# open SNP

personalized genotype & phenotype data in the public domain



#### 23andMe store





# DTC Genetic Testing



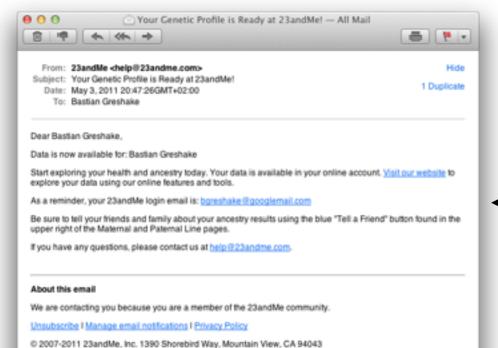






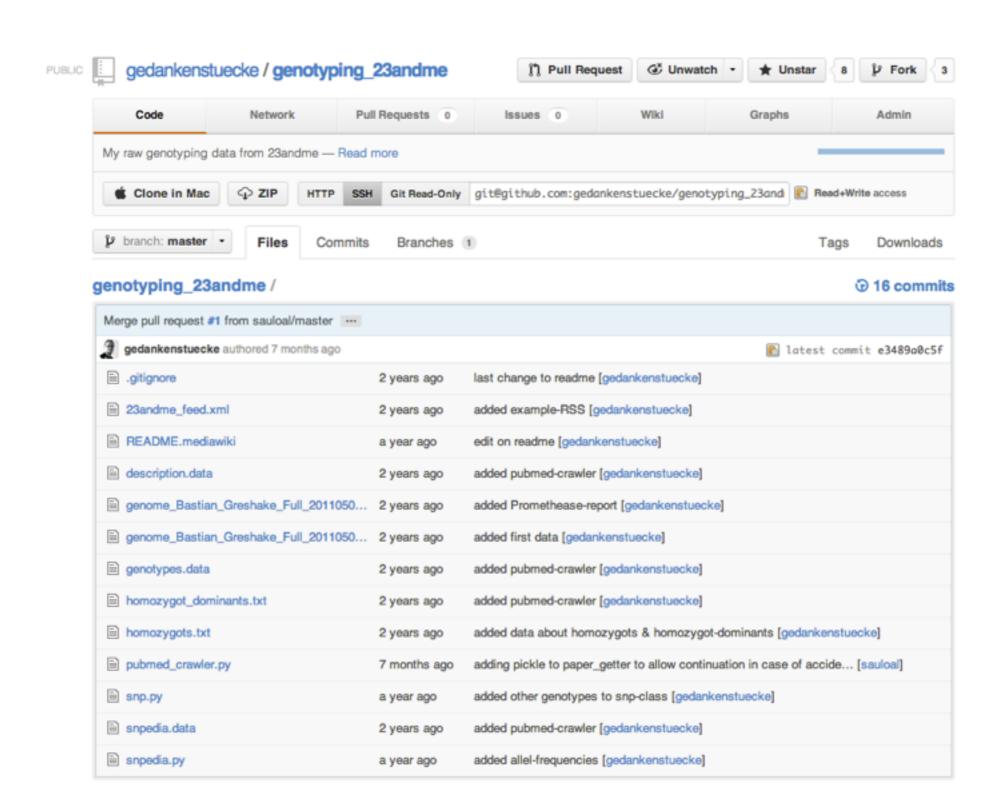




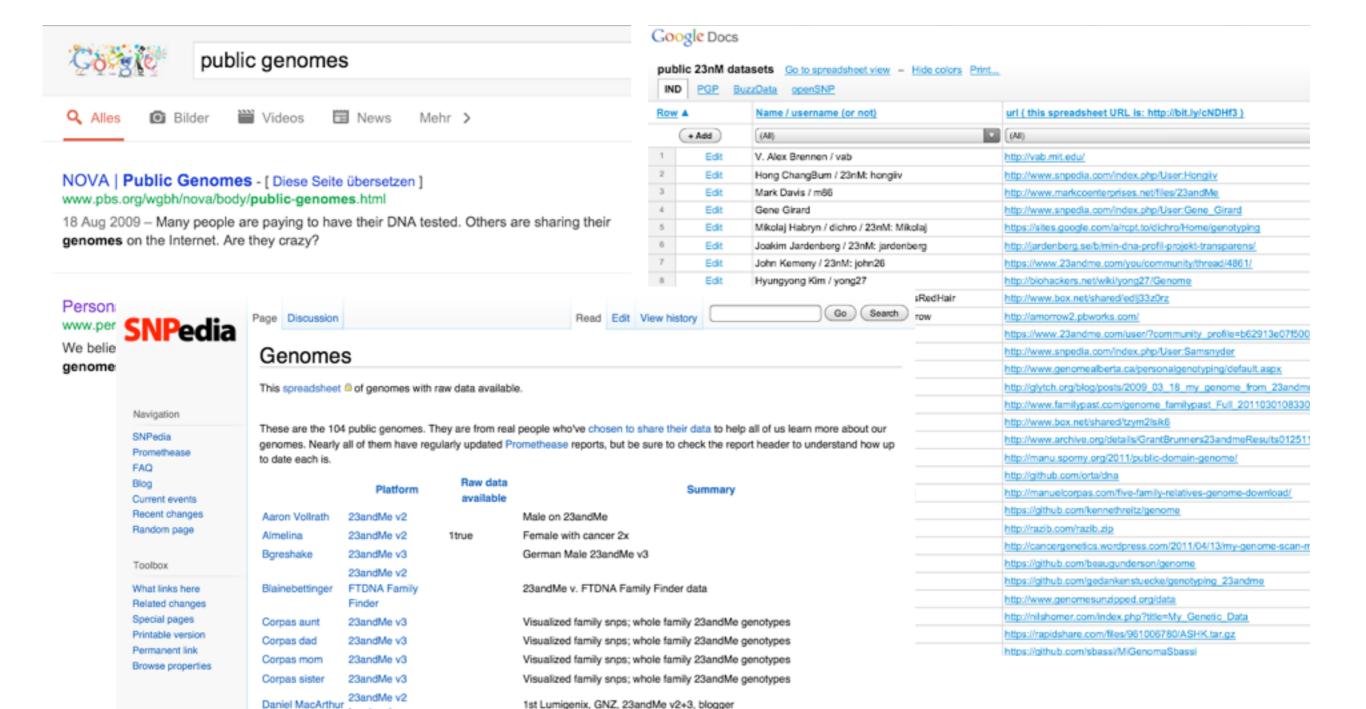


#### Potential

- 23andMe already close to 1 mio customers
- AncestryDNA projected to reach 1.3 mio customers at the end of 2015
- What if (at least some) people share their data?



## More Data?



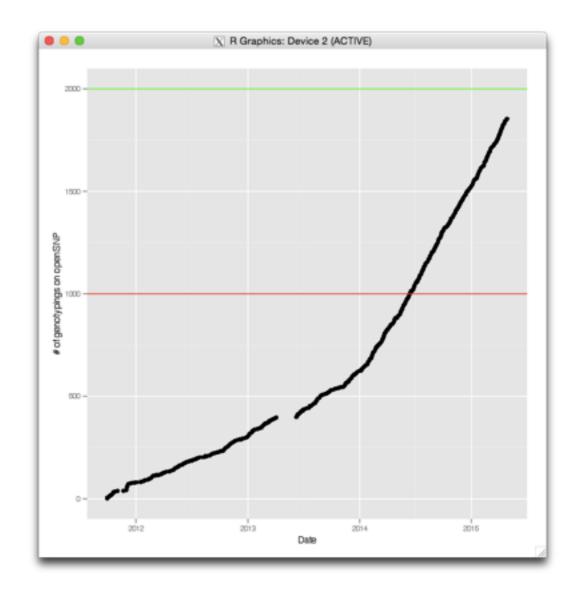


## open SNP

- Genotypes
- Phenotypes
- Metadata about SNPs

## Genotypes

- close to 2000 data sets
- formats:
  - 23andMe
  - AncestryDNA,
  - FamilyTreeDNA
  - (deCODEme)



## Phenotypes

```
Phenotype: Eye color

    Brown Download genotyping-files of all corresponding users 

    Brown-green Download genotyping-files of all corresponding users 

 · Blue-green Download genotyping-files of all corresponding users
 . Blue-grey Download genotyping-files of all corresponding users

    Green Download genotyping-files of all corresponding users 

    Blue Download genotyping-files of all corresponding users 

    Hazel Download genotyping-files of all corresponding users 

    Mixed Download genotyping-files of all corresponding users

    Gray-blue Download genotyping-files of all corresponding users 

    Blue-grey; broken amber collarette Download genotyping-files of all corresponding users 

    Dark blue Download genotyping-files of all corresponding users 

    Dark brown Download genotyping-files of all corresponding users

    Blue-green Download genotyping-files of all corresponding users 

    Light-mixed green Download genotyping-files of all corresponding users 

    Hazel/light brown Download genotyping-files of all corresponding users
```

- completely crowdsourced
- so far: free text entries (messy) + picture uploads
- ~350 different categories
- additionally: Fitbit data (weight, sleep, steps) + pictures

#### Metadata for SNPs

• Effect of a given SNP according to SNPedia



Effect of a SNP according to NHGRI-EBI GWAS catalog



- Effect according to GET Evidence of Personal Genome Project Harvard
- Papers that mention a given Rs-ID







## Access: Genotypes/SNPs

- Distributed Annotation System
- Ugly JSON-"API":
  - downloadlinks for complete files per user
  - genotypes at a SNP for a given user/all users

#### Data Points:

- Rs-ID
- Chromosome & Location
- Allele & Genotype-Frequency in our DB
- Individual Genotypes

#### Access: Phenotypes

- JSON-API:
  - By Phenotype
  - All phenotypes for a given user

#### Data Points:

- Phenotype Name
- Phenotype Description
- Known Variations
- Users & their variations

#### Access: Metadata

#### Papers:

- Title
- First Author
- Publication Date
- DOI/URL
- Open Access?
- # of Readers

#### Others:

- Gene
- Trait
- Impact
- Summary
- Inheritance
- p-value
- CI

## missing so far

- API access to Images & Fitbit data
- curated phenotypes as additional level to crowdsourced phenotypes

#### Thx!



WE CAN'T BE SURE ABOUT
THIS, BUT WE'VE ANALYZED
GENES ON SEVERAL OF YOUR
CHROMOSOMES, AND IT'S HARD
TO AVOID THE CONCLUSION:



