

Enactment 2

A patient comes to you (the physician) with a report from a DTC testing company showing that she is at increased risk of diabetes and wants you to help her interpret the report. Before you talk to the patient, you consult with a medical geneticist about these results.

T2DM genetic risk report

- Caucasian
- Female
- 40-50 yrs

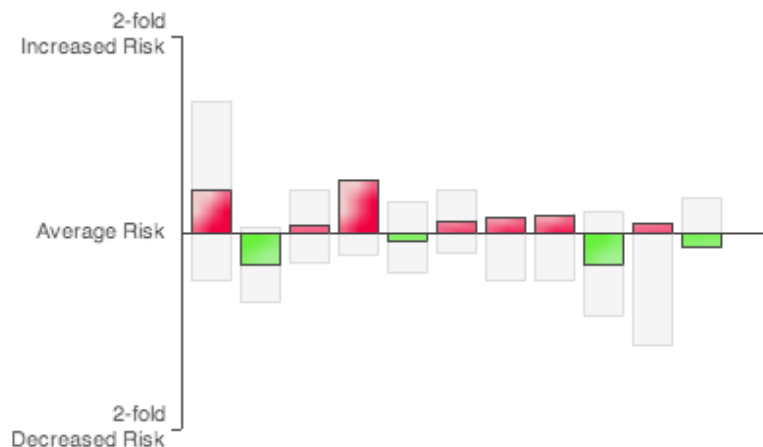
Type 2 Diabetes

Jeanette:  25%

Average:  21%

This is the estimated lifetime incidence of Type 2 Diabetes for someone with Jeanette's genotype compared to average.

Marker Effects



What does this chart show?

The chart shows the approximate effects of the selected person's genotype at the 11 reported markers. Higher, **red bars** indicate **increased risk** from the average, while lower, **green bars** indicate **decreased risk** from the average. The light gray bars show the maximum possible effects for the possible genotypes at the marker.

Mouse over individual bars to view additional information about each marker. Click on a bar to view detailed information about that marker below. You can read more about all markers in the [technical report](#).

Factors that impact risk estimates from multiSNP panels

- Assumptions about average population risk of disease
- # of SNPs used to predict disease
- How SNP effects are combined

Table 1: Average population risks and number of SNPs used by 23andMe, deCODEme, and Navigenics in the prediction of risks for six multifactorial diseases

Diseases	Average population risk (%)			Number of SNPs		
	23andMe	deCODEme	Navigenics	23andMe	deCODEme	Navigenics
Age-related macular degeneration	6.5	8	3.1	3	6	6
Atrial fibrillation	27.2	25	26	2	6	2
Celiac disease	0.12	1	0.06	4	8	10
Crohn disease	0.53	0.5	0.58	12	30	27
Prostate cancer	17.8	16	17	12	26	9
Type 2 diabetes	25.7	25	25	11	21	18

SNPs, single nucleotide polymorphisms.

Comparison across DTC genetic testing results from different companies

Table 4: Agreement among the three companies in assigning individual consumers to the same risk category, according to the risk categories used by 23andMe

Diseases	Assigned to the same risk category by all three companies			Assigned to the same risk category by two companies			Assigned to different risk categories
	↓↓↓	---	↑↑↑	↑↑- ↓↓-	--↑ --↓	↑↑↓ ↓↓↑	↑-↓
Age-related macular degeneration	52.3	0.5	15.2	6.1	6.0	12.5	7.4
Atrial fibrillation	42.4	6.7	16.7	27.3	5.7	1.2	0.0
Celiac disease	75.3	0.0	13.8	9.0	0.4	1.3	0.3
Crohn disease	51.8	0.2	3.5	13.8	3.7	19.9	7.2
Prostate cancer	15.6	4.5	13.5	29.4	21.7	6.5	9.0
Type 2 diabetes	22.2	7.8	14.7	24.1	23.1	3.2	5.0

23andMe categorizes disease risks as decreased (↓), elevated (↑), and typical (-) risks if the risks of disease are lower than 20% below the average population risk, higher than 20% above the average population risk, and in between, respectively. Values are percentages. For example, ↓↓↓ indicates the percentage of individuals that were at decreased risk according to all three companies, and ↑-↓ indicates the percentage of individuals for which the three companies predicted risks in three different risk categories.

Perfect concordance across all 3 companies: 33-89%

Where to find objective information about tests



<http://currents.plos.org/genomictests/>

Also...

- Professional guidelines
- Technology assessments

EGAPP: Evaluation of genomic applications in practice and prevention



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

T2DM multigene review from EGAPP



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

A screenshot of the "Genetics in Medicine" journal website. The header shows the journal title "Genetics in Medicine" and its subtitle "Official Journal of the American College of Medical Genetics". Navigation links include "Home", "Current Issue", "Archive", "Podcasts", "For Authors & Referees", and "About the journal". Below this is a breadcrumb trail: "Archive > Volume 15 > Issue 8 > Article". The main heading of the article is "GENETICS IN MEDICINE | EGAPP RECOMMENDATION STATEMENT". The article title is "Recommendations from the EGAPP Working Group: does genomic profiling to assess type 2 diabetes risk improve health outcomes?". Below the title is the author information: "Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group*". The citation information is "Genetics in Medicine (2013) 15, 612–617 | doi:10.1038/gim.2013.9". At the bottom, it says "Received 24 September 2012 | Accepted 16 January 2013 | Published online 14 March 2013".

Genetics in Medicine Official Journal of the American College of Medical Genetics

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Archive > Volume 15 > Issue 8 > Article

GENETICS IN MEDICINE | EGAPP RECOMMENDATION STATEMENT

Recommendations from the EGAPP Working Group: does genomic profiling to assess type 2 diabetes risk improve health outcomes?

Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group*

Genetics in Medicine (2013) 15, 612–617 | doi:10.1038/gim.2013.9

Received 24 September 2012 | Accepted 16 January 2013 | Published online 14 March 2013

EGAPP Recommendation Statement: The EGAPP Working Group (EWG) found **insufficient evidence to recommend testing** for predictive variants in 28 variants (listed in Table 1 of the recommendation) to assess risk for Type 2 Diabetes in the general population, based on studies in populations of northern European descent. The EWG found that the magnitude of net health benefit from use of any of these tests alone or in combination is close to zero. The EWG discourages clinical use unless further evidence supports improved clinical outcomes.

Key concepts from Enactment 2

- Multi-marker genetic risk panels should be interpreted with caution
- Genes identified to date poorly explain genetic underpinnings of complex diseases
- Several resources exist to find objective information/guidance on tests
- Genetic testing should not distract from modifiable risk factors (diet, lifestyle, medication)
- There are two sides to the controversy over direct-to-consumer testing