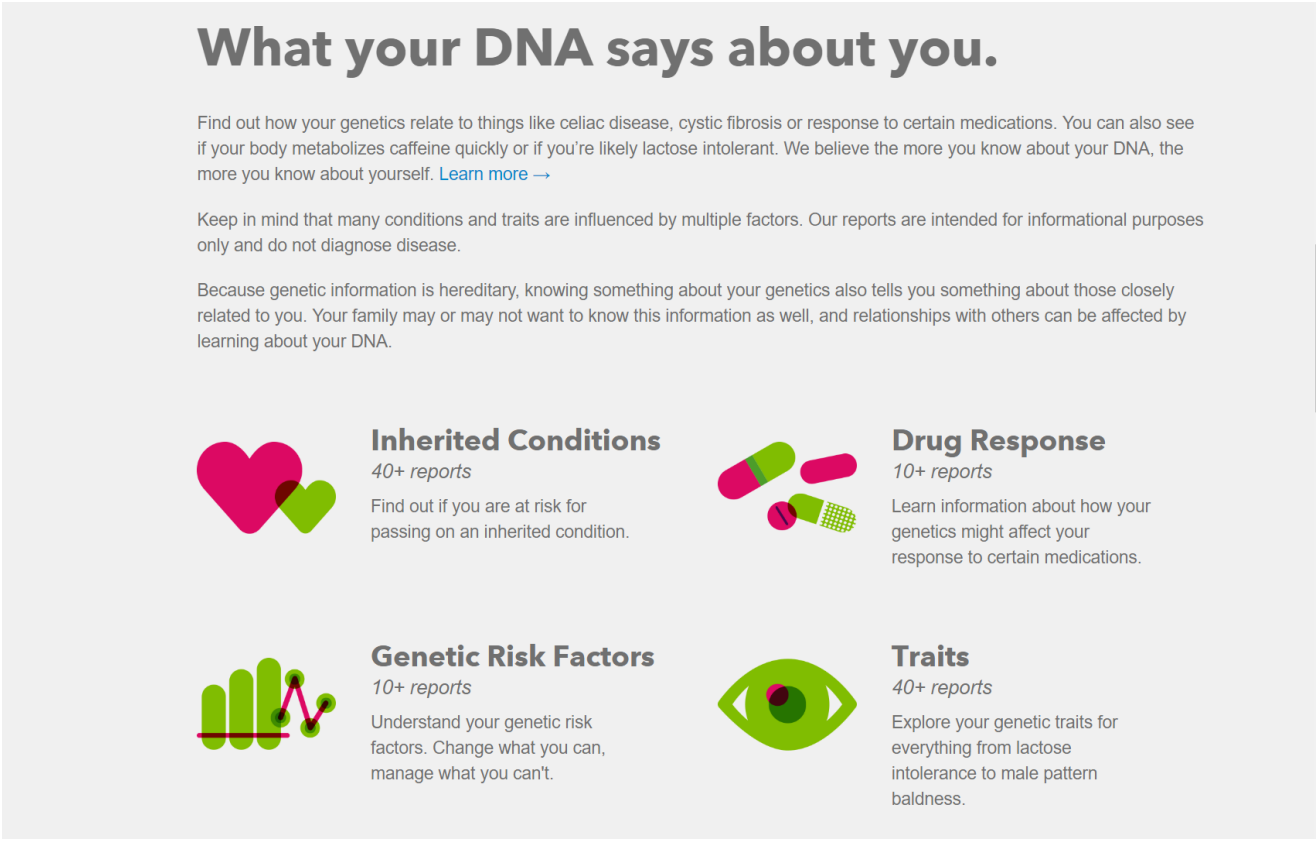
https://www.23andme.com/en-int/



미국에서는 제한된 유전질환 보인자 검사를 시행하고 있다.

미국 이외에는 해당 국가에 따라 유전 예측검사를 하고 있다.

https://www.23andme.com/en-ca/



한국의 상황

Research Article

Comparison of Commercial Genetic-Testing Services in Korea with 23andMe Service

Sollip Kim, Ki-Won Eom, Chong-Rae Cho, and Tae Hyun Um

Department of Laboratory Medicine, Ilsan Paik Hospital, Inje University College of Medicine, Joowha-ro 170, Ilsanseo-gu, Goyang, Gyeonggi 411-706, Republic of Korea

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Introduction. Genetic testing services for disease prediction, drug responses, and traits are commercially available by several companies in Korea. However, there has been no evaluation study for the accuracy and usefulness of these services. We aimed to compare two genetic testing services popular in Korea with 23andMe service in the United States. *Materials and Methods*. We compared the results of two persons (one man and one woman) serviced by Hellogene Platinum (Theragen Bio Institute), DNAGPS Optimus (DNAlink), and 23andMe service. *Results*. Among 3 services, there were differences in the estimation of relative risks for the same disease. For lung cancer, the range of relative risk was from 0.9 to 2.09. These differences were thought to be due to the differences of applied single nucleotide polymorphisms (SNPs) in each service for the calculation of risk. Also, the algorithm and population database would have influence on the estimation of relative disease risks. The concordance rate of SNP calls between DNAGPS Optimus and 23andMe services was 100% (30/30). *Conclusions*. Our study showed differences in disease risk estimations among three services, although they gave good concordance rate for SNP calls. We realized that the genetic services need further evaluation and standardization, especially in disease risk estimation algorithm.

Table 2: Comparison of relative risks among 3 genetic tests.

Category	Relative risk for participant 1 (man)			Relative risk for participant 2 (woman)		
	Hellogene Platinum	DNAGPS Optimus	23andMe	Hellogene Platinum	DNAGPS Optimus	23andMe
Lung cancer	2.09	1.97	0.8*	2.09	0.92	0.8*
Esophageal squamous cell carcinoma	1.25	2.41	1.21	0.72	0.52	0.8
Colorectal caner	1.36	0.73	1.1	0.98	0.97	1.1
Bladder cancer	0.77	0.8	Typical	0.77	0.9	Typical/lower
Breast cancer	NA	NA	NA	0.84	0.77	0.98
Atopic dermatitis	1.16	1.13	Typical	1.16	0.74	Lower
Rheumatoid arthritis	NA	NA	0.71	0.97	2.95	1.1
Systemic lupus erythematosus	NA	NA	NA	0.97	1.47	1.09
Psoriasis	0.97	0.46	0.87*	0.69	1.06	0.44*
Diabetes mellitus, type 1	0.77	0.97	0.08*	1.11	1.08	0.1*
Body mass index (obesity)	0.91		0.85*	0.91		0.83*
Atrial fibrillation	0.94	0.68	1.12*	0.94	1.39	1.93*
Coronary artery disease	1.06	0.51	0.78*	0.62	0.34	0.71*
Parkinson's disease	1.09	0.84	1.05	0.95	1.04	0.77
Narcolepsy	1.41	1.29	Lower*	1.41	1.29	Typical*
Warfarin maintenance dose	Normal dose	Moderate dose	Decreased dose	Normal dose	Increased dose	Slightly decreased dose

NA: not applicable.
* Relative risk in European. There was no Asian data.

S Kim et al, BioMed Research International 2014, ID 539151

The New York Times

May 14, 2013

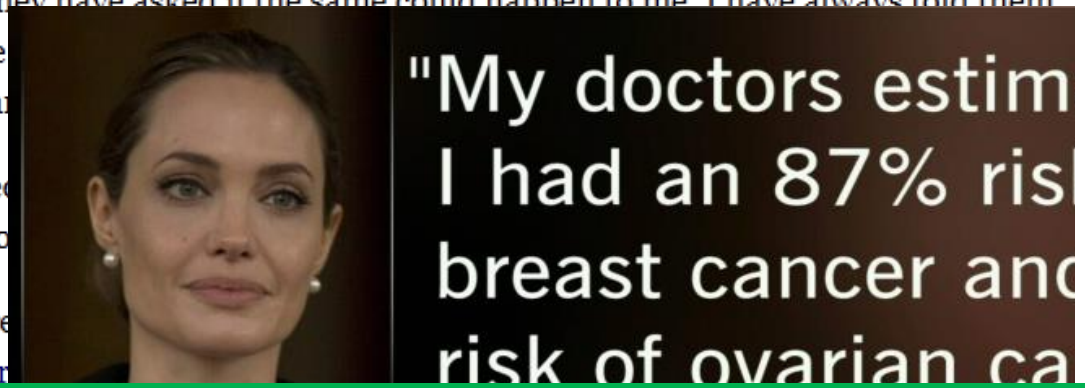
My Medical Choice

By ANGELINA JOLIE

LOS ANGELES

MY MOTHER fought cancer for almost a decade and died at 56. She held out long enough to meet the first of her grandchildren and to hold them in her arms. But my other children will never have the chance to know her and experience how loving and gracious she was.

We often speak of "Mommy's mommy," and I find myself trying to explain the illness that too her away from us. They have asked if the same could happen to me. I have always told them not to worry, but the developing breast ca



My doctors estimate ovarian cancer, altho

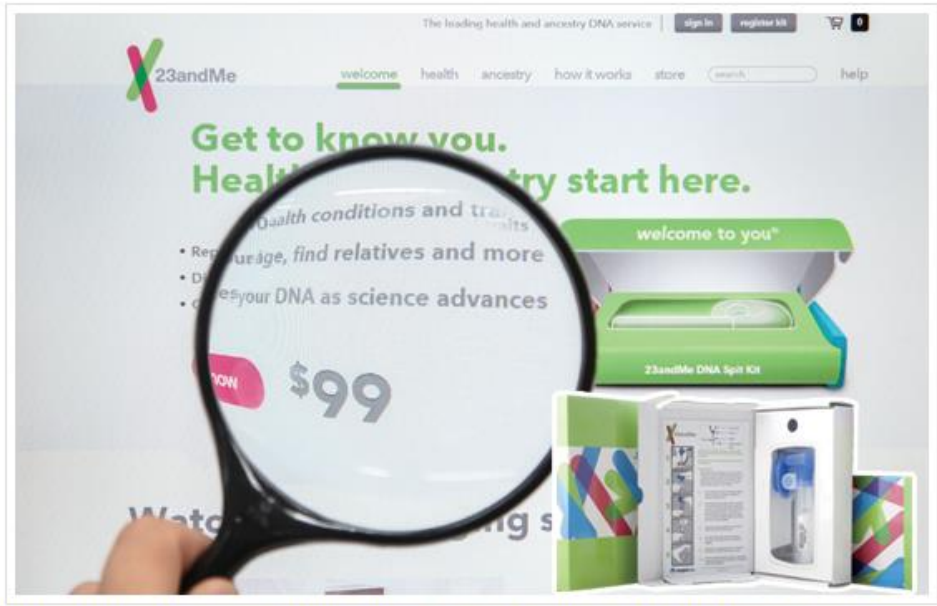
Only a fraction of bre BRCA1 have a 65 per

매경이코노미 제1726호(13.10.02~10.08일자) 기사

MK 뉴스 인쇄하기

[유전자 비즈니스의 세계]기술 진일보된 유전자 진단…피 한 방울이면 유전자 진단 완료

기사입력 2013.10.07 09:38:42 | 최종수정 2013.10.07 18:21:33



BRCA1/2 유전자 검사는 이미 의료기관에서 수행하고 있는 검사이다.
23andMe에서 제공하는 유전자검사는 BRCA 유전자를 포함하지 않는다.
23andMe에서 제공하는 유전자검사는 다인자 질환(암, 심장병등)의 예측검사이다.

생명윤리 및 안전에 관한 법률 (개정 2015.12.29)

제50조(유전자검사의 제한 등) ① 유전자검사기관은 과학적 증명이 불확실하여 검사 대상자를 오도(誤導)할 우려가 있는 신체 외관이나 성격에 관한 유전자검사 또는 그 밖에 국가위원회의 심의를 거쳐 대통령령으로 정하는 유전자검사를 하여서는 아니 된다.

② 유전자검사기관은 근이영양증이나 그 밖에 대통령령으로 정하는 유전질환을 진단하기 위한 목적으로만 배아 또는 태아를 대상으로 유전자검사를 할 수 있다.

③ 의료기관이 아닌 유전자검사기관에서는 다음 각 호를 제외한 경우에는 질병의 예방, 진단 및 치료와 관련한 유전자검사를 할 수 없다. <개정 2015.12.29.>

1. 의료기관의 의뢰를 받은 경우

2. 질병의 예방과 관련된 유전자검사로 보건복지부장관이 필요하다고 인정하는 경우

④ 유전자검사기관은 유전자검사에 관하여 거짓표시 또는 과대광고를 하여서는 아니 된다. 이 경우 거짓표시 또는 과대광고의 판정 기준 및 절차, 그 밖에 필요한 사항은 보건복지부령으로 정한다.

의료기관이 아닌 유전자검사기관이 직접 실시할 수 있는 유전자검사 항목에 관한 규정

제정 2016. 6. 20. 보건복지부고시 제2016-97호

의료기관이 아닌 유전자검사기관이 실시할 수 있는 유전자검사의 내용은 다음 각 목과 같다.

가. FTO, MC4R, BDNF 유전자에 의한 체질량지수 유전자검사

.....

바. OCA2, MC1R 유전자에 의한 색소침착 유전자검사

사. chr20p11(rs1160312, rs2180439), IL2RA, HLA-DQB1 유전자에 의한 탈모 유전자검사

아. EDAR 유전자에 의한 모발굵기 유전자검사

자. AGER 유전자에 의한 피부노화 유전자검사

차. MMP1 유전자에 의한 피부탄력 유전자검사.....

2. 제1호자목부터 타목까지의 유전자검사 내용은 이 고시 시행일로부터 2년 후 그 검사의 적정성 여부를 재검토하여야 한다.

3. 의료기관이 아닌 유전자검사기관은 제1호에 따른 유전자검사를 실시하는 경우 검사결과의 한계, 과학적 근거 등을 결과지에 명시하고 검사대상자에게 충분히 설명하여야 한다.

4.「훈령·예규 등의 발령 및 관리에 관한 규정」(대통령훈령 제334호)에 따라 이 고시 발령 후의 법령이나 현실여건의 변화 등을 검토하여 이 고시의 폐지, 개정 등의 조치를 하여야 하는 기한은 2019년 6월 30일까지로 한다.

부 칙

이 고시는 2016년 6월 30일부터 시행한다.

차. MMP1 유전자에 의한 피부탄력 유전자검사

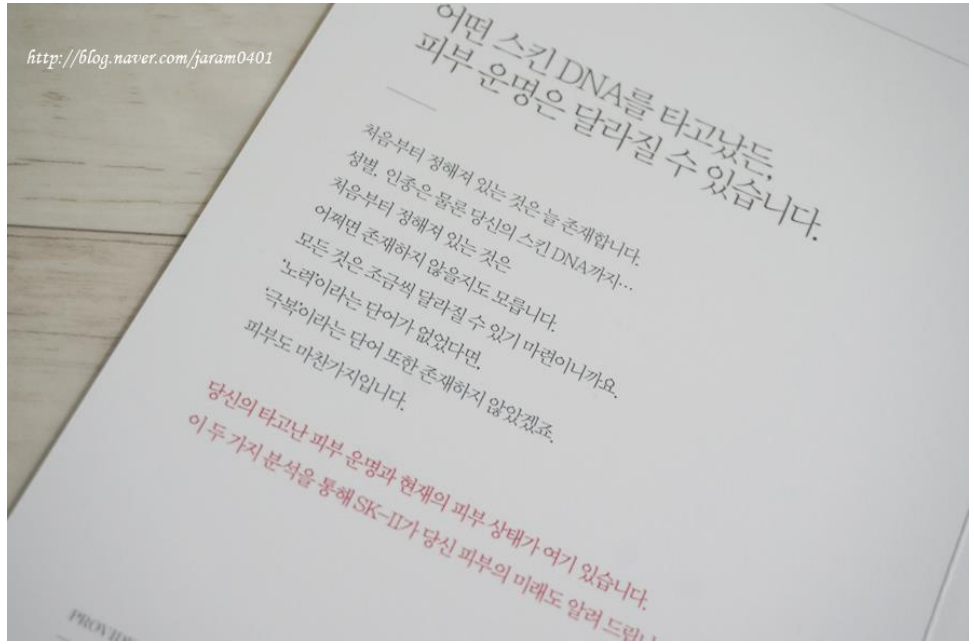
Matrix metalloproteinase-1 and skin ageing in smokers

Christine Lahmann, Jörg Bergemann, Graham Harrison, Antony R Young

Smokers look older than non-smokers of the same age. We have compared the concentrations of mRNA for matrix metalloproteinase 1 (MMP-1) in the buttock skin of smokers and non-smokers with quantitative real-time polymerase chain reactions. MMP-1 degrades collagen, which accounts for at least 70% of the dry weight of dermis. We report significantly more MMP-1 mRNA in the skin of smokers than non-smokers whereas no difference was seen for the tissue inhibitor of metalloproteinases 1 (TIMP-1) or the housekeeping gene *GAPDH* (glyceraldehyde-3-phosphate dehydrogenase). We suggest that smoking-induced MMP-1 might be important in the skin-ageing effects of tobacco smoking.

Lancet. 2001 Mar 24;357(9260):935-6.

http://blog.naver.com/jaram0401



http://blog.naver.com/jaram0401

| 스킨 DNA 분석

톤과 기미에 관련된 GPX1 유전자는 우수하고 탄력에 영향을 주는 항산화 유전자 SOD2는 양호하나, 피부결과 주름에 주로 관여하는 MMP1 유전자는 취약한 편. 따라서 피부결과 주름을 제외한 피부결, 광채, 기미, 탄력은 우수한 피부 유전자로 분석됨

| 현재 피부 상태

피부의 5가지 요소(피부결, 광채, 기미, 주름, 탄력)가 균형 있게 우수하게 잘 관리된 상태임

| SK-II 솔루션

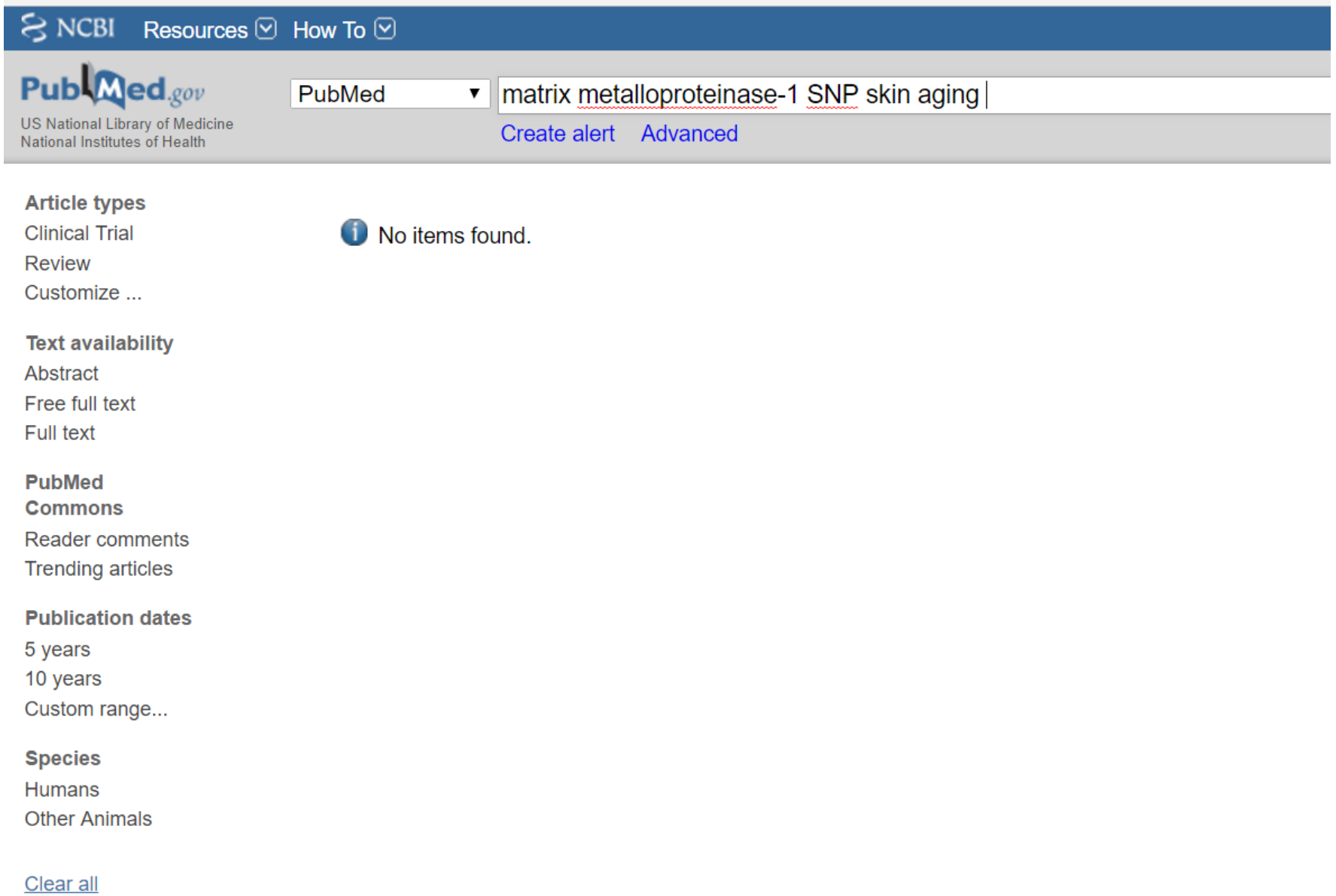
[1] **장기적인 케어가 필요한 부분** 40대가 되면 건조함, 모공 처짐, 뾰루지등을 동반한 심각한 피부결 트러블이 나타날 수 있으니 지속적인 관리가 필요함

[2] **시급한 케어가 필요한 부분** 현재 기미 부분이 잘 관리되고 있으나, 타고난 피부가 기미가 잘 생기지 않는 유전자를 가지고 있으므로 조금만 더 신경써서 집중적인 관리를 시작하면 비교적 빠른 시간에 더욱 호전된 가능성이 있음

[3] **꼭 필요한 제품** 페이셜 트리트먼트 리페어 C, 페이셜 트리트먼트 에센스, 스템파워 크림

http://blog.naver.com/jaram0401

Pubmed 분석 결과: (2016.10.31): 찾을 수 없었음.



보건복지부 장관 고시 항목의 문제

- 일부 항목에서 관련성은 과학적으로 인정되고 있다.
- 고시된 모든 종목의 임상적 유용성은 확인되지 않았다.

과학적 관련성과 임상적 유용성은 같은 의미인가?

예측 유전검사의 임상 적용 개발 단계

위험 유전 인자의 발굴

GWAS, 유전체 염기서열 분석 등 유전체 연구 방법 사용

상대 위험도의 계산

A 유전인자가 있는 사람이 없는 사람보다 Y 배 질환 위험도가 높다.

절대 위험도의 계산

A 유전인자를 가진 사람은 Z% 의 발병 위험도를 가진다.

위험도 예측 모델 개발

A,B,C.....유전인자와 P,Q,R 환경, 생활 습관 인자의 복합계산 모델 개발

실제 대상 인구에 적용하여 효용성(utility) 평가

고시된 항목은 모두 이 단계 혹은 이전 단계에 있다.

Adapted from Chatterjee et al, Nature Review Genetics 2016(7)

DTC(Direct-To-Consumer) 검사의 고려사항

1. 검사 (Genetic Test)
    1. 분석 단계 (Analytical Phase)

        #1. 분석결과는 정확해야 한다.

    2. 해석 단계 (Interpretation Phase)

        #2. 해석은 명확해야 한다.

DTC(Direct-To-Consumer) Test의 사례

- 임신 반응 검사, 혈당 검사
  1. 검사의 예민도(sensitivity), 특이도(specificity)의 평가와 평가 정보 제공
  2. 결과 수치의 표준화: 기기에 따른 편차가 없어야 함. 편차 정보의 제공
  3. 사용자에 따른 오차 범위 제거: 사용자에 따라 허용 오차 기준 이내 충족

혈당 측정기 (예)

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혈당 측정기 (예)

ACCU-CHEK® Performa

Controls

REF 04861736

(EN)

Suitable for self-testing/For in vitro diagnostic use

Intended Use

For performance checks on the Accu-Chek Performa, Accu-Chek Performa Nano, Accu-Chek Performa Connect, Accu-Chek Performa Combo, and Accu-Chek Performa Insight systems with the Accu-Chek Performa test strips. For performance checks on the Accu-Chek Performa (with code chip slot), Accu-Chek Performa Nano, and the Accu-Chek Inform II systems with the Accu-Chek Inform II test strips.

Testing control solutions with known glucose levels establishes that the operator and the system are performing acceptably. Control results must be within the defined acceptable ranges before valid patient testing is allowed.

Important Information

WARNING: Choking hazard. Small parts. Keep away from children under the age of 3 years.

DO NOT INGEST! Seek immediate medical attention if swallowed.

Contents of the pack

Pack containing 2 × 2.5 mL control solution and package inserts. The Accu-Chek Performa control solution pack contains two control solutions, one for the hypoglycaemic range (control solution 1, grey cap) and one for the hyperglycaemic range (control solution 2, white cap).

Disposal

All components of the pack can be discarded in domestic waste. Consult local ordinances as they may vary by country. Because the reactive substances are in such small quantities, they are not considered hazardous materials under EU regulations. If you have any questions, contact the local Roche representative.

Control solution storage and handling

- Refer to the meter User's Manual for system operating conditions.
- The printed use by date is valid if the unopened control solutions are stored between 2 and 32 °C.
- The control solutions do not need to be kept in the refrigerator. DO NOT freeze the control solution.
- Write the date the control solution bottle was opened on the bottle label. The control solution must be discarded 3 months from the date the control solution bottle was opened (discard date) or on the use by date on the bottle label, whichever comes first.
- Control solutions taken directly from the refrigerator must

4. Was the test strip used immediately after it was removed from the test strip container?
5. Were the test strips and control solutions stored in a cool, dry place?
6. Did you follow the directions?
7. Was the correct control solution level selected when the test was performed?

Testing Intervals

Follow your facility's policy for control testing intervals.

Control tests should be performed:

- the first time before using the meter for patient blood glucose testing.
- at intervals established by the facility.
- when a new test strip box is opened.
- if the test strip container was left open.
- if test strips were improperly stored.
- if there is a question about a blood glucose result.
- to check the performance of the system.
- if the meter was dropped.

Your facility may require that control tests be successfully performed **after** any of the following occur and **before** patient testing resumes:

- Previous control results were outside the acceptable range.
- Control tests were not performed at the proper interval.

Control results must be within the designated range on the test strip container label, or as defined by your facility, before being considered acceptable. Patients can be tested after control tests have been acceptably performed at the proper testing interval.

Performance Characteristics

Reproducibility (day-to-day imprecision): The mean imprecision is <1.7 %. In a typical series of tests, a coefficient of variation of 1.6 % was obtained.

Ingredients

Component	Control Solution Level 1 (Low)	Control Solution Level 2 (High)
Glucose	0.06 %	0.30 %
Buffer	4.84 %	4.84 %
Biological Salt	3.39 %	3.39 %
Preservative	0.30 %	0.30 %
Non-reactive ingredients	10.01 %	10.01 %
FD&C Blue #1	0.08 %	0.08 %
Percentages in weight/weight		

NOTE

For an explanation of symbols used refer to the end of the insert.

Visit our website at www.accu-chek.com to contact the local Roche representative for more information.

LAST UPDATE: 2014-05

Research Article

Comparison of Commercial Genetic-Testing Services in Korea with 23andMe Service

Table 2: Comparison of relative risks among 3 genetic tests.

Category	Relative risk for participant 1 (man)			Relative risk for participant 2 (woman)		
	Hellogene Platinum	DNAGPS Optimus	23andMe	Hellogene Platinum	DNAGPS Optimus	23andMe
Lung cancer	2.09	1.97	0.8*	2.09	0.92	0.8*
Esophageal squamous cell carcinoma	1.25	2.41	1.21	0.72	0.52	0.8
Colorectal caner	1.36	0.73	1.1	0.98	0.97	1.1
Bladder cancer	0.77	0.8	Typical	0.77	0.9	Typical/lower
Breast cancer	NA	NA	NA	0.84	0.77	0.98
Body mass index (obesity)	0.91		0.85*	0.91		0.83*
Atrial fibrillation	0.94	0.68	1.12*	0.94	1.39	1.93*
Coronary artery disease	1.06	0.51	0.78*	0.62	0.34	0.71*
Parkinson's disease	1.09	0.84	1.05	0.95	1.04	0.77
Narcolepsy	1.41	1.29	Lower*	1.41	1.29	Typical*
Warfarin maintenance dose	Normal dose	Moderate dose	Decreased dose	Normal dose	Increased dose	Slightly decreased dose

NA: not applicable.

*Relative risk in European. There was no Asian data.

이 유전자 검사들은 직접 소비자에게 허용하기에 적절한 것인가?

S Kim et al, BioMed Research International 2014, ID 539151

보건복지부 장관 고시 항목의 문제

- 아직 임상적 유용성이 확인되지 않은 항목을 정부가 제도적으로 허용하고 있다.
- 검사 항목의 안정성과 유효성을 정부가 책임지고 있다.
- 미국은 이들 검사를 일반인에 서비스하지 않고 있다.

혁신의 항목과 틀을 정부가 정할 수 있을 만큼 정부는 예측을 할 수 있고, 전문적인가?

문제의 재조명:

정확하게 예측할 수 없는 검사를 허용할 수 있을까?

정부의 보증 → 소비자의 책임으로 전환

핵심 전제 조건

소비자의 알 권리, 투명성

국내 유전자 검사 기관의 관련 제도

의료기관

식약처: 의료기기 승인

NECA: 신의료기술 유효성, 안전성 평가

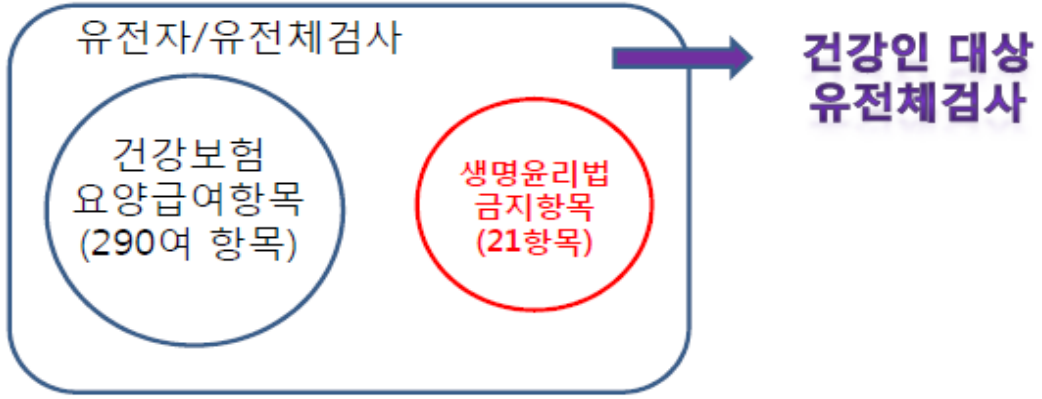
심평원: 수가등재

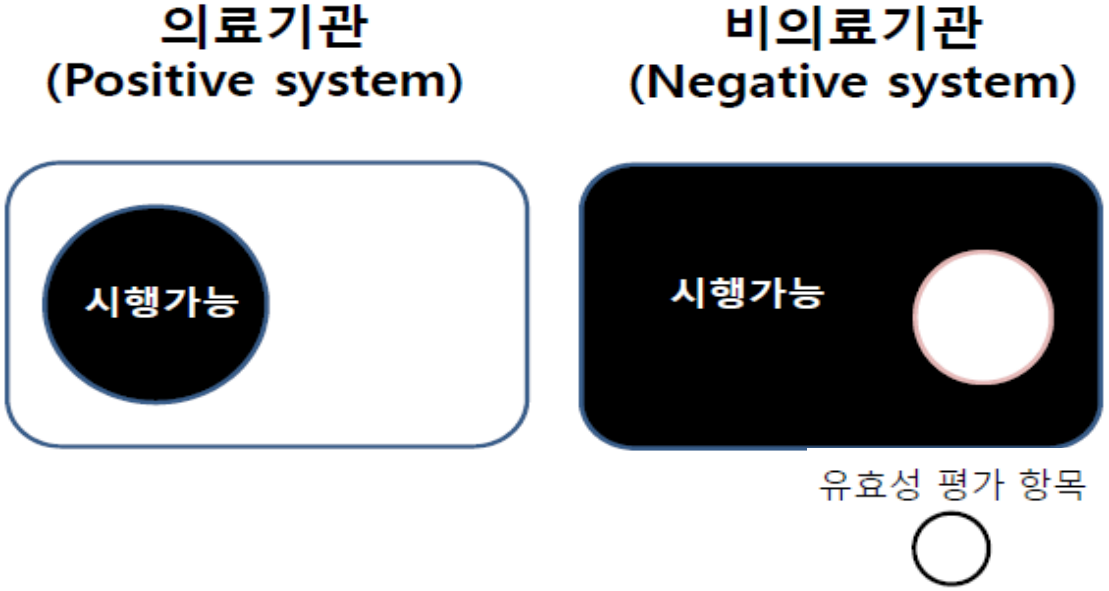
건강보험요양급여 항복 시행(290여 항목)

비의료기관

질병관리본부: 유전자검사 기관 및 항목 신고

생안법 규정금지/제한검사(21 항목) 제외한 모든 항목 의료기관의 의뢰로 시행 가능 복지부장관 고시 직접유전자검사항목 시행





2015년 국내 유전자 검사 실적

	Total	의료기관	비의료기관
진단	422,121	218,368(52%)	203,753(48%)
치료	233,254	178,137(76%)	55,117(24%)
예방	661,537	17,116(3%)	644,421(97%)
기타	1,683	1,035(61%)	648(39%)
계)	1,332,717	414,662(31%)	918,055(69%)

국내 의료기관과 비의료기관의 비중은 비슷하다.

유전자 검사기관 179개, 의료 90개, 비의료 89개

정밀의료 관련 사회, 제도적 환경: 미국 Vs 한국

- 미국
  - 느슨한 진입장벽: 규제항목이 아니면 모두 허용한다.
  - 강력한 사후 모니터링과 규제
  - 신뢰를 우선으로 한다.
  - 개별 사건-→ 개별 사건에 대한 대책 수립, 강력한 처벌, 규제의 일반화를 하지 않음.
- 한국
  - 강력한 진입장벽: 규정에 허용되지 않은 항목은 모두 못한다.
  - 느슨한 사후 모니터링과 규제: 일단 승인된 것은 큰 문제가 아니면, 감독이 없다.
  - 신뢰(?)
  - 개별 사건 → 개별 사건의 해결이 아닌 규제의 일반화

정밀의료와 디지털헬스케어 의료서비스의 정책 제언

- 네가티브 규제 방식
  - 규제가 필요한 항목 이외에는 모두 허용한다.
- 소비자에게 알권리, 참여를 촉진하도록 한다.
- 강력한 사후 감독
- 의료기관과 비의료기관의 관리 제도의 일원화