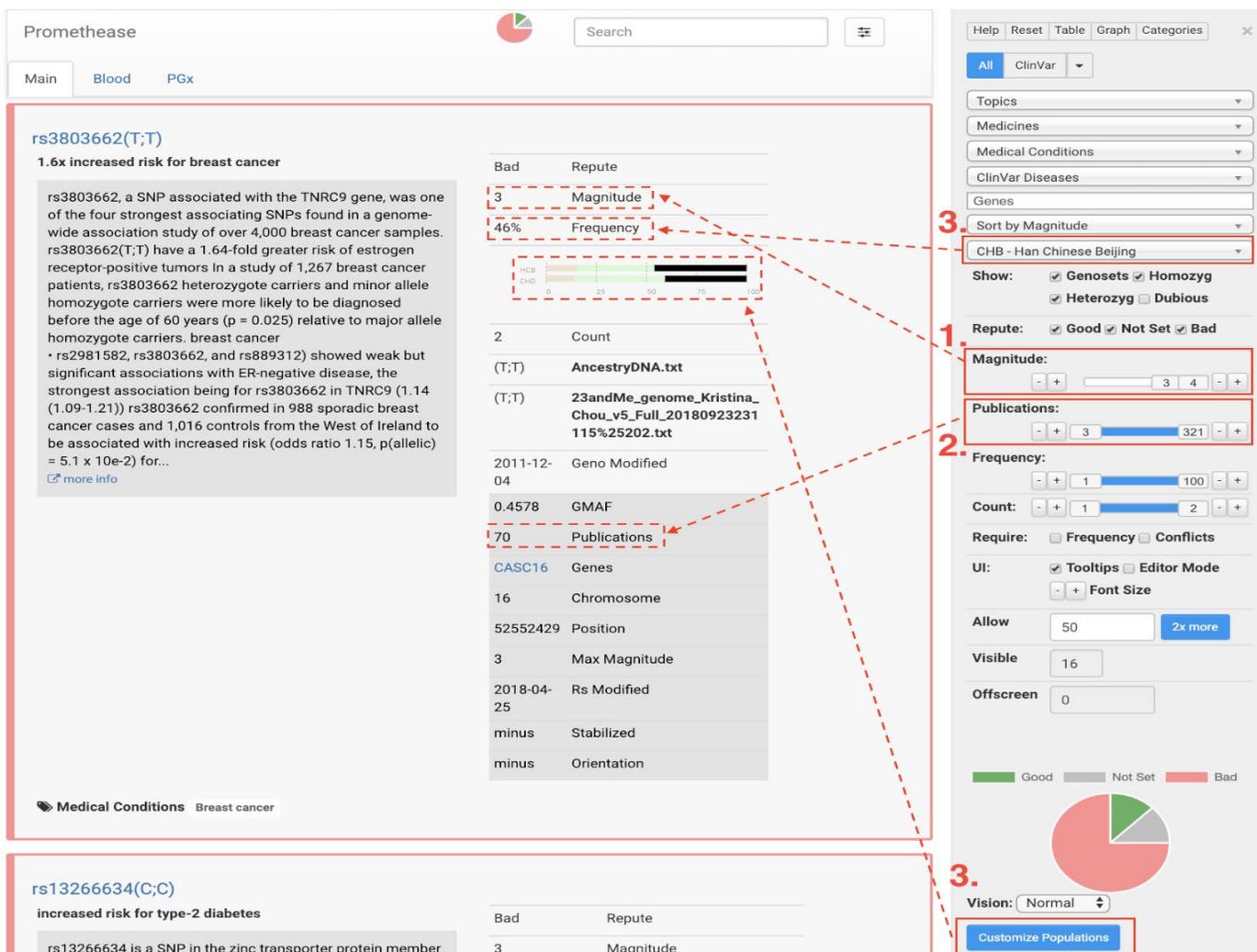


Genetic Testing With 23 and Me and Promethease

As medicine becomes more personalized, genetic information might be used to inform decisions regarding risk factors and treatment options. We aren't using it routinely in clinical practice just yet, but we will, as our understanding increases and more data becomes available. Until then, patients may choose to get their own genetic testing report from a commercial provider of testing and potentially consider it as a resource that may be useful in decision making. This information may be used by a patient to estimate risks for genetically influenced disorders and possibly modify risk factors to decrease that chance that they might develop an illness.

This guide was developed to assist in obtaining the testing and access to the searchable database. Below is a screenshot of the database.



Promethease

Search:

Main Blood PGx

rs3803662(T;T)
1.6x increased risk for breast cancer

rs3803662, a SNP associated with the TNRC9 gene, was one of the four strongest associating SNPs found in a genome-wide association study of over 4,000 breast cancer samples. rs3803662(T;T) have a 1.64-fold greater risk of estrogen receptor-positive tumors in a study of 1,267 breast cancer patients, rs3803662 heterozygote carriers and minor allele homozygote carriers were more likely to be diagnosed before the age of 60 years ($p = 0.025$) relative to major allele homozygote carriers. breast cancer

- rs2981582, rs3803662, and rs889312) showed weak but significant associations with ER-negative disease, the strongest association being for rs3803662 in TNRC9 (1.14 (1.09-1.21)) rs3803662 confirmed in 988 sporadic breast cancer cases and 1,016 controls from the West of Ireland to be associated with increased risk (odds ratio 1.15, $p(\text{allelic}) = 5.1 \times 10^{-2}$) for...

[more info](#)

Bad Repute

3 Magnitude

46% Frequency

2 Count

(T;T) AncestryDNA.txt

(T;T) 23andMe_genome_Kristina_Chou_v5_Full_20180923231115%25202.txt

2011-12-04 Geno Modified

0.4578 GMAF

70 Publications

CASC16 Genes

16 Chromosome

52552429 Position

3 Max Magnitude

2018-04-25 Rs Modified

minus Stabilized

minus Orientation

Medical Conditions Breast cancer

rs13266634(C;C)
increased risk for type-2 diabetes

rs13266634 is a SNP in the zinc transporter protein member

Bad Repute

3 Magnitude

3. Vision: Normal

Customize Populations

Help | Reset | Table | Graph | Categories

All ClinVar

Topics

Medicines

Medical Conditions

ClinVar Diseases

Genes

Sort by Magnitude

CHB - Han Chinese Beijing

Show: Genosets Homozyg Heterozyg Dubious

Repute: Good Not Set Bad

Magnitude: 3 4

Publications: 3 321

Frequency: 1 100

Count: 1 2

Require: Frequency Conflicts

UI: Tooltips Editor Mode

Font Size

Allow: 50 2x more

Visible: 16

Offscreen: 0

Good Not Set Bad

Step by Step Instructions

- You can buy the kit from <https://23andme.com> for around \$200.00.
- There is a report that is ancestry alone and there is one with ancestry and health. Please make sure you are buying the ancestry and health version.
- Order the kit, send it back for analysis and wait for an email with results.
- Once your results are received you will download your raw genetic data from 23andMe.
- You will be provided with a report and access to your “raw genome”. This data can be uploaded to a searchable database with links to literature related to specific genes.
- Go to the online site called Promethease (<https://www.promethease.com/>). Once on the website you will upload your raw genetic data, after about 15 minutes, your database will be complete, and you can download it for personal use. There is a cost associated with this. Usually around \$25.00.
- Download and save the database on your desktop, so that it is accessible for you.
- The database provides information on the genes tested in the panel. The testing looks for genotype and for SNP’s (single-nucleotide-polymorphisms), it reports your genetic type, brief description of the importance of the gene and a link to “snipedia” which has additional links to published data regarding the SNP.
- The database is organized based on “beneficial” (green) genetic variants and “harmful” (red) variants. Red variants are ranked by magnitude, the most harmful genes are magnitude 6 and the least, a magnitude of 1.
- To search the data, enter the term (“-----”) in the search box at the top of the page. Hit enter and the result should pull up, (If it is setup in the database). If your search term doesn’t pull up in the database, you may be searching for the wrong term, go into your browser’s search engine and search for the term you entered and see if another term pulls up. Genes in the database are typically reported as “rs94850495 (T;T)” The “rs” with numbers is the identification for the gene and the letters at the end are your genotype.
- This information can be used to help make decisions regarding risk factors but is not a substitute for medical advice or diagnosis. The test does not mean that you WILL or WILL NOT have a certain condition. Most health concerns are caused by several factors and genetics are only one factor. Never stop a medication without discussing with your physician or health care provider. Consult a trained professional with questions or concerns about your genetic information. This information is for educational purposes only and is NOT a substitute for medical treatment or consultation.
- Example of how to use this information: Patient looks at the database and sees that they have a gene associated with a 2.5 times average risk of developing melanoma. Patient decides to be cautious in the sun, to wear sunscreen and participate in annual skin checks with their dermatologist.
- Warning: Your genetic information will be “out there” and accessible to those with permission to access it. You may be able to see others who are biologically related to you and vice versa. You may learn that you have a risk factor for a very significant illness or disease, and this may cause psychological distress or anxiety. Please be aware of these concerns and thoughtfully proceed.