Crowdsourcing Genome Wide Association Studies

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28.12.2011

Overview

What are GWAS?

- Genome-wide Association Studies

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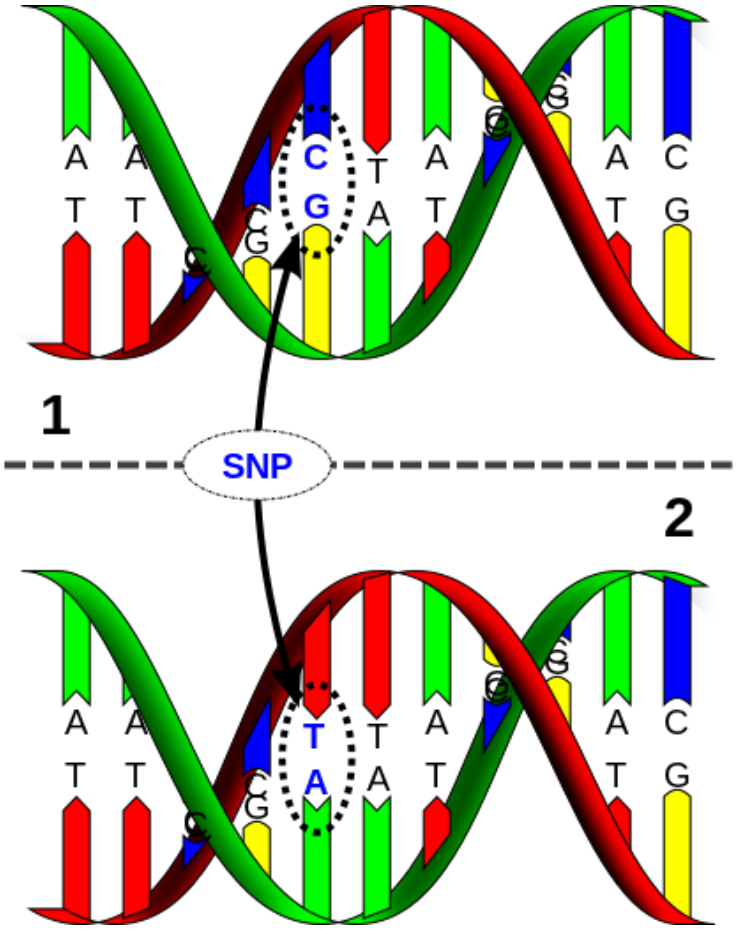
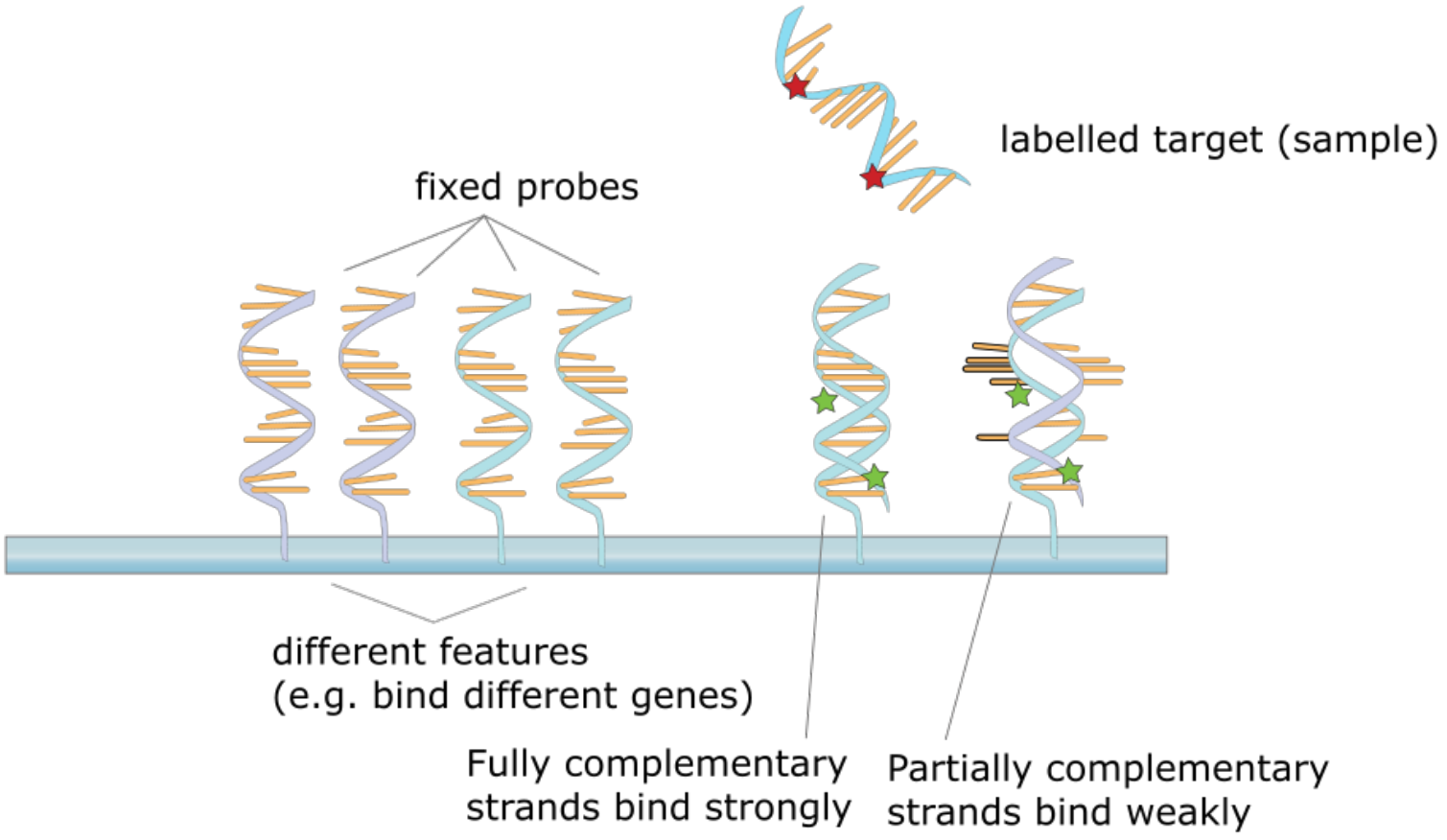
- Genome-wide Association Studies
- Link genetic variants (SNPs) to certain traits like eye or hair colour or to diseases like Diabetes, types of cancer

Single Nucleotide Polymorphism



Source: http://en.wikipedia.org/wiki/File:Dna-SNP.svg

How to analyse SNPs?



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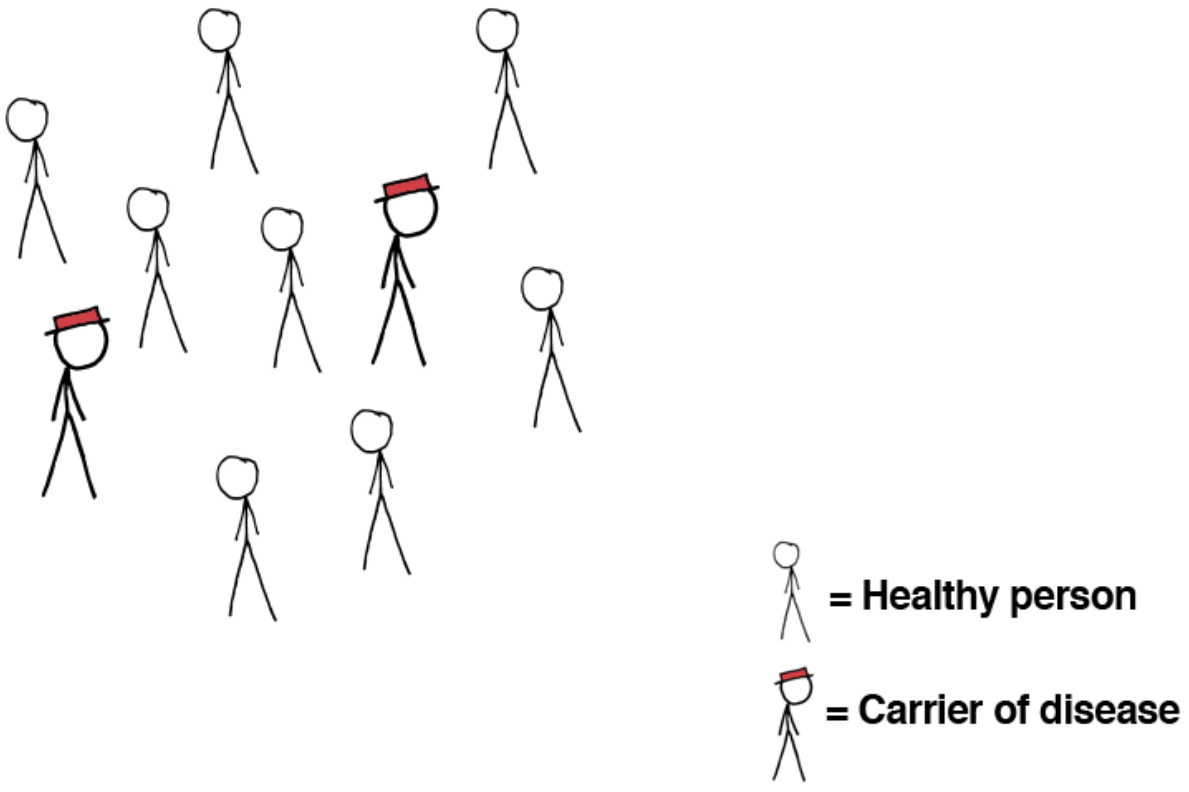
How do GWAS work?



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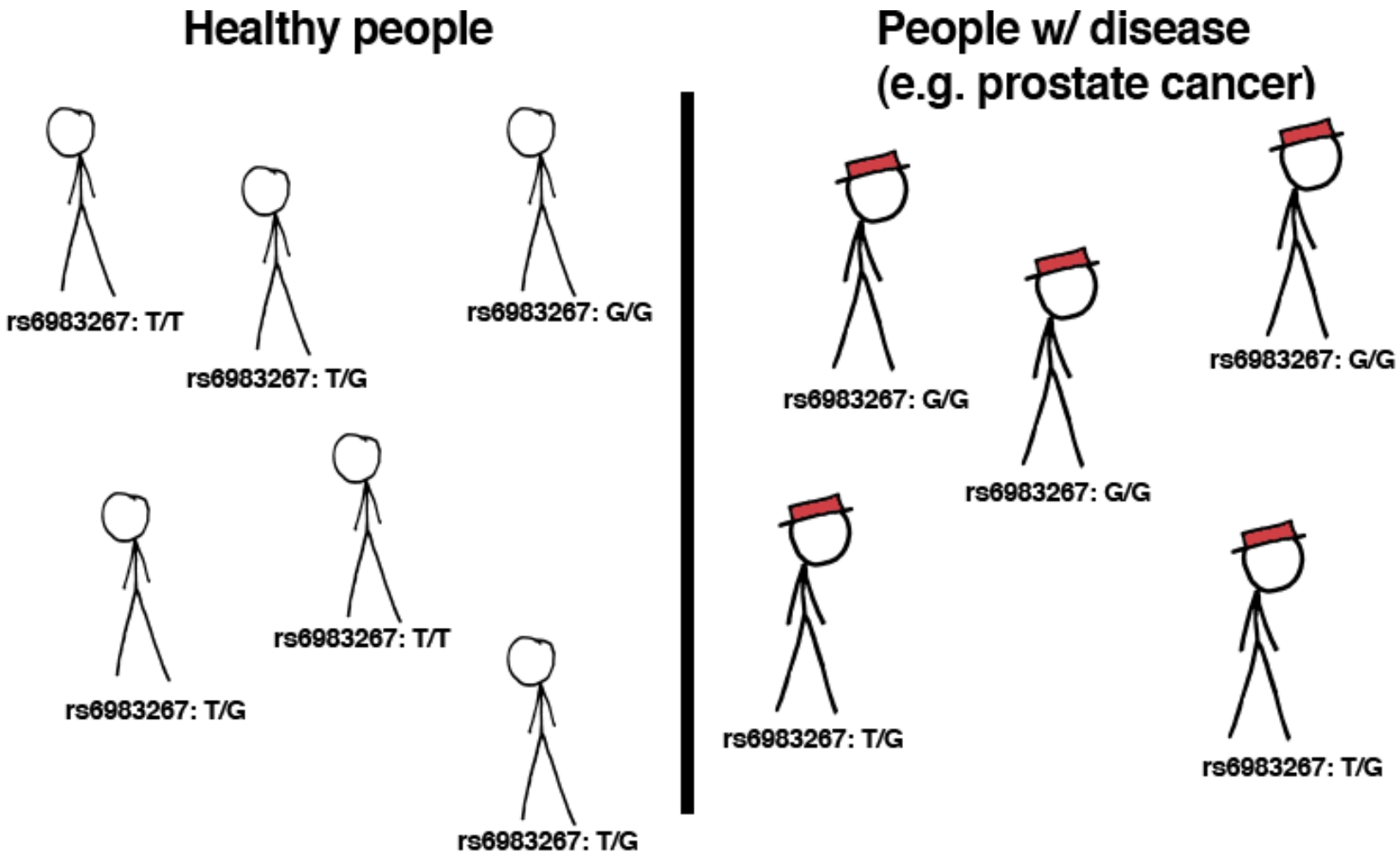

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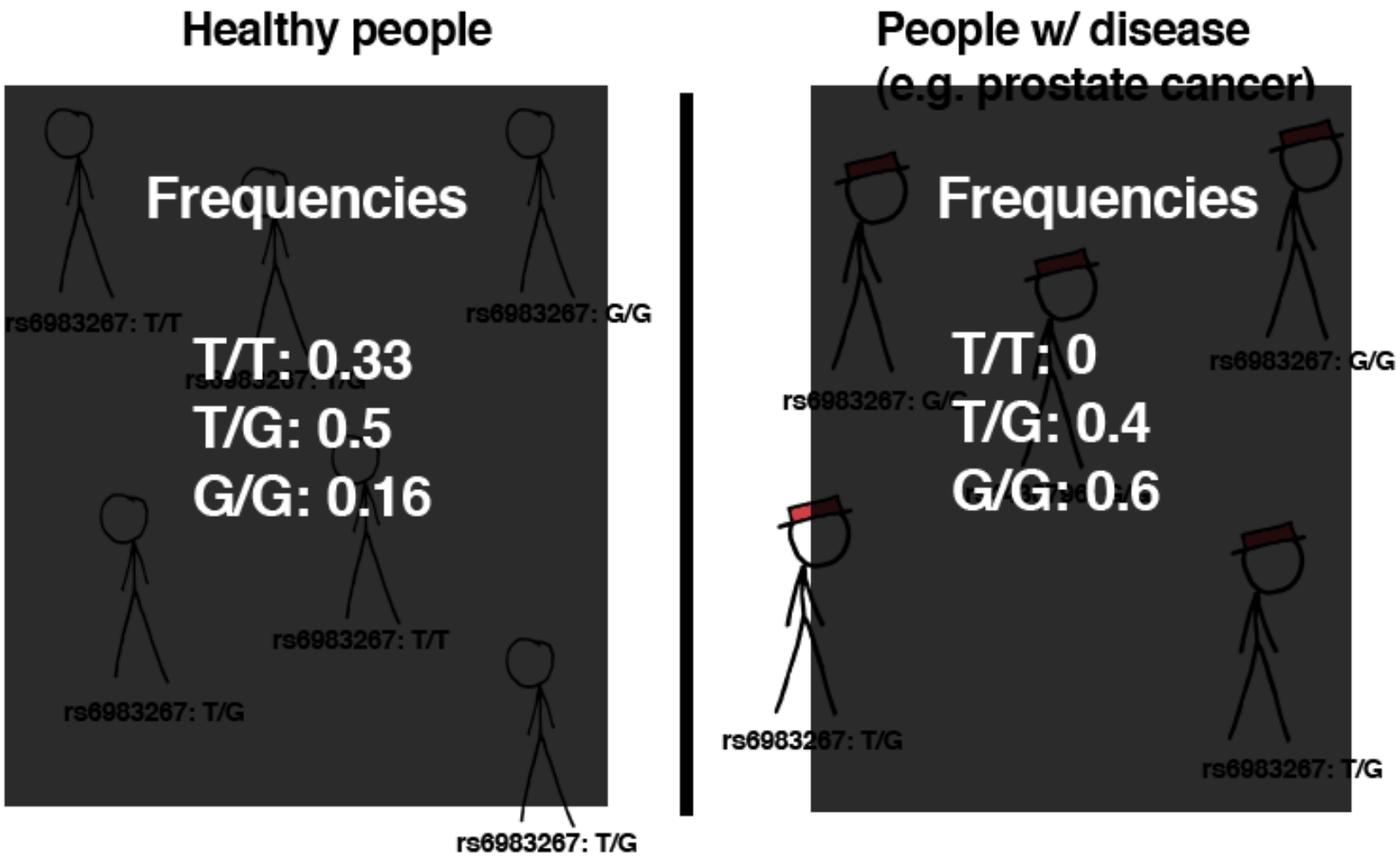

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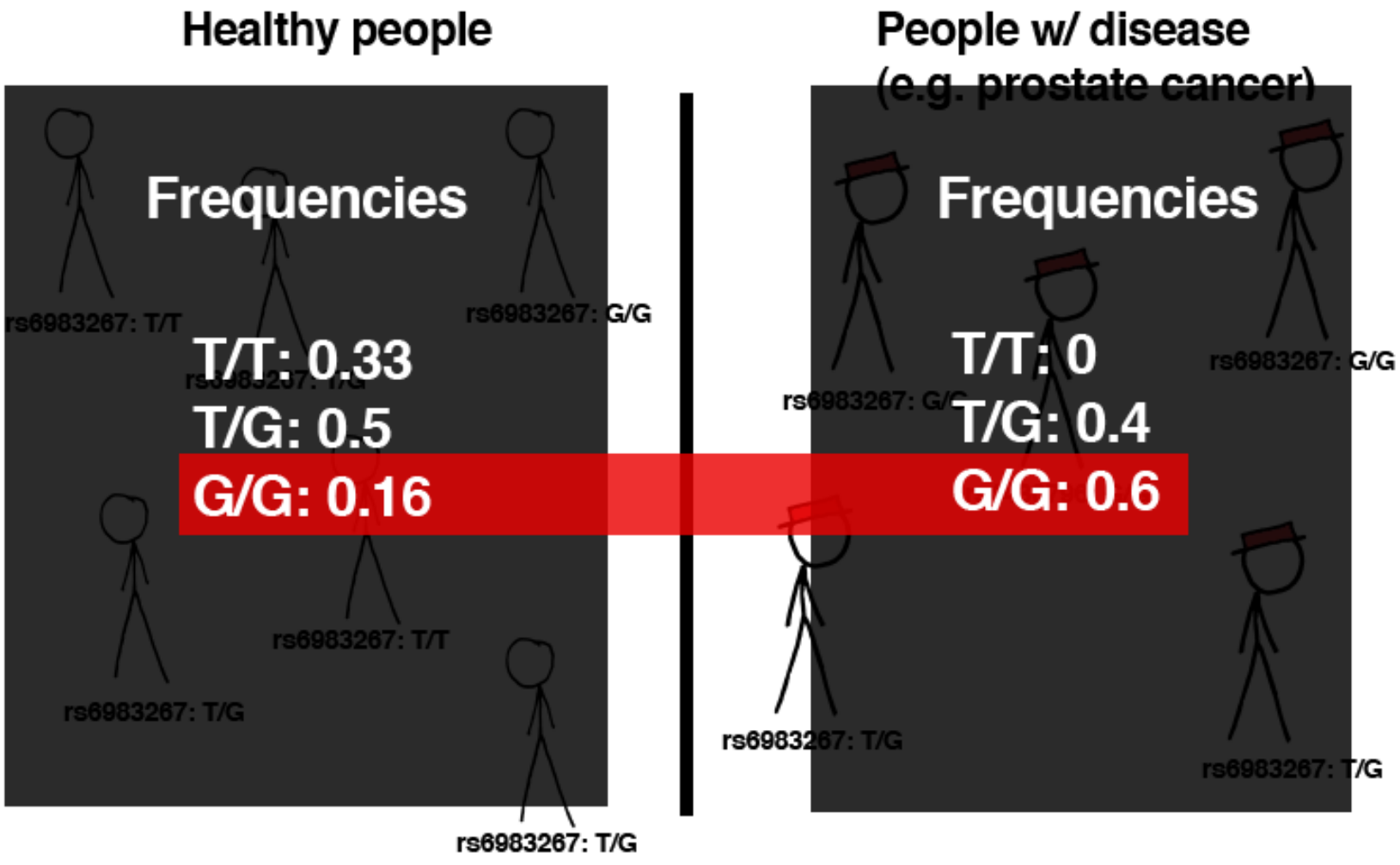

Some GWAS-examples

- Sladek *et al.* (2007) identified four gene locations linked to heightened type 2 diabetes risk

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- The Wellcome Trust Case Control Consortium (2007) linked 24 locations to 7 major diseases

Problems with GWAS

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- Correlation != Causation

Putting GWAS to use

- Direct-To-Consumer genetic testing
- Analyse about 1 million SNPs and provide summary of disease risks & ancestry
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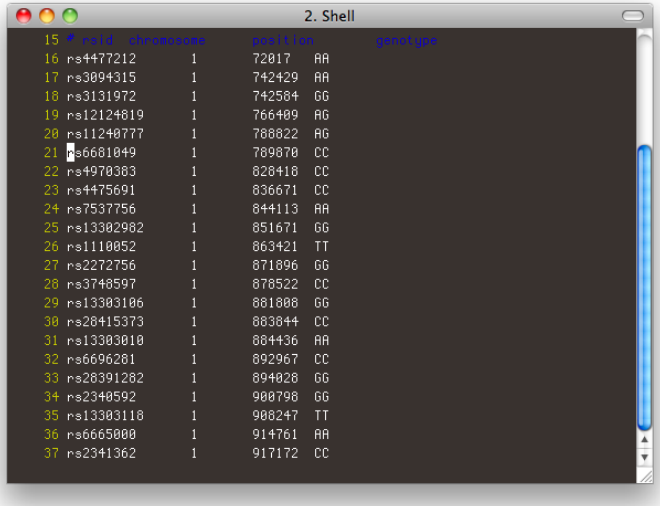
- Providers: 23andMe, deCODEme, FamilyTree DNA, ...
- You get access to the raw data!



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- 59 % of them share phenotypic information with 23andMe

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- Finding new associations for Parkinsons disease

Data sharing

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- The *Personal Genome Project*: Open data, but closed participation

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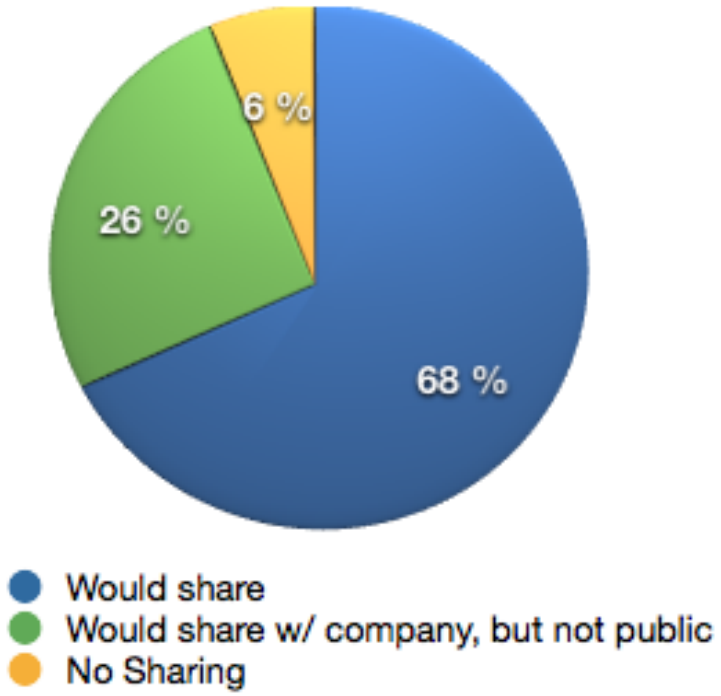

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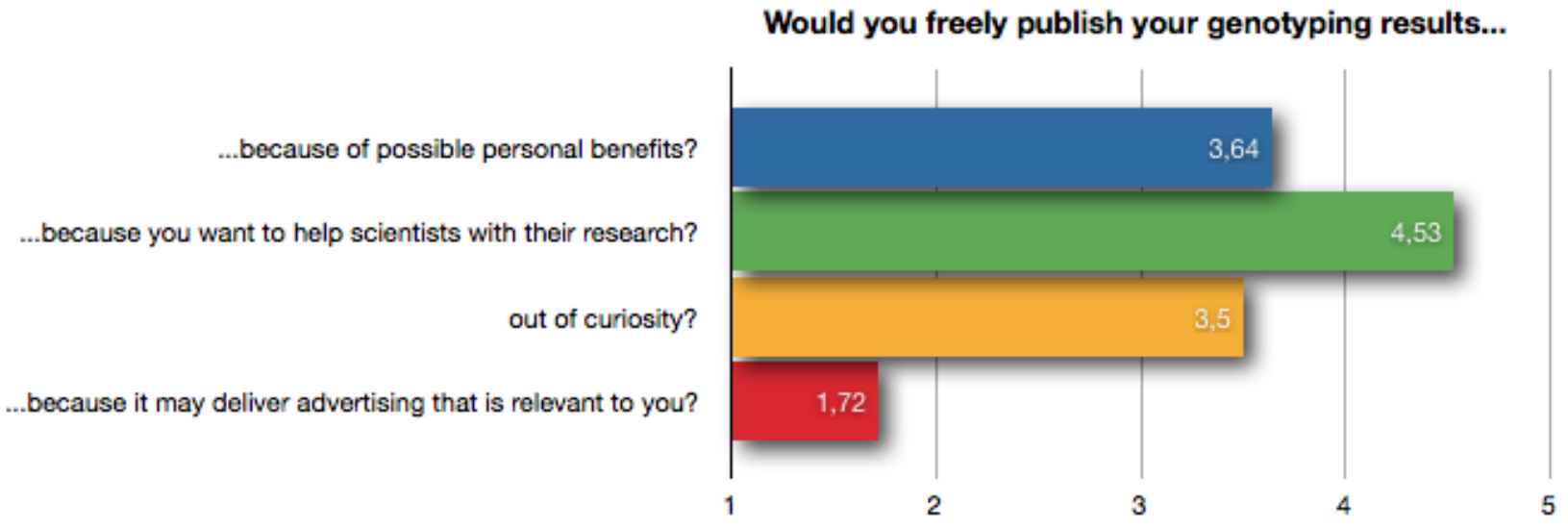

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- Up to you to decide whether you want to open your data

Positive consequences

- More knowledge about yourself

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- More knowledge about yourself
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- Great data-source for citizen scientists

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- Knowledge isn't static: Future research could show new, negative (or positive) associations.
- Personal SNPs very similar to parents and relatives

Somebody Else's Problem? A case study

Me: rs1219648(A/A)

rs1219648(A/A): 1.64x risk for breast cancer
rs1219648(A/G): 1.20x risk for breast cancer
rs1219648(G/G): "normal" risk

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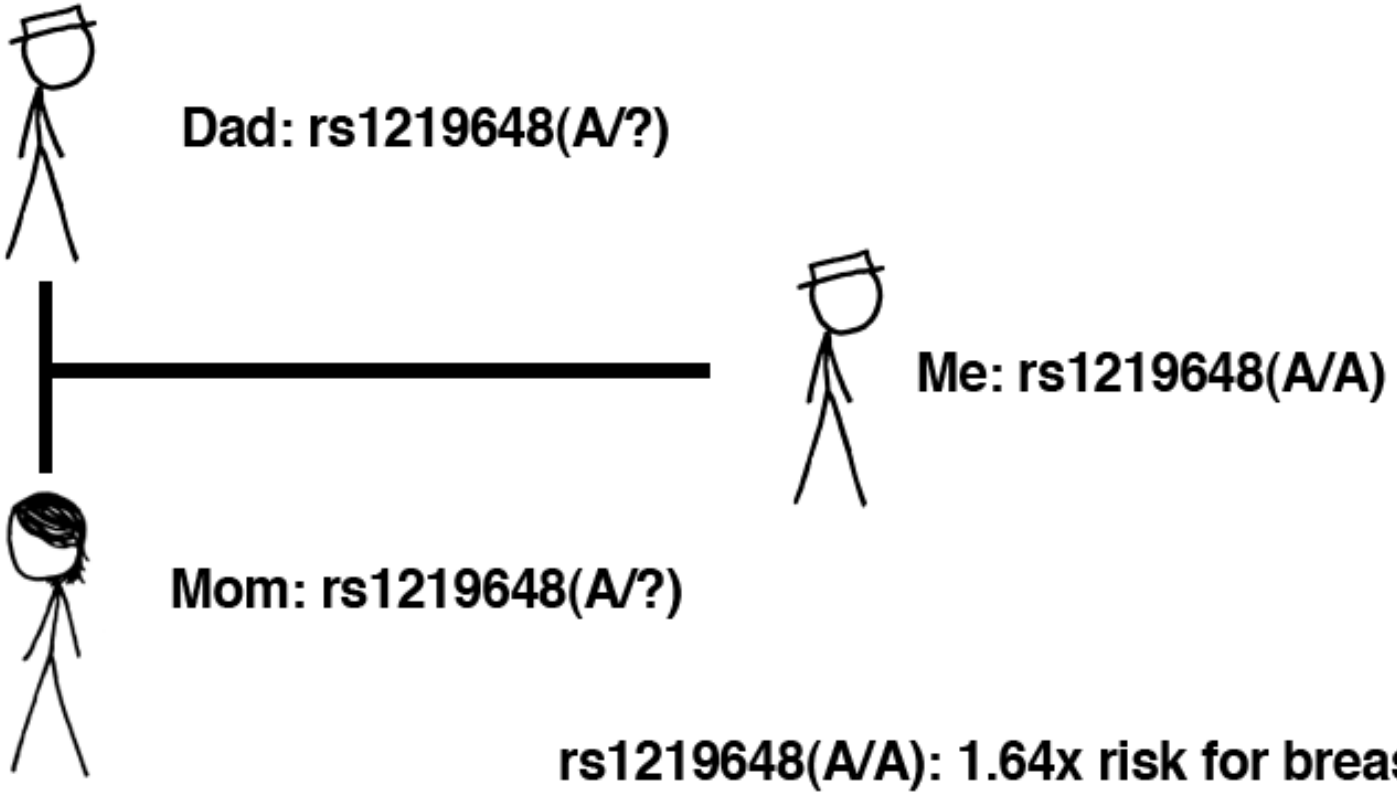

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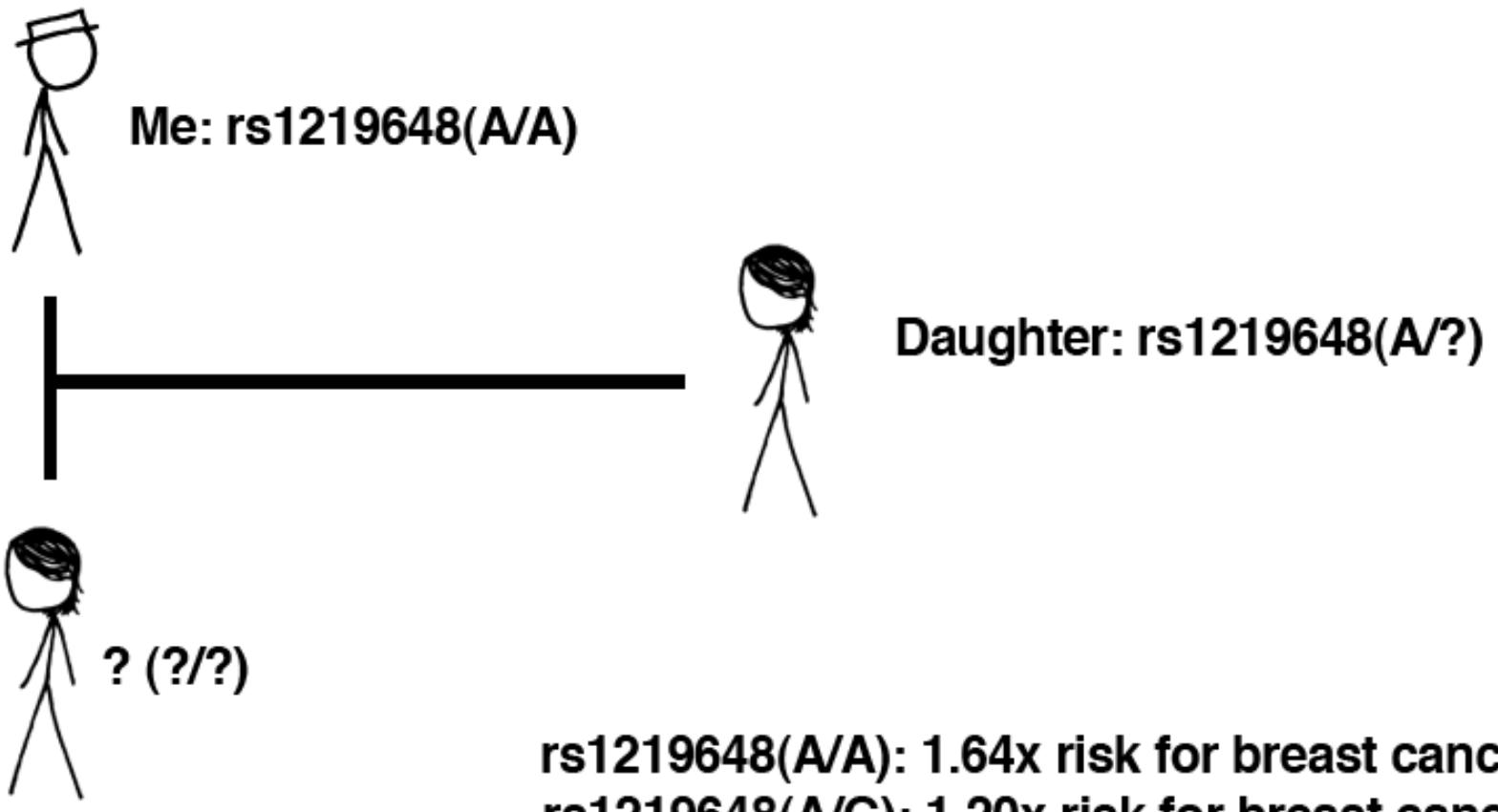

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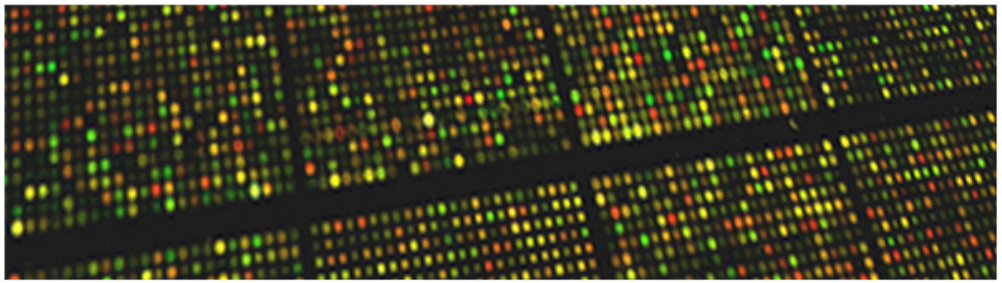
  - Germany: Gendiagnostikgesetz (GenDG, 2010)

For those who still want to share: Open GWAS

openSNP News Genotypes Phenotypes SNPs Users Search here Sign in FAQ

Welcome to *openSNP*



openSNP **allows customers of direct-to-customer genetic tests to publish their test results, find others with similar genetic variations, learn more about their results, find the latest primary literature on their variations and help scientists to find new associations.**

Sign Up!

For Genotyping Users | For Scientists | FAQ

Upload Your Genotyping File

Upload the genotyping raw-data you got from 23andMe or deCODEme to the database of *openSNP* to make it available for everybody.

Share Your Phenotypes & Traits

Share as many phenotypes, characteristics and traits with other *openSNP* users and find others with similar characteristics.
And maybe help scientists to discover new genetic associations.

Share your stories on variations & phenotypes

openSNP lets you share your stories on your genetic variations & phenotypes with others. Discover the stories of other users.
Find others to exchange experiences about your variations.

Find literature on genetic variation

openSNP gets the latest open access journal articles on genetic variations via the Public Library of Science. Additionally popular articles are indexed via the social reference manager Mendeley. Summaries are provided by SNPedia.

openSNP

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- We've created openSNP.org

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- open source repository for CC0-genotypings from 23andme, deCODEme and others

... continued

- Allows users to annotate with phenotypes (hair colour, nicotine dependence, SAT-scores...)

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- So far: 81 genotypings and 207 users

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- It's in the hands of users to make or break the situation
- Chance to take science into our own hands

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- We've won the PLoS/Mendeley Binary Battle

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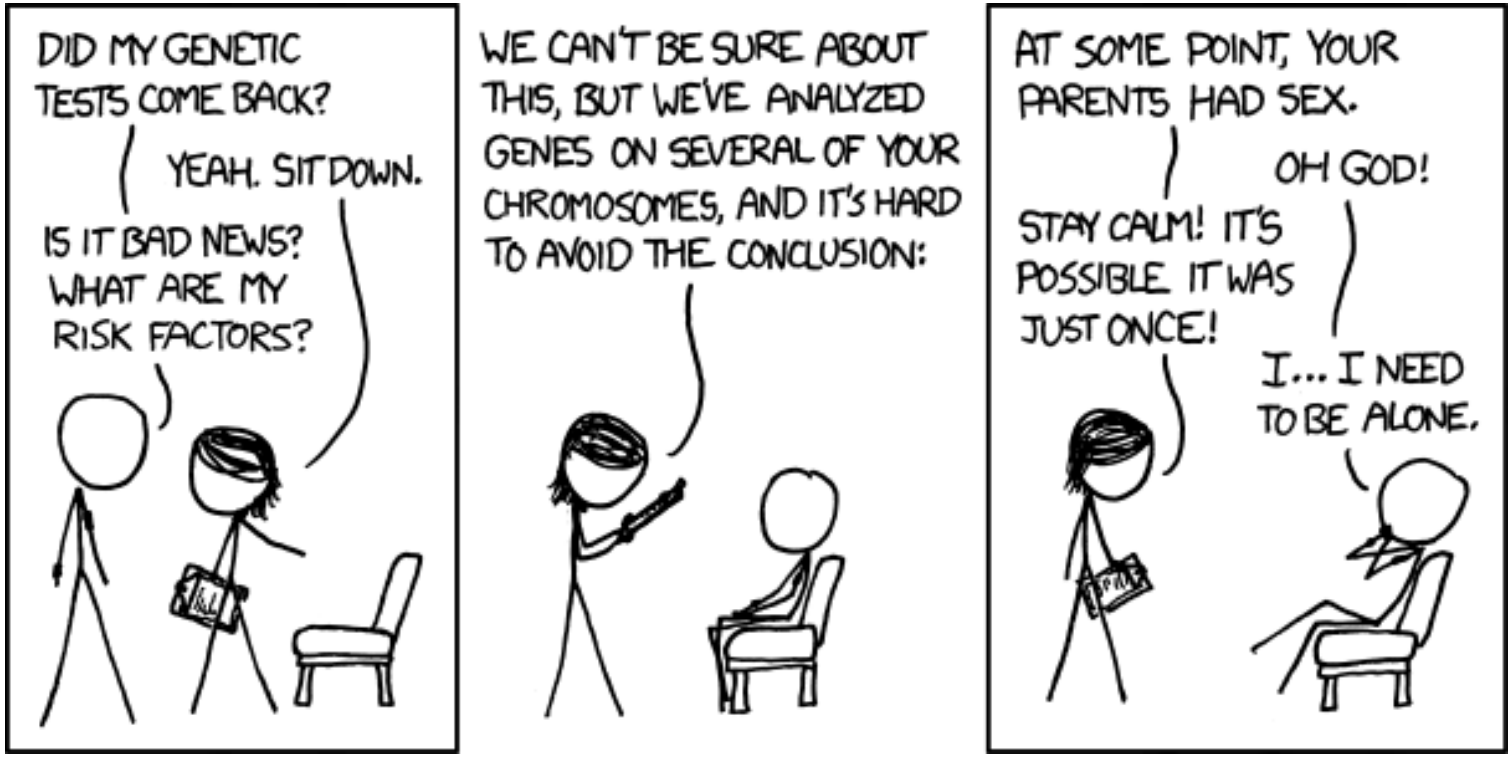
- Constantly improving the project (and are happy if somebody wants to help)

The end



Thanks for listening. Any questions?
For further questions: @gedankenstuecke
or @PhilippBayer

References

Do *et al.* (2011) Web-Based Genome-Wide Association Study Identifies Two Novel Loci and a Substantial Genetic Component for Parkinson's Disease. PLoS Genetics 7(6): e1002141. doi:10.1371/journal.pgen.1002141
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