

# What is Personal Genomics?

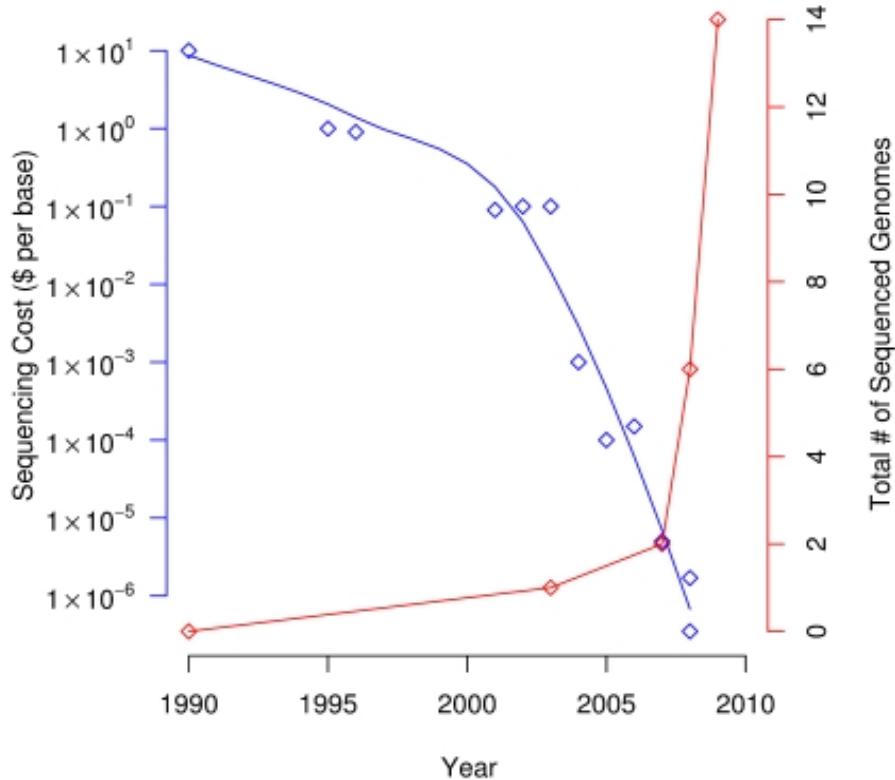


- Sequencing and analysis of the genome of individuals
- Partial or full genome sequencing
- Many diseases are genetically inherited (e.g. sickle cell anaemia, thalassemia and cystic fibrosis)
- Most common diseases are caused by multiple genes, multiple pathways and multiple environmental factors.
- Used to identify
  - genetic predisposition of an individual for common diseases
  - carrier status for inherited diseases
  - familial traits
  - efficacy and adverse reactions to common drugs.

# Personal Genomics



Sequencing Cost & Number of Sequenced genomes



Cost of DNA sequencing and cumulative number of genomes sequenced as a function of time. The blue points and the fitted line show the per-base sequencing cost, and the red points show the total number of sequenced genomes.

## Personal genome sequencing: current approaches and challenges

Michael Snyder, Jiang Du, and Mark Gerstein *Genes Dev.* 2010 Mar 1; 24(5): 423–431. doi: [10.1101/gad.1864110](https://doi.org/10.1101/gad.1864110)

Main steps in determining a genome sequence:

- identifying single-nucleotide polymorphisms [SNPs]
- structural variations [SVs]
- assembling new sequences
- phasing haplotypes)
- Challenges and performance metrics for evaluating the accuracy of the reconstruction.
- Individual and societal benefits of personal genome sequences

# Personal Genome Project (PGP)



- long term
- large cohort study
- aims to sequence and publicize the complete **genomes** and medical records of 100,000 volunteers, in order to enable research into **personal genomics** and personalized medicine.



## Sharing Personal Genomes

The Personal Genome Project was founded in 2005 and is dedicated to creating public genome, health, and trait data. Sharing data is critical to scientific progress, but has been hampered by traditional research practices—our approach is to invite willing participants to publicly share their personal data for the greater good.

[Personal Genome Project - Wikipedia](#)

[https://en.wikipedia.org/wiki/Personal\\_Genome\\_Project](https://en.wikipedia.org/wiki/Personal_Genome_Project)

<http://www.personalgenomes.org/>

# Personal Genome Project Data



Personal Genome Project: Harvard

About Team Volunteer

Data

News

Participant log in

Data & Samples

<http://www.personalgenomes.org/harvard/data>

## Genome data

In addition to whole genome sequencing, the Harvard PGP has a variety of donated genetic data (ranging from externally-performed genomes and exomes to direct-to-consumer genotyping).

[Get Genetic Data](#)

## Genome Reports

To provide some insight into whole genome sequences, the Harvard PGP has developed GET-Evidence to facilitate interpretation of genetic variants. Genome reports generated by GET-Evidence are publicly available for PGP genomes.

[View Genome Reports](#)

## Trait and survey data

The Harvard PGP data is uniquely valuable because it combines trait and health information with genetic data in a publicly available resource. Our trait surveys query participants regarding 239 self-reported traits.

[Trait & Survey Data](#)

## Participant profiles

Participant profiles present all public data associated with a participant, presenting genetic and trait data in the same place. Profiles also include medical data imported from electronic health records and diverse other types of data uploaded by participants.

[Browse Participant Profiles](#)

## Microbiome data

Microbiome profiles investigate the types of bacteria in and on a participant's body. Understanding the microbiome helps build a more complete profile of a participant: it is a form of biological profiling that captures personal environment and may be an important aspect in human health.

[Microbiome Data](#)

# What is Direct to Consumer Genetic Testing? (Personal Genomics)



 MyHeritage DNA

 GPS<sup>®</sup> ORIGINS<sup>™</sup>

 Living DNA  
YOUR ANCESTRY

 FamilyTreeDNA

 23andMe



Your Guide to Understanding  
Genetic Conditions

Healthcare providers order the appropriate test from a laboratory, collect and send the samples, and interpret the test results.

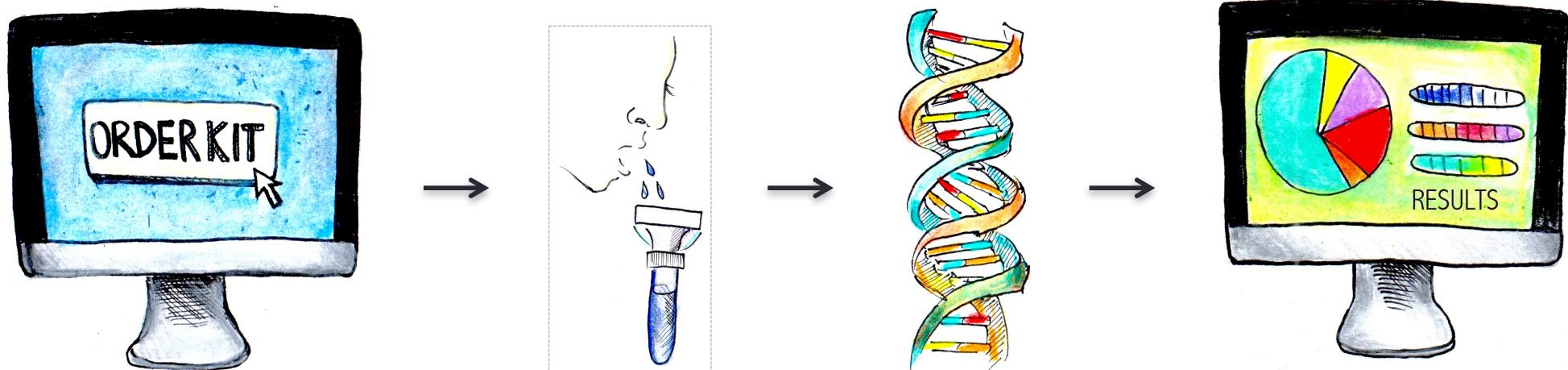
Direct-to-consumer genetic testing refers to genetic tests that are marketed directly to consumers via television, print advertisements, or the Internet. Apr 4, 2017

**Please follow the link for more information.**

<https://ghr.nlm.nih.gov/primer/testing/directtococonsumer>

<http://www.top10bestdnatesting.com/>

# How does direct-to-consumer genetic testing work?



1. Order kit online.

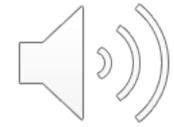
2. Spit into tube  
and send to lab.

3. The lab  
analyzes DNA.

4. Customer  
receives results.

Note: Some tests require a doctor's approval. In this case, results are typically sent to both the customer and the doctor, so they may discuss. 6

# Canvas Discussion topics:



- 1.What are the potential benefits to knowing more about your likelihood of developing a genetic disease?
- 2.What are the possible downsides to knowing?
- 3.Aside from health and medical information, what else might you be curious to learn about from your DNA?

# Why might personal genetics matter to you?

- Genome analysis available for under \$1,000 dollars
- Broadly available in the next 5-10 years
- Transforming medical care for the next generation



Image: James Hart, CC BY-NC-ND 2.0



<http://www.genome.gov/sequencingcosts/>



Veritas \$999 myGenome fully sequenced genome available on your smart phone

<http://www.nanalyze.com/2016/03/does-full-genome-sequencing-really-cost-1000-now/>

# Should you have access to your own data?



## POLICYFORUM

### INFORMATION ACCESS

#### Raw Personal Data: Providing Access

Jeanine E. Lunshof,<sup>1,2\*</sup> George M. Church,<sup>1</sup> Barbara Prainsack<sup>3</sup>

Hated debates on responsibilities in biomedical research currently focus on the end of the data and information pipeline: They revolve around issues of returning results to participants and patients (*1–3, 4*). Although these debates are timely, they miss a crucial point at the beginning of the pipeline: the question of whether sample donors are able to access the raw data derived directly from their stored sample. The U.S. Presidential Commission recently reviewed 32 reports from the United States and worldwide on returning of findings in diverse contexts (*4*); it is striking



Donors should have access to raw data derived from their contributions to research or clinical repositories to increase personal choice and reciprocity.

for data sharing, a requirement that may be very challenging in a competitive setting with diverse research and commercial interests. Databases that provide public open access, as, e.g., the Personal Genome Project (*5*), avoid such problems, and for them, issues of donor access are moot. But also studies for which public access is not an option should pay urgent attention to the structures and conditions necessary to provide donors access to their raw data.

In actual practice, the financial and logistic challenges related to providing access might result in significant additional costs, costs that



### LETTERS

edited by Jennifer Sills

#### Raw Data: Access to Inaccuracy

IN "RAW PERSONAL DATA: PROVIDING ACCESS" (POLICY FORUM, 24 January, p. 373), J. E. Lunshof and colleagues argue that donors should have access to raw data derived from their contribution to research or clinical repositories. Fairness, reciprocity, and respect for autonomy are compelling ethical reasons for access, if not for one major problem: the intrinsic inaccuracy of most research data.

Even the best-documented population studies cannot guarantee accurate data for individual participants. Limited research budgets force researchers to decide between assessing a few variables at high quality or many variables at lower quality, and they typically choose the latter. More data means more research opportunities, and suboptimal data quality is perfect enough when conclusions are drawn for populations at large. Yet, the data cannot be used to inform about individual participants.



To illustrate the moral obligation for granting access, the authors draw an excellent analogy with money banks, but the example actually undercuts their point. Money banks would never provide customers access to their bank accounts if they had even the slightest doubt about the accuracy of the balances. Inaccurate account data not only harm individual customers, who then remain uncertain about their financial position, but also destroy public trust in money banks. This is a risk that banks would not even think of taking, and scientists should not either.

High-quality online genome data interpretation tools, health professionals, and other independent experts cannot make sense of data when they cannot rely on the quality. A disclaimer concerning data accuracy, as the authors propose, does not solve that problem. If researchers respect their participants, take them seriously, and want to do more good than harm (*1*), they do not give them all they have, but give something valuable in a responsible way (*2*). And that is not merely access to data.

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#### References

1. The National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research. Ethical Principles and Guidelines for the Protection of Human Subjects of Research (1979); [www.hhs.gov/ohrp/humansubjects/guidance/belmont.html](http://www.hhs.gov/ohrp/humansubjects/guidance/belmont.html).
2. J. P. Evans, B. B. Rothschild, *Genet. Med.* **14**, 358 (2012).

#### Raw Data: Research and Health Care Goals Differ

IN THEIR POLICY FORUM "RAW PERSONAL data: Providing access" (24 January, p. 373), J. E. Lunshof *et al.* suggest that more should be done to enable persons who had their genome sequenced, either as research subjects or as patients, to actively access their raw data. They argue that routinely providing them with personal access codes to those data would be a matter of transparency and respect for autonomy. We think Lunshof *et al.* underestimate the dangers of actively handing out data that we know are not fully reliable and can lead to misinterpretation.

In our view, the proposed policy would be at odds with the responsibility of health professionals.

In health care, clinical utility should have priority over social utility. An appeal to reciprocity between donors and users of genomic data does not change this argument. Patients are not the same as data donors. If patients become data donors by consenting to have their data stored in research registries, they should be aware that they have entered a different relationship, in which they primarily contribute to the benefit of future patients.

Lunshof *et al.* ignore the crucial difference between health care and research when they criticize the recent recommendations of the European Society of Human Genetics

(ESHG). To avoid the unnecessary generation of incidental findings, these recommendations advise the use of targeted forms of testing if that is sufficient to address the patient's problem (*1*). According to Lunshof *et al.*, this is problematic as it "systematically precludes the possible discovery of complex genetic causation." We disagree: Discovery is the aim of research, not of health care. Conflicting these aims risks turning patients into research subjects without their consent.

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[www.sciencemag.org](http://www.sciencemag.org) SCIENCE VOL 343 24 JANUARY 2014

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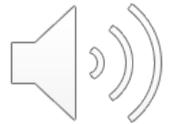
1: Lunshof JE, Church GM, Prainsack B. Information access. Raw personal data: providing access. *Science*. 2014 Jan 24;343(6169):373-4. doi: 10.1126/science.1249382. PubMed PMID: 24458627.

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28 FEBRUARY 2014 VOL 343 SCIENCE www.sciencemag.org  
Published by AAAS

<http://science.sciencemag.org/content/343/6169/373.full>  
<http://science.sciencemag.org/content/343/6174/968.1>

# The Beery twins' story highlights the promise of personalized medicine

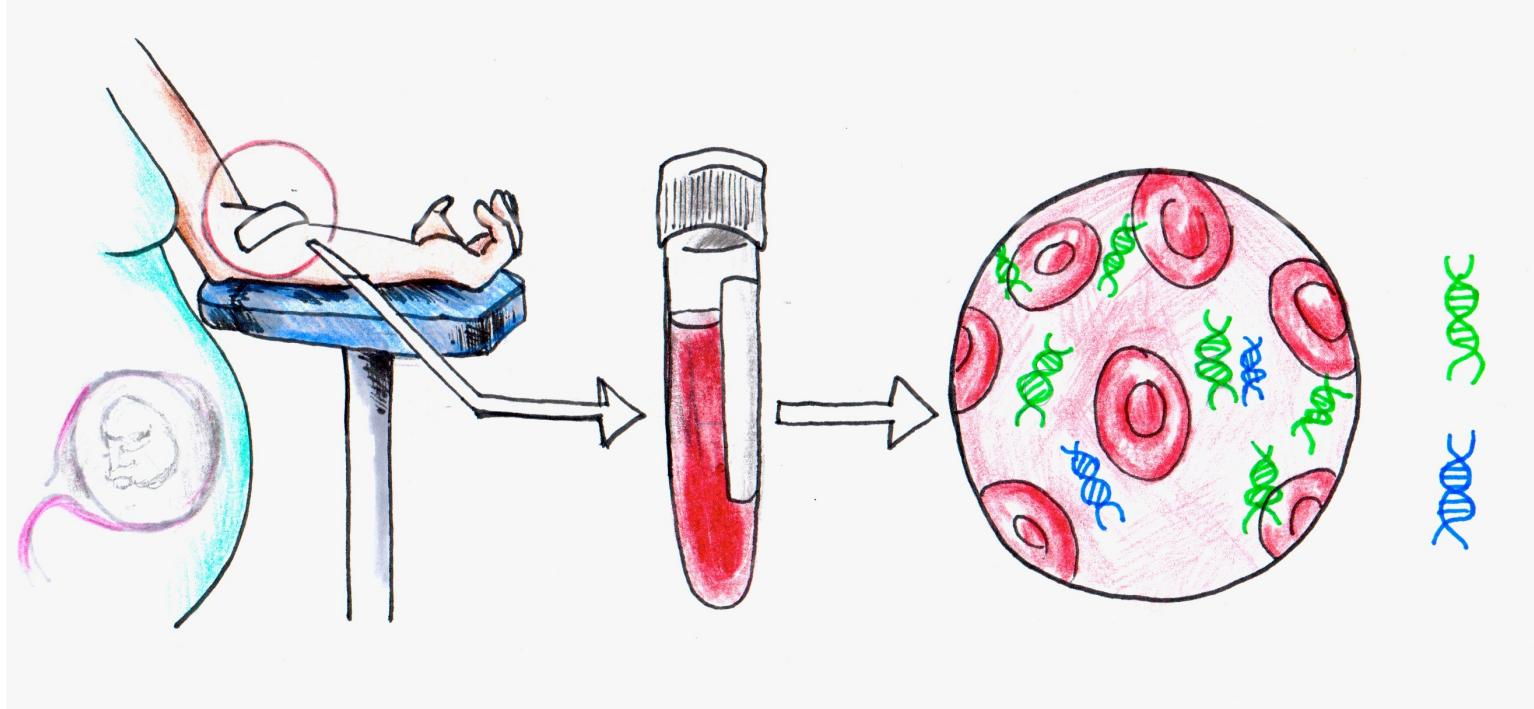


# Personal choices based on genetic information



- Angelina Jolie reveals she chose to undergo a double mastectomy.
- Jolie had a genetic test and found she carried a mutation in the BRCA1 gene. Doctors estimated there was a very high chance she would get breast cancer.

# Non-Invasive Prenatal Testing (NIPT)



- Fetus and mother share a blood supply.
- Fetal cells release DNA that enters the maternal bloodstream.
- Maternal blood now contains a mixture of maternal cell-free DNA and fetal cell-free DNA.

# DNA tests locate genetic branches on African-Americans' family trees

March 29, 2006 12:00 AM



By Ervin Dyer Pittsburgh Post-Gazette

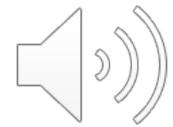
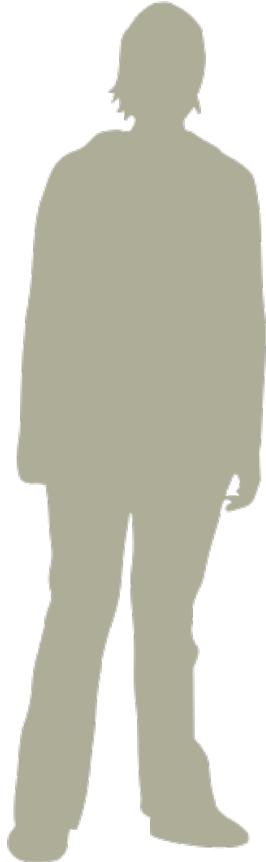
A few decades ago, the closest Gary Payne came to filling out his family tree were the little seeds of oral histories sown by his mom, Flabia Payne.



Steve Mellon, Post-Gazette

**A DNA test Gary Payne of Penn Hills took showed he has roots from China.**

# What could you learn from genetic testing?



## - Risk for conditions including:

- Alzheimer's Disease
- Heart Disease
- Cancer

## - Carrier Status

- Which medications could work best for you and/or which could harm you

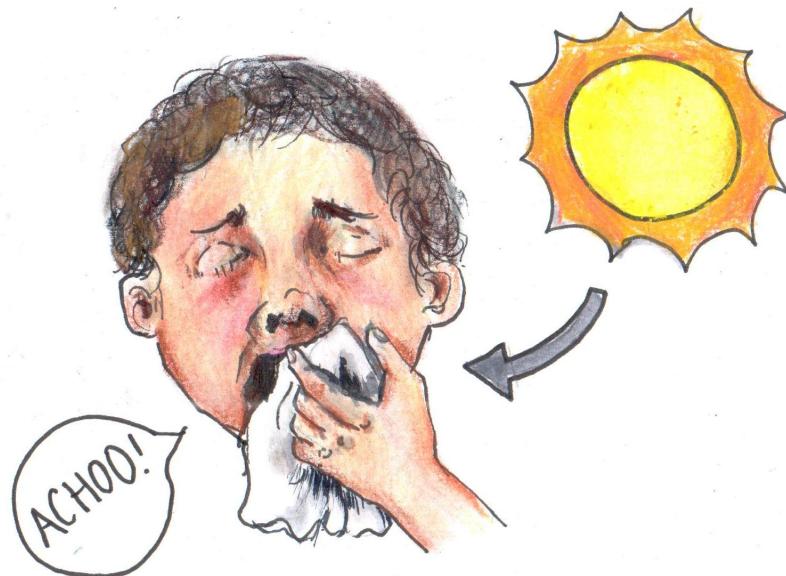
## - Ancestry



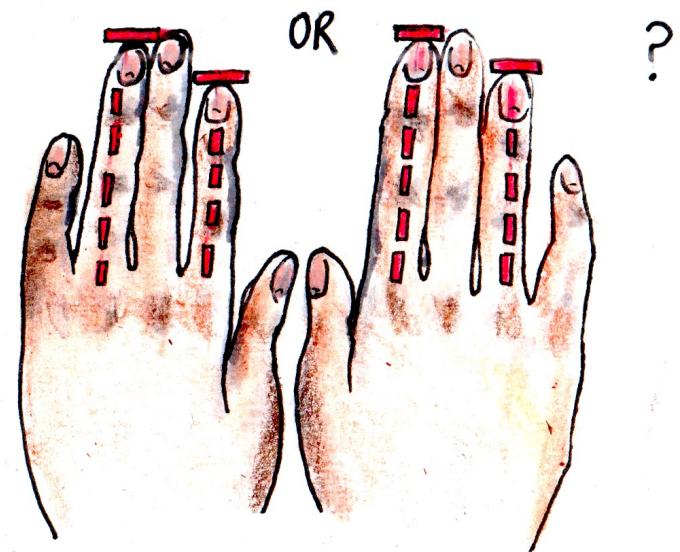
# The “lighter side” of DNA analysis



Photic Sneeze Reflex



Finger Length Ratio



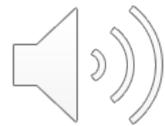
Cheek dimples

# **Challenges in personal genomics**

## **Where can data science help?**



1. How likely is it you or your doctor can take action based on genetic information?
2. How might this information impact you and your family?
3. How can we ensure access for everyone?
4. Will people understand that our environment (health care, family, society, etc.) also shapes who we are?



## Some of the slides are from or adapted from **Introduction to Personal Genetics – lots more details there!**

Personal Genetics Education Project (pgEd), Harvard Medical School [www.pged.org](http://www.pged.org) 2016

<https://pged.org/lesson-plans/#intro> PowerPoint Slides: [http://pged.org/wp-content/uploads/2016/03/2016a\\_IntroToPersonalGeneticsSlides\\_pgEd.ppt](http://pged.org/wp-content/uploads/2016/03/2016a_IntroToPersonalGeneticsSlides_pgEd.ppt)

### Image credits

"Kidzania – Doctor" by James Hart (<https://www.flickr.com/photos/jameshart/3216713992/>, accessed Jan 25, 2016). Available under a Creative Commons Attribution-NonCommercial-NoDerivs 2.0 Generic License (<https://creativecommons.org/licenses/by-nc-nd/2.0/>).

Retty Beery, The Beerys Dystonia Support Site Web Site (<http://dystonia.thebeerys.com/>, accessed Jan 25, 2016).

"Angelina Jolie" by Gage Skidmore (<https://www.flickr.com/photos/gageskidmore/4860509634/>, accessed Jan 25, 2016). Available under a Creative Commons Attribution-ShareAlike 2.0 Generic license (<https://creativecommons.org/licenses/by-sa/2.0/>).

Personal Genetics Education Project (Patricia Hautea)

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National Human Genome Research Institute (NHGRI), National Institutes of Health (NIH) (<http://www.genome.gov/sequencingcosts/>, accessed Jan 11, 2016).

Adapted from Madeleine Price Ball ([https://commons.wikimedia.org/wiki/File:Silhouette\\_of\\_Woman\\_with\\_Short\\_Hair\\_and\\_Jeans.svg](https://commons.wikimedia.org/wiki/File:Silhouette_of_Woman_with_Short_Hair_and_Jeans.svg), accessed Feb 16, 2016).

Personal Genetics Education Project (Dana Waring)