Gen-ETHICS

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LSUHSC

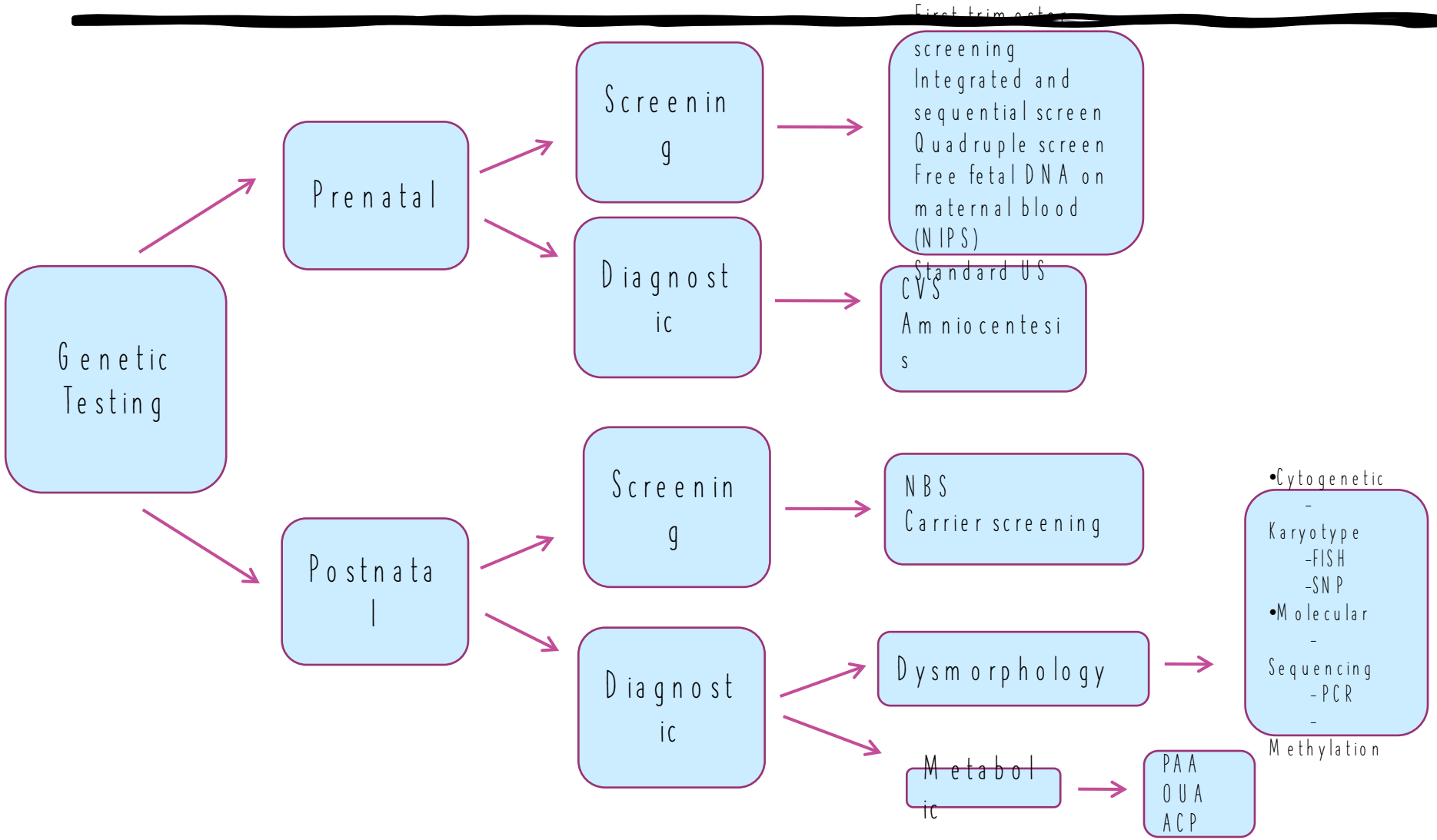
Disclosures

- Alexion Pharmaceutical Speaker Bureau (NF1 PN)
- Chiesi Pharmaceutical Advisory Board alpha-mannosidosis

Objectives

- Understand the complexity of the practice of Genetics and the impact of genetic information/testing in our life
- Recognize ethical, legal and social issues relevant to genetic counseling and testing
- Address ethical dilemmas related to testing and advances in medical genetics

Genetic testing



Genetic testing (Pros and cons)

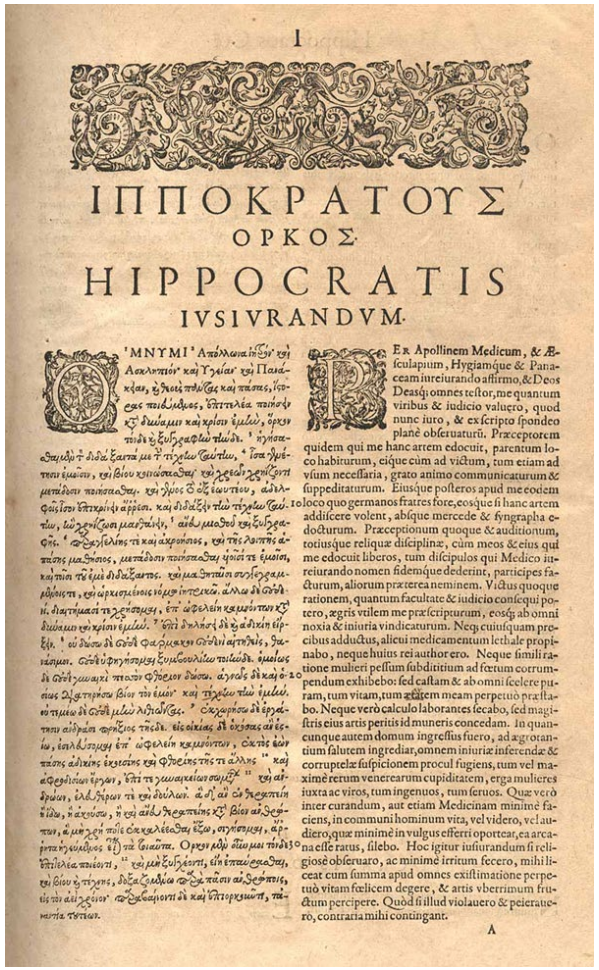
- One of the cornerstone of a comprehensive genetic evaluation is testing
- Genetic testing results not only impact our patient's care, but also their immediate and extended family
- Establish or confirm a clinical diagnosis
- Determine course of action or treatment
- Determine prognosis

- Technological limitations
- Identification of variants of unknown clinical significance (VUS)
- Emotional, social, or financial consequences of the test results
- Genetic discrimination for employment or insurance (GINA Law)

Informed consent (IC)

- The process of gathering autonomous authorization for a medical intervention or medical research participation
- IC is a fundamental component of medical practice; it is both an ethical and legal requirement
- What is needed for IC?
  - decision-making capacity
  - voluntariness
  - comprehension and
  - adequate information
- Does this apply to the Genomic era?

Evolution of IC



ΙΠΠΟΚΡΑΤΟΥΣ
ΟΡΚΟΣ

HIPPOCRATIS
IVSIVRANDVM.

ΟΜΝΥΜΙ Ἀπόλλωνα ἰητρὸν καὶ Ἀσκληπιὸν καὶ Ὑγείαν καὶ Πανάκεαν, καὶ θεοὺς πάντας τε καὶ πάσας, ἵστορας ποιεύμενος, ἐπιτελέα ποιήσειν κατὰ δύναμιν καὶ κρίσιν ἐμὴν, ὅρκον τόνδε καὶ ξυγγραφὴν τήνδε. Ἡγήσασθαι μὲν τὸν διδάξαντα με τὰ τέχνην ταύτην, ἴσα γενέτῃσιν ἐμοῖσιν, καὶ βίου κοινώσασθαι καὶ χρεῶν χρηίζοντι μετάδοσιν ποιήσασθαι, καὶ γένος τὸ ἐξ ἐωυτοῦ, ἀδελφοῖσιν ἐπικρινῆ ἄρρεσι, καὶ διδάξειν τὴν τέχνην ταύτην, ἢν χρηίζωσι μανθάνειν, ἄνευ μισθοῦ καὶ ξυγγραφῆς. Παραγγελίης τε καὶ ἀκροσιος, καὶ τῆς λοιπῆς ἁπάσης μαθήσιος, μετάδοσιν ποιήσασθαι υἱοῖσί τε ἐμοῖσι, καὶ τοῖσι τοῦ ἐμὲ διδάξαντος, καὶ μαθηταῖσι συγγεγραμμένοισι, καὶ ὡρκισμένοισι νόμῳ ἰητρικῷ, ἄλλου δὲ οὐδενός. διαιτήμασί τε χρήσομαι, ἐπ' ὠφελείῃ καμνόντων κατὰ δύναμιν καὶ κρίσιν ἐμὴν, κακῷ δὲ κληῖσαι καὶ ἀδικίην ὑπέρξειν. οὐ δώσω δὲ οὐδὲ φάρμακον οὐδενὶ αἰτηθεὶς, θανάσιμον. οὐδὲ ὑφηγήσομαι ξυμβουλίην τοιήνδε. ὁμοίως δὲ οὐδὲ γυναικὶ πεσσὸν φθόριον δώσω. ἁγνῶς δὲ καὶ ὁσίως διατηρήσω βίον τὸν ἐμὸν καὶ τέχνην τὴν ἐμήν. οὐ τεμέω δὲ οὐδὲ μὴν λιθιῶντας, ἐκχωρήσω δὲ ἐργάτῃσιν αἰδρείῃσι τοιῆδε. εἰς οἰκίας δὲ ὁκόσας ἂν ἐσίω, ἐσελεύσομαι ἐπ' ὠφελείῃ καμνόντων, ἐκτὸς ἐὼν πάσης ἀδικίης ἑκουσίης καὶ φθορίης τῆς τε ἄλλης καὶ ἀφροδισίων ἔργων, ἐπί τε γυναικείων σωμάτων καὶ ἀνδρώων, ἐλευθέρων τε καὶ δούλων. ἃ δ' ἂν ἐν θεραπείῃ ἢ ἰδω, ἢ ἀκούσω, ἢ καὶ ἄνευ θεραπείης κατὰ βίον ἀνθρώπων, ἃ μὴ χρὴ πού ἐκκαλέεσθαι ἔξω, σιγήσομαι, ἄρρητα ἡγεύμενος εἶναι τὰ τοιαῦτα. Ὅρκον μὲν οὖν μοι τόνδε ἐπιτελέα ποιοῦντι, καὶ μὴ ξυγχέοντι, εἴη ἐπαύρασθαι καὶ βίου καὶ τέχνης, δοξαζομένῳ τοῖσι πᾶσιν ἀνθρώποις εἰς τὸν ἀεὶ χρόνον· παραβαίνοντι δὲ καὶ ὑπορκοῦντι, τἀναντία τούτων.

ER Apollinem Medicum, & Æsculapium, Hygiamque & Panaceam iureiurando affirmo, & Deos Deasq; omnes testor, me quantum viribus & iudicio valuero, quod nunc iuro, & exscripto spondeo planè obseruaturũ. Præceptorem quidem qui me hanc artem edocuit, parentum loco habiturum, eique cùm ad victum, tum etiam ad vsum necessaria, grato animo communicaturum & suppeditaturum. Eiusque posteros apud me eodem loco quo germanos fratres fore, eosque si hanc artem addiscere volent, absque mercede & syngrapha edocturum. Præceptionum quoque & auditionum, totiusque reliquæ disciplinæ, cùm meos & eius qui me edocuit liberos, tum discipulos qui Medico iureiurando nomen fidemque dederint, participes facturum, aliorum præterea neminem. Victus quoque rationem, quantum facultate & iudicio consequi potero, ægris vtilem me præscripturum, eosq; ab omni noxia & iniuria vindicaturum. Necq; cuiusquam precibus adductus, alicui medicamentum lethale propinabo, neque huius rei author ero. Neque simili ratione mulieri pessum subdititium ad fœtum corrumpendum exhibebo: sed castam & ab omni scelere puram, tum vitam, tum artem meam perpetuò præstabo. Neque verò calculo laborantes secabo, sed magistris eius artis peritis id muneris concedam. In quascunque autem domum ingressus fuero, ad ægrorantium salutem ingrediar, omnem iniuriæ inferendæ & corruptelæ suspicionem procul fugiens, tum vel maximè rerum venerearum cupiditatem, erga mulieres iuxta ac viros, tum ingenuos, tum seruos. Quæ verò inter curandum, aut etiam Medicinam minimè faciens, in communi hominum vita, vel videro, vel audiero, quæ minimè in vulgus efferri oporteat, ea arcana esse ratus, silebo. Hoc igitur iusiurandum si religiosè obseruaro, ac minimè irritum fecero, mihi liceat cum summa apud omnes existimatione perpetuò vitam fœlicem degere, & artis vberrimum fructum percipere. Quòd si illud violauero & peieratero, contraria mihi contingant.

A

- You would think this concept should be as old as the practice of medicine
- But throughout the history of medicine, medical ethicists and physicians were more concerned with howbest to conceal potentially stress-inducing information from patients to reduce harm and anxiety
- Even the Hippocratic oath encouraged physicians to keep their patients in the dark as a means of protecting them from undue stress
- It was not until 1947 that the Nuremberg code,

Genetic literacy

› Public Health Genomics. 2017;20(6):343-348. doi: 10.1159/000489117. Epub 2018 May 31.

Assessing Genetic Literacy Awareness and Knowledge Gaps in the US Population: Results from the Health Information National Trends Survey

Melinda Krakow [1], Chelsea L Ratcliff [2], Bradford W Hesse [1], Alexandra J Greenberg-Worisek [3]

Affiliations + expand

PMID: 29852491 PMCID: PMC6095736 DOI: 10.1159/000489117

Free PMC article

Abstract

Background/aims: Public understanding of the role of genetics in disease risk is key to appropriate disease prevention and detection. This study assessed the current extent of awareness and use of genetic testing in the US population. Additionally, the study identified characteristics of subgroups more likely to be at risk for low genetic literacy.

Methods: The study used data from the National Cancer Institute's 2017 Health Information National Trends Survey, including measures of genetic testing awareness, genetic testing applications and genetic testing usage. Multivariable logistic regression models estimated associations between sociodemographics, genetic testing awareness, and genetic testing use.

Results: Fifty-seven percent of respondents were aware of genetic tests. Testing awareness differed by age, household income, and race/ethnicity. Most participants had heard of using tests to determine personal disease risk (82.58%) or inherited disease risk in children (81.41%), but less were familiar with determining treatment (38.29%) or drug efficacy (40.76%). Among those with genetic testing awareness, actual testing uptake was low.

Conclusions: A large portion of the general public lacks genetic testing awareness and may benefit from educational campaigns. As precision medicine expands, increasing public awareness about genetic testing applications for disease prevention and treatment will be important to support population health.

Keywords: Awareness; Genetic education; Genetic literacy; Genetic testing; Health communication; Health information; Knowledge; Knowledge gap; Population survey.

› J Community Genet. 2019 Jan;10(1):73-84. doi: 10.1007/s12687-018-0363-7. Epub 2018 Mar 28.

New literacy challenge for the twenty-first century: genetic knowledge is poor even among well educated

Robert Chapman [1], Maxim Likhanov [2], Fatos Selita [2 3], Ilya Zakharov [4], Emily Smith-Woolley [5], Yulia Kovas [6 7]

Affiliations + expand

PMID: 29589204 PMCID: PMC6325037 DOI: 10.1007/s12687-018-0363-7

Free PMC article

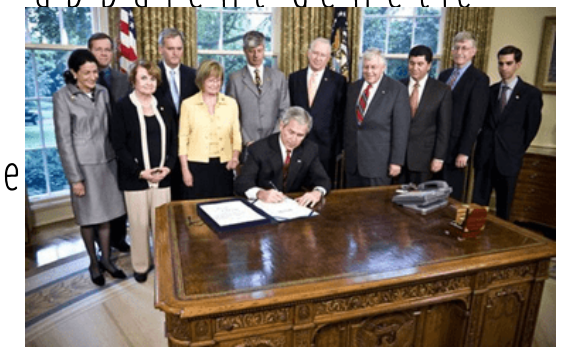
Abstract

We live in an age of rapidly advancing genetic research. This research is generating new knowledge that has implications for personal health and well-being. The present study assessed the level of genetic knowledge and personal engagement with genetics in a large sample (N = 5404) of participants. Participants received secondary education in 78 countries, with the largest samples from Russia, the UK and the USA. The results showed significant group differences in genetic knowledge between different countries, professions, education levels and religious affiliations. Overall, genetic knowledge was poor. The questions were designed to assess basic genetic literacy. However, only 1.2% of participants answered all 18 questions correctly, and the average score was 65.5%. Genetic knowledge was related to peoples' attitudes towards genetics. For example, those with greater genetic knowledge were on average more willing to use genetic knowledge for their personal health management. Based on the results, the paper proposes a number of immediate steps that societies can implement to empower the public to benefit from ever-advancing genetic knowledge.

Keywords: Demographic differences; Genetic knowledge; Genetic literacy; Genetic testing; Health.

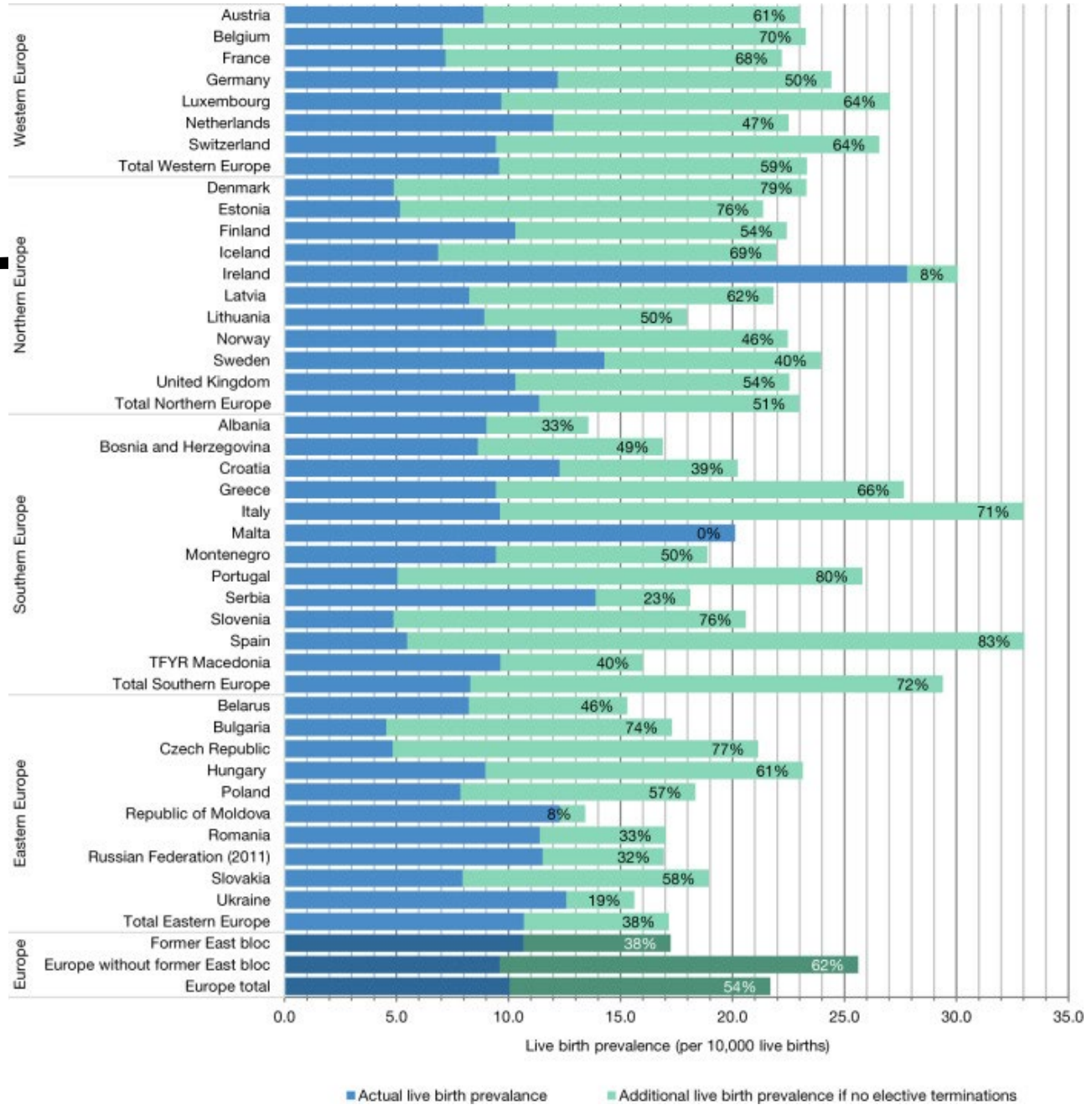
Genetic discrimination

- Discrimination based solely on the nature of an individual's genotype
- Historical discriminatory practices occurred commonly in the US based on misuse of genetic information (GI)
  - 1907 Indiana sterilization law
  - By 1981, many States adopted sterilization laws to "correct" apparent genetic traits or tendencies (fortunately now repealed)
  - In the 1970's GI was misused to target populations with increased genetic disorders (Black community (SS))
- Genetic Information Non-Discrimination act of 2008



Food for thought...

- Is prenatal GT a form of genetic discrimination?
- Let's use DS as an example, data form a study published in 2020 showed in 2011-2015, they estimate 8,031 annual live births of children with DS, which would have been around 17,331 births annually, absent selective



Food for thought...

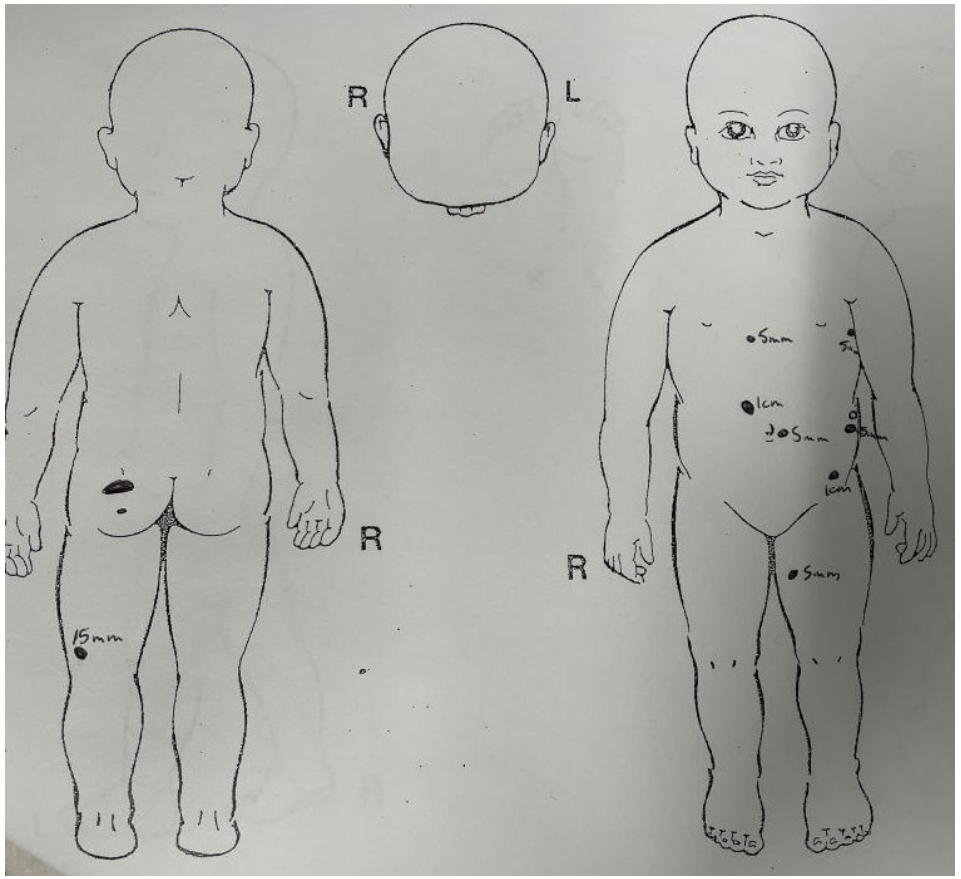
- What about Newborn screening programs (NBS)?
  - Because NBS can be the only way to tell if a newborn has certain conditions, it is mandated (required) by state lawin the United States (U.S.). (Newborn Screening Saves Lives Act of 2007, GWB)
  - In Louisiana Newborn Screening panel is required by law
  - Wyoming is the only US state that requires parental consent for NBS
  - Most states allowopting out due to a religious objection, but they must sign a written refusal statement
  - Most birthing hospitals do not inform mothers of NBS and often it is a shock when a PPNBS is reported...

Genetic testing challenges...

- Emotional Impact:
  - Anxiety and Uncertainty: Receiving genetic test results can be emotionally overwhelming. People may feel anxious, depressed, or guilty, especially if the results reveal a risk for a serious condition.
  - Unexpected Information: Sometimes, genetic testing uncovers unexpected information about health risks, family relationships, or ancestry. Dealing with this newfound knowledge can be stressful1.
- Social and Family Dynamics:
  - Family Tensions: Genetic testing results don't just affect the individual being tested; they can also reveal information about other family members. This can create tension within families.
  - Disclosure Challenges: Deciding whether to share genetic information with family members can be complex. Balancing privacy and the need for informed decisions is crucial.
- Financial Considerations:
- Ethical Dilemmas:
  - Privacy: Balancing the need for privacy with the importance of sharing genetic information for medical purposes is an ongoing challenge.
  - Informed Consent: Ensuring that individuals fully understand the implications of genetic testing and provide informed consent is critical.
- Clinical Limitations:
  - Limited Treatment Options: Even when a genetic disorder is diagnosed, treatment options may be scarce or nonexistent. This can be frustrating for patients and healthcare providers.
  - Predictive Value: Genetic tests can't always predict the severity of symptoms or disease progression. They provide probabilities rather

Let's talk business...

- Case #1
  - 11 mo old male seen with father for an evaluation due to CALs, at the time of initial evaluation, he did not meet clinical criteria (only had >6 CAL that measured more than 5mm
  - We continued to monitor him clinically
  - At the age of 18 months still did not meet criteria, we discussed molecular testing, a sample was sent that identified a VUS in NF1, to help resolve this VUS we requested parental testing, mother and



Case #1

ORDER SUBMITTED	SPECIMEN RECEIVED	LAB PROCESSING	ANALYSIS AND INTERPRETATION	REPORT RELEAS
12/23/2022	1/10/2023	1/10/2023	1/17/2023	1/24/2023

PATIENT [redacted]

+ Start new order for this patient

CLINICAL TEAM Children's Hospital of New Orleans Genetics

Ordered by: Rainbow Laurant

Ordering physician: Regina Zambrano, NPI: 1659574739

Shared with 4 colleagues

TEST SELECTION Family follow-up test

BILLING No additional charge

REPORTS AND DOCUMENTS

Clinical report (01/24/2023)

Requisition form

Upload document

MESSAGES

SEND MESSAGE TO INVITAE

father of [redacted]; results for father [redacted] were not consistent w/parent-chil d relationship

Case #2

- Called to do a consultation at WJMC for a 1 day old child with prenatal diagnosis of Trisomy 13
- Born at 37 WGA to a G3P1A1 mother after a pregnancy complicated by abnormal prenatal ultrasound that showed:
  - 2 vessel cord
  - Intracardiac echogenic foci
  - Microcephaly
  - SGA
- Mother underwent NIPS, a sample was sent to Sequenom for MaterniT21 Plus test
  - Abnormal result, positive for trisomy 13, no Y material (female fetus)

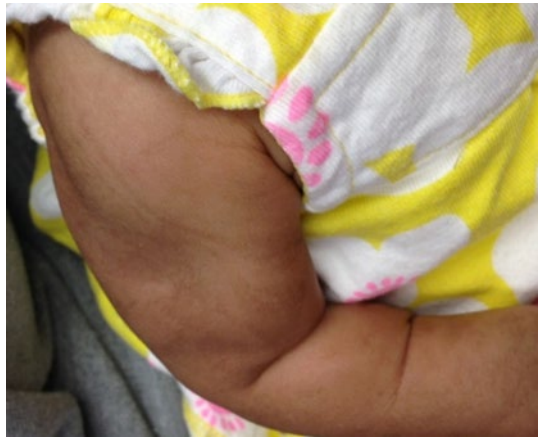
Case #2

- Mother was counseled by her MFM and they discussed the severity and short life span in children with trisomy 13, she was offered delivery at 38 weeks gestation

- Mother indicated to her OB/GYN she wished for comfort care for her child...

Case #2

- Upon arrival to the unit, I found confused nurses, confused mother …
- Positive findings on PE included:
  - Microcephaly
  - Slopping forehead and mild bi-temporal narrowing
  - Low set, dysplastic ears
  - Mild DS PF
  - CV, murmur and echocardiographic evidence of ASD, PDA, VSD and PFO
  - Skin examination with areas of streaky hyperpigmentation
- I suspected mosaic trisomy 13 and sent extended post-natal karyotype that showed:

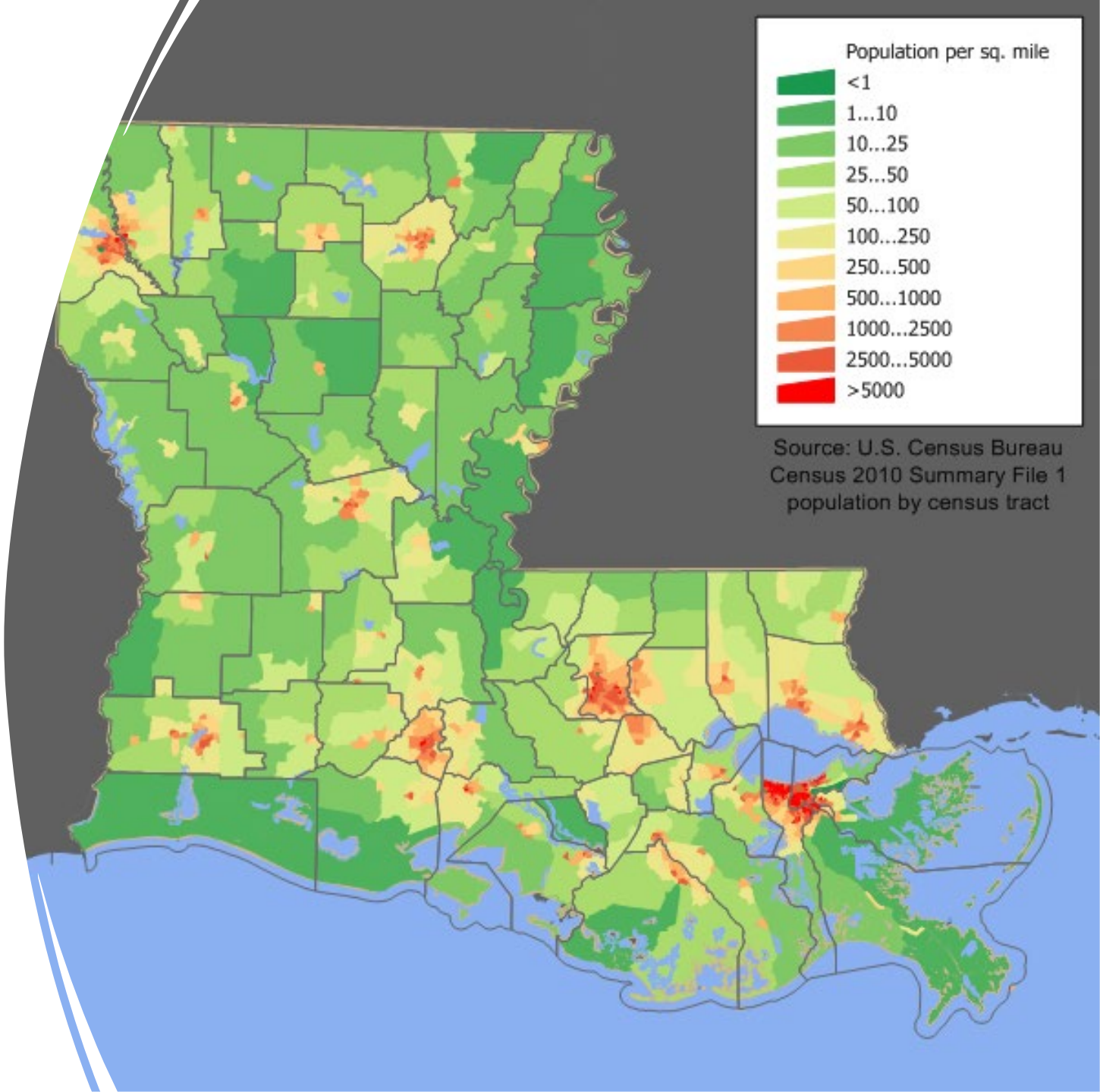


Ethical considerations

- Full Trisomy 13 is a devastating diagnosis, with short life span, in this case PNS and PNT supported this diagnosis
- Upon arrival to this world, she was expected to be severely ill, hence the confusion in the unit, and upon examination it was determined she did not have full trisomy 13, but she was a mosaic
- Children with mosaicism could have a less severe presentation, survival rates could be higher
- Appropriate counseling considering this possibility (mosaicism) could have helped in this case

Limitations of access to genetic care

- According the official last US Census (2010) 4,533,372 people lived in Louisiana at that time
- In 2024 there are 6 practicing clinical Geneticists in the state
- There is approximately 1.32 geneticists per 1,000,000 in the state



Genetic testing of minors for adult onset disorders

- A consensus exists between professional medical societies including the ASHG, ACMG, ACOG, AMA, NSGC regarding GT of minors for adult onset conditions:
  - *… encourages deferring predictive genetic testing of minors for adult-onset conditions when results will not impact childhood medical management or significantly benefit the child. Predictive testing should optimally be deferred until the individual has the capacity to weigh the associated risks, benefits, and limitations of this information, taking his/her circumstances, preferences, and beliefs into account to presserve his/her autonomy and right to an open future*
- Reasons supporting this statement include:
  - Possibility of discrimination
  - Stigma (alteration of self image and the expectations of those close to them)
  - Compromise their rights of autonomy and privacy
  - No ability to control who has access to that information

Case #3

- 5yo and 3yo females with an extensive family history of Lynch syndrome (*MLH1*):
  - father positive genetic testing at age 21, diagnosed with stage 3 CRC at age 35, *MLH1*
  - paternal uncle (age 38, had positive genetic testing but has not been diagnosed with cancer),
  - paternal grandfather (diagnosed with CRC age 35),
  - paternal paternal great-grandmother (diagnosed with CRC age 45, per report may have had diagnosis of breast cancer prior to CRC diagnosis),
  - paternal paternal great uncle (diagnosed at age 50) and great aunt (diagnosed with breast and colorectal cancer), and
  - paternal paternal maternal great-great grandmother (diagnosed with CRC at age 50)

Ethical considerations

- Extensive genetic counseling (CGC) was not enough to deter parents from GT
- They agreed on not disclosing the information to their children up until they felt they would have the maturity to deal with the implications of a positive test
- But this result is nowpart of her medical record, this could be shared by a healthcare professional not aware of their plan for later disclosure, risking psychological damage
- In my opinion testing should have not been done, as it does not change their immediate care

What about incidental / secondary findings?

- Whole exome sequencing has become a first-line approach for children with suspected genetic disease and/or developmental delay
- In 2013 the ACMG published their first guidelines on reporting secondary findings (56 genes, Cancer predisposition, CTD, malignant hyperthermia, cardiac disease): No option of opting out
- In 2015 the college revised its guidelines to permit patients to opt out after counseling
- Still, this is a contradiction to the recommendation on deferring testing in minors… .

Case #4

- 6yo male referred for an evaluation due to a history of developmental delays (mostly speech), macrocephaly, CALs, megaureter, ADD, he initially did not meet clinical criteria as only 1 sign was identified, and because of the other features (not

Result: Positive

- Causative variant(s) in disease genes associated with reported phenotype: **POSITIVE**
- Variant(s) in disease genes possibly associated with reported phenotype: **None Identified**
- ACMG Secondary Findings: **POSITIVE**
- mtDNA Test Results: **Mitochondrial DNA sequencing and deletion results are reported separately.**

Causative Variant(s) in Disease Genes Associated with Reported Phenotype:

Gene	Disease	Mode of Inheritance	Variant	Zygosity	Inherited From	Classification
NF1	NF1-related neurofibromatosis	Autosomal Dominant	c.4380_4383dup p.(A1462Yfs*4)	Heterozygous	Unknown	Pathogenic Variant

ACMG Secondary Findings:

Gene	Disease	Mode of Inheritance	Variant	Zygosity	Inherited From	Classification
MUTYH	MUTYH-related polyposis	Autosomal Recessive	c.1187 G>A p.(G396D)	Homozygous	Maternal + Unknown	Pathogenic Variant

DIY Genetics (Direct to consumer GT)

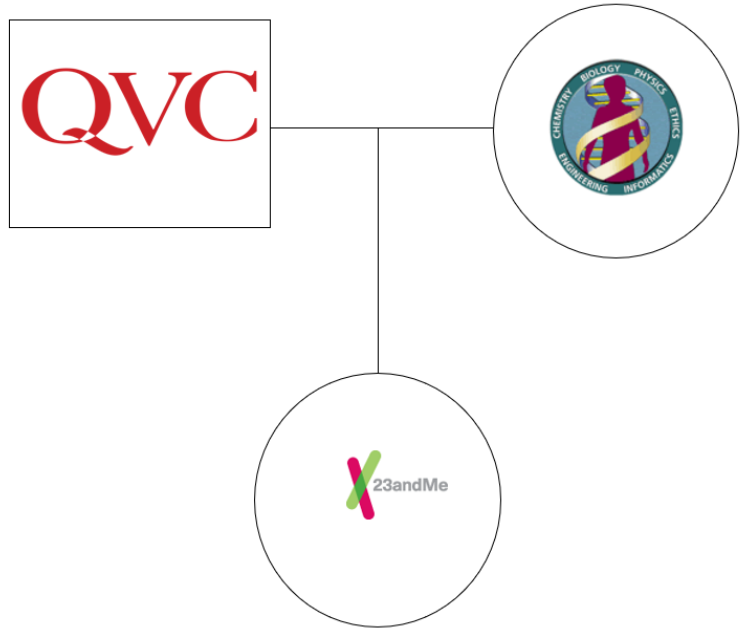
- Genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet
- These tests provide access to a person's genetic information without necessarily involving a doctor or insurance company in the process
- Consumers are notified of their results by mail or over the telephone, or the results are posted online or accessed via an app
- The global direct-to-consumer genetic testing market size was estimated at USD 1.93 billion in 2023 and is

DIY Genetics

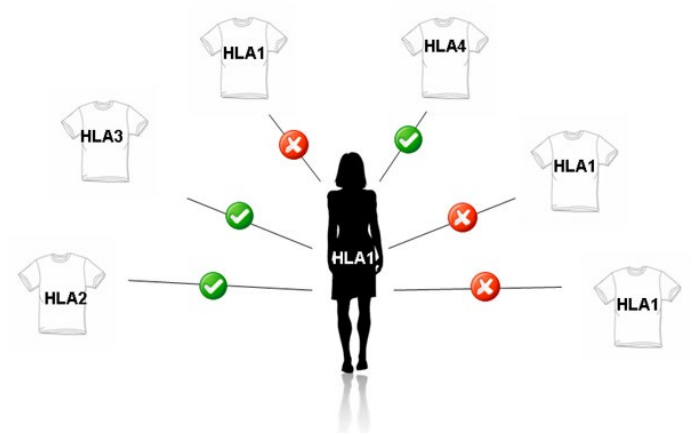
Key Direct-to-Consumer Genetic Testing Companies:

- 23andMe
- Family Tree DNA
- Ancestry
- Genesis HealthCare
- EasyDNA,
- Veritas
- Myriad Genetics Inc.
- Full Genomes Corporation, Inc
- Living DNA Ltd.
- Color Health, Inc.

- Ancestry
- Carrier testing
- Predictive testing
- Data mining
- Pharmacogenomics
- Nutrigenomics



Find your partner for life?



GenePartner

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Love is no coincidence!

Matching people by analyzing their DNA

TIME CNN abc NEWS GMA Galileo

→ Order a GenePartner Test

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- Press coverage
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Your perfect match is just one click away

Order your test now for only $249

Cons of DTC GT

- Quality of the tests offered (some labs are not CLIA approved)
- Accuracy of the information provided to the consumer
- Risk of misleading claims putting the consumer at risk of making harmful healthcare decisions on the basis of tests results
- PCP in the spotlight having to

Forensic Genetic Geneology

EMERGED IN 2018 AS A NOVEL INVESTIGATIVE TOOL THAT HAS BEEN APPLIED TO HUNDREDS OF UNRESOLVED COLD CASES IN THE UNITED STATES TO GENERATE LEADS AND IDENTIFY

WITH THE RISE IN POPULARITY OF CONSUMER DNA TESTING THERE NOW EXIST PUBLIC GENETIC GENEALOGY DATABASES POPULATED WITH LARGE AMOUNTS OF GENETIC DATA FROM

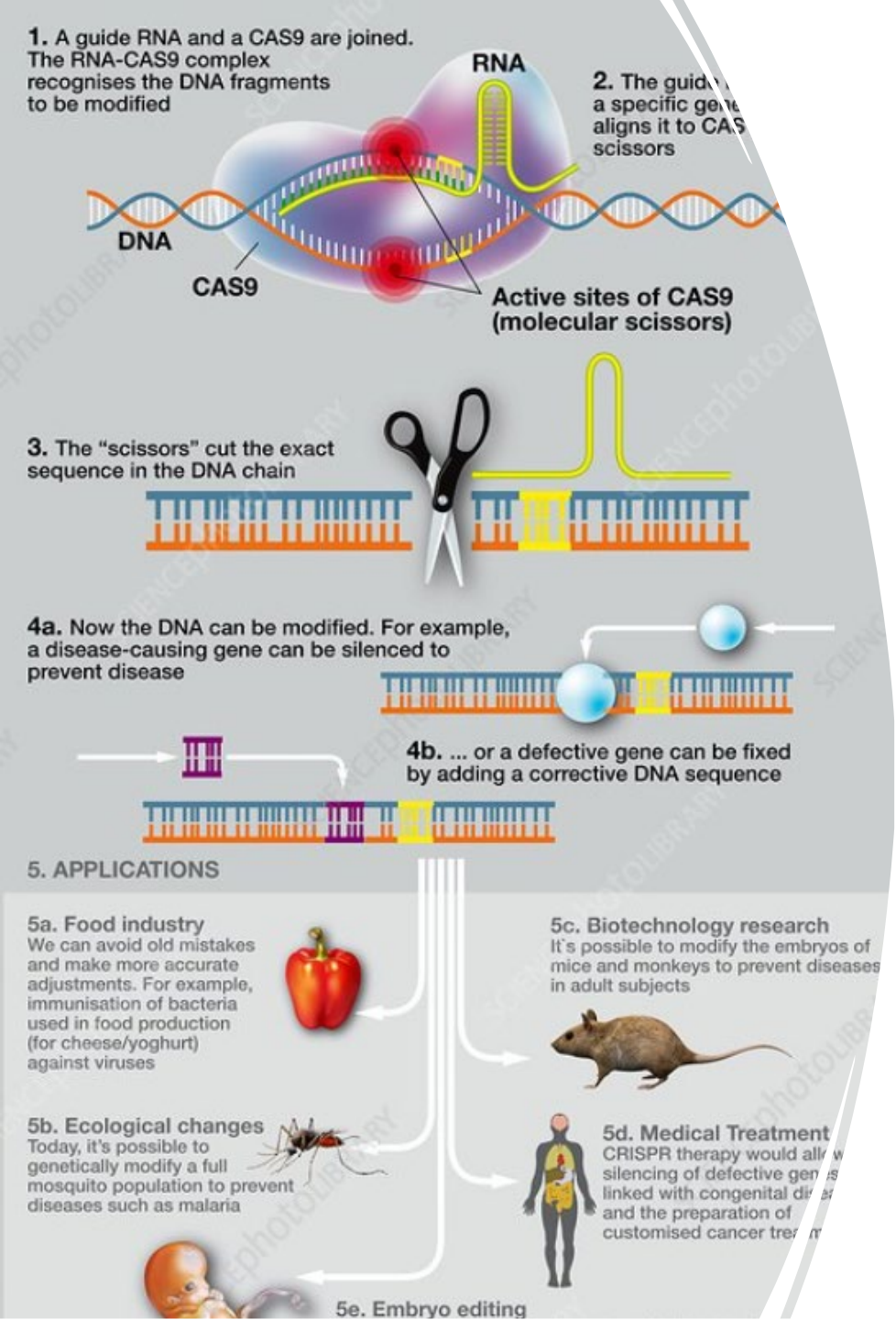
THE USE OF FGG TO RESOLVE THE IDENTITY OF THE GOLDEN STATE KILLER IN 2018 IS WIDELY CREDITED AS THE CASE THAT BROUGHT FGG TO THE FOREFRONT

FGG INVOLVES SEARCHING GENETIC GENEALOGY DATABASES FOR INDIVIDUALS THAT SHARE SEGMENTS OF MATCHING, OR VERY SIMILAR, DNA WITH AN UNKNOWN DNA SAMPLE AND ARE THEREFORE

THE NUMBER OF USERS IN THE DTC DATABASES (AS OF 16 JULY 2022) IS ESTIMATED AT; ~21 MILLION IN ANCESTRYDNA, ~12.8 MILLION IN 23ANDME, ~6 MILLION IN MYHERITAGEDNA,

Ethical considerations

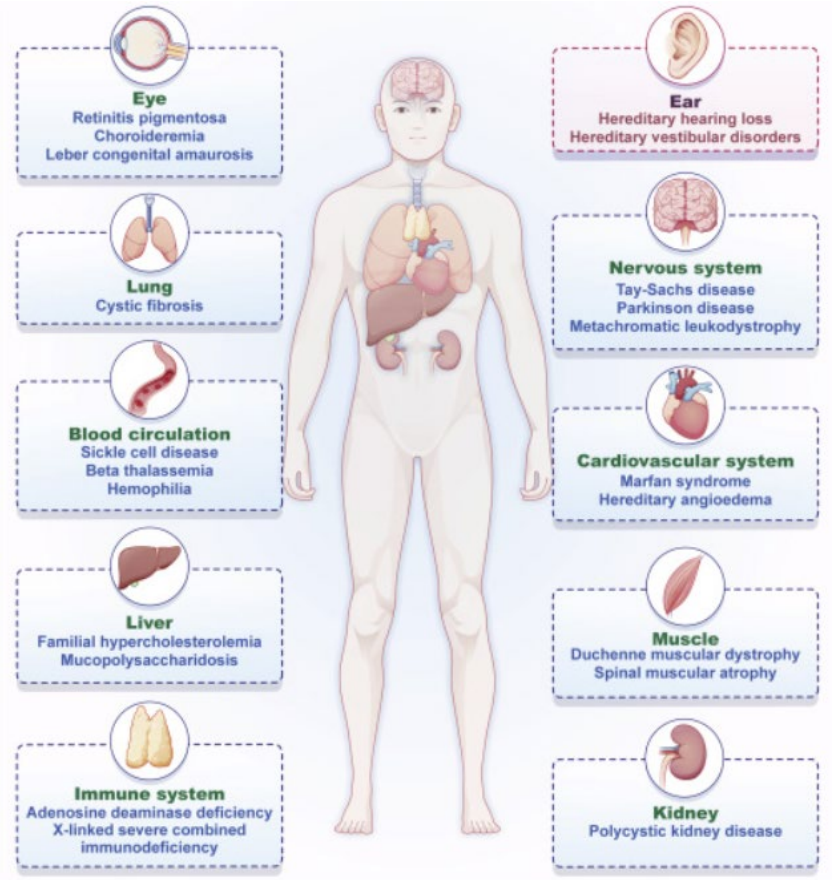
- Privacy issues: should a criminal be afforded privacy?
- Protection of the populations from violent offenders
- Identification of remains
- Resolution of cold cases



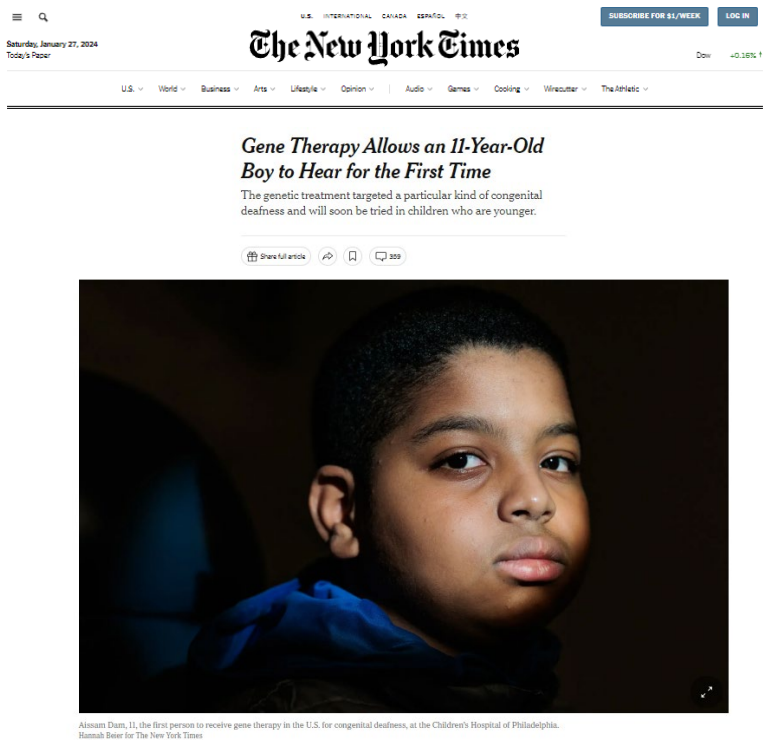
The not-so-distant future

- Genome editing (also called gene editing) is a group of technologies that give scientists the ability to change an organism's DNA
- Using the enzyme CRISPR/Cas-9 which was adapted from a naturally occurring genome editing system that bacteria use as an immune defense
- It is being explored in research and clinical

Gene therapy



- Monogenic disorders are current targets for gene therapy
- More than 300 clinical trials are underway using gene therapy to target monogenic disease
- A recent article in the media highlighted success with gene therapy in an 11-year old boy



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Gene Therapy Allows an 11-Year-Old Boy to Hear for the First Time

The genetic treatment targeted a particular kind of congenital deafness and will soon be tried in children who are younger.

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Aissam Dam, 11, the first person to receive gene therapy in the U.S. for congenital deafness, at the Children's Hospital of Philadelphia.
Hannah Beier for The New York Times

Ethical considerations

- Gene editing techniques have the potential of alter a human genome
- Most of the changes introduced with genome editing are limited to somatic cells
- Germline cell and embryo genome editing bring up a number of ethical challenges, including whether it would be permissible to use this technology to enhance normal human traits (such as height or intelligence).
- Based on concerns about ethics and safety, germline cell and

He Jiankui Controversy

- In December 2018, Chinese researcher He Jiankui shocked the world by announcing the birth of twin girls whose embryos he had genetically edited to be resistant to HIV
- While He Jiankui egregiously violated university regulations and ethical standards, his actions ignited a global dialogue about the permissibility of human embryonic gene editing
- Apprehended and sentence to 3 years in jail
- As of July 24, 2024 He has relocated to Wuhan as the Director of The Institute of Genetic Medicine at Wuchang University of Technology to work on gene editing to treat DMD, then got fired and now is at a private independent lab in Hainan (medical tourism city in China)

The End!

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