

Make a perfect man



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[Pull requests](#) [Issues](#) [Gist](#)



Manu Sporny

msporny

Founder, CEO, and advisor working on financial inclusion and next generation Web technologies (identity, payments, linked data)

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Joined on 25 Jul 2009

130 14 0

Overview

Repositories

Public activity

Follow

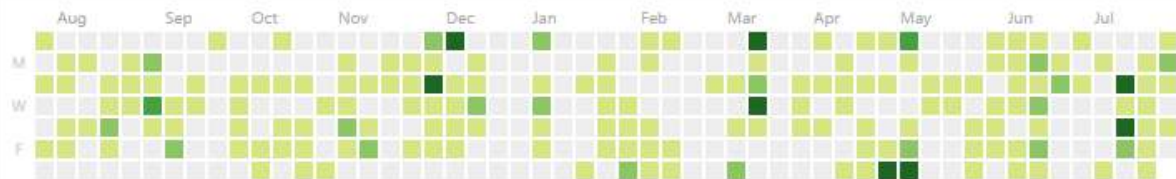
Block or report



Popular repositories

dna	Manu Sporny's genetic information (roughly 1 million SNP markers)	371 ★
live-loop	Live Loop lets you play with HTML+RDFa markup and instantly see the data that your markup produces.	8 ★
coductivity	A tool to measure productivity over a set of github repositories	5 ★
fuzz	Fuzz is a native Semantic Web Processor that understands RDFa. It is available as a Firefox Add-on. Fuzz is most useful ...	4 ★
ruby-sparql	Integrated Ruby SPARQL library	2 ★

719 contributions in the last year



Summary of pull requests, issues opened, and commits. Learn how we count contributions.

Less More

#	rsid	chromosome	position	genotype
	rs4477212	1	72017	AA
	rs3094315	1	742429	AA
	rs3131972	1	742584	GG
	rs12124819	1	766409	AA
	rs11240777	1	788822	AG
	rs6681049	1	789870	CC
	rs4970383	1	828418	AA
	rs4475691	1	836671	CT
	rs7537756	1	844113	AG
	rs13302982	1	851671	GG
	rs1110052	1	863421	GG
	rs2272756	1	871896	AG
	rs3748597	1	878522	CC
	rs13303106	1	881808	AA
	rs28415373	1	883844	CC
	rs13303010	1	884436	AA
	rs6696281	1	892967	CC
	rs28391282	1	894028	GG
	rs2340592	1	900798	AA
	rs13303118	1	908247	GT
	rs6665000	1	914761	AA
	rs2341362	1	917172	CC
	rs9777703	1	918699	TT
	rs1891910	1	922320	GG
	rs9697457	1	924208	GG
	rs35940137	1	930066	GG
	rs3128117	1	934427	CT
	rs2465126	1	936897	AA
	rs2341365	1	938555	AA
	rs15842	1	938784	CC
	rs6657048	1	947503	CC
	rs2710888	1	949705	CT
	rs3128126	1	952073	AG

SNP Data Bases

- dbSNP



- F-SNP

F-SNP: a collection of functional SNPs,
specifically prioritized for disease association studies

- ClinVar

ClinVar

- SNPedia

SNPedia

- GWAS central



Promethease is a literature retrieval system that builds a personal DNA report based on connecting a file of DNA genotypes to the scientific findings cited in [SNPedia](#).

Biomedical researchers, healthcare practitioners and customers of DNA testing services (such as [23andMe](#), [Ancestry.com](#), [FamilyTreeDNA](#), etc.) use Promethease to retrieve information published about their DNA variations. Most reports cost \$5 and are produced in under 10 minutes. Much larger data files (such as imputed full genomes from [dna.land](#)) cost \$10 and have increased runtime.



[main page](#)

Promethease Report

Version: 0.1.166

Generated: 2016-07-27 11:30

Infile: C:\Users\Daniel\Documents\ManuSporny-genome.txt

Reference Population GIH

25004 genotypes annotated

[Help](#) interpreting your results.

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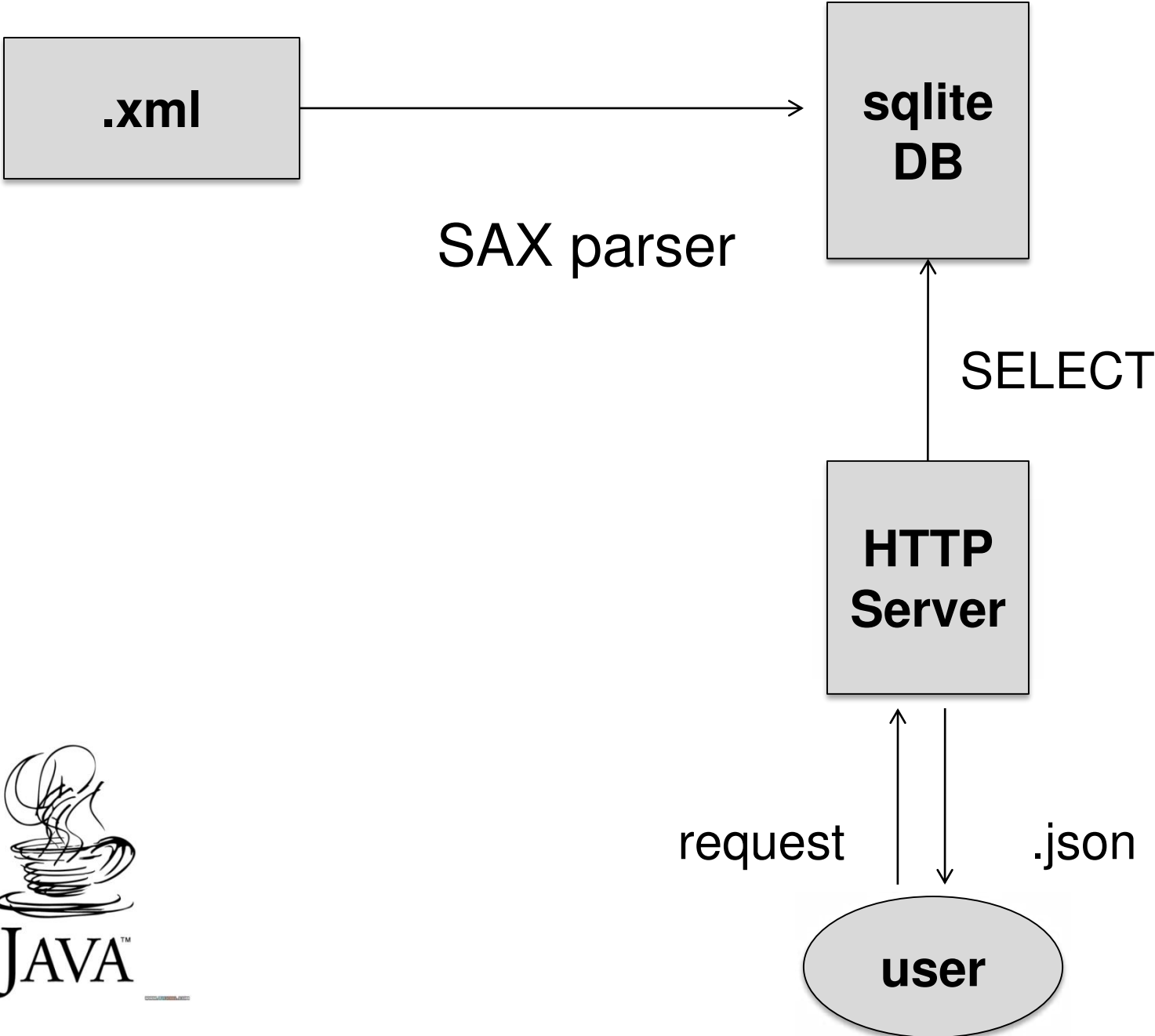
Good news

[\(hide\)](#)

rs273 Magnitude: 3 Repute: Good	Lowest risk (13% of white women) of Atrial Fibrillation reported by 23andMe The two atrial fibrillation SNPs mentioned by 23andMe are both normal. This causes the heart to form normally and gives the lowest risk of Atrial Fibrillation (quivering of the top part of the heart) that 23andMe reports. It also seems to lower the risk of Cardioembolic ischemic stroke (blocked blood flow to the brain) although 23andMe doesn't mention that. For European women, 23andMe reports this as a 13.0% risk, compared to the 15.9% average. The risk is higher for men. Based on many quality studies, and confirmed for Europeans and partly for Asians.
rs5848(G;G) Magnitude: 3 Frequency: 56.6% Repute: Good References: 26	lower dementia risk Lower risk of frontotemporal dementia, alzheimer's disease and parkinson's disease. ...more...
rs1800497(C;C) Magnitude: 3 Frequency: 66.4%	Normal (A2/A2): Better avoidance of errors. Normal OCD risk, normal Tardive Diskinesia risk, lower ADHD risk. Less Alcohol dependence. Higher risk of Postoperative Nausea. Lower obesity. Bupropion is effective. This DRD2 TaqIA A2/A2 version causes a normal amount of Dopamine Receptors. Learns from mistakes more easily. Men may have a higher risk of Obsessive Compulsive Disorder but lower risk of ADHD. Women have higher

SNP id	SNP → Normal	Risk of disease
rs10490924	(T;T) → (G;G)	risk for age related macular degeneration
rs13202464	(A;G) → (A;A)	risk for B27 Syndromes
rs1800562	(A;G) → (G;G)	risk for hemochromatosis
rs2145418	(G;G) → (T;T)	risk for thyroid cancer
rs2981582	(C;T) → (C;C)	risk for ER+ breast cancer
rs7754840	(C;G) → (G;G)	risk for type-2 diabetes
rs1333049	(C;G) → (G;G)	risk for CAD
rs53576	(A;G) → (G;G)	lack of empathy
rs210138	(G;G) → (A;A)	testicular cancer
rs10830963	(C;G) → (C;C)	risk for type-2 diabetes
rs6983267	(G;T) → (T;T)	risk for prostate cancer
rs4988235	(C;C) → (C;T)/(T;T)	risk for lactose intolerant as an adult
rs891512	(A;G) → (G;G)	Higher blood pressure than G;G

SNP id	SNP → Normal	Risk of disease
rs7574865	(G;T) → (G;G)	risk of rheumatoid arthritis, type-1 diabetes, and primary biliary cirrhosis
rs9886784	(A;G) → (A;A)	risk for Alzheimer's disease
rs13266634	(A;G) → (G;G)	risk for Alzheimer's disease
rs13266634	(C;T) → (T;T)	risk for type-2 diabetes
rs3738919	(C;C) → (A;A)	risk of developing rheumatoid arthritis
rs1421085	(C;T) → (T;T)	risk for obesity
rs1121980	(C;T) → (C;C)	risk for obesity
rs664143	(C;T) → (C;C)	higher risk for number of cancers
rs36053993	(A;G) → (G;G)	risk for colorectal cancers



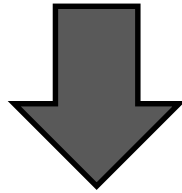
JAVA™

Example

<p>← → ↻ localhost:9000</p> <p>SNP. 21 chromosome.</p> <p>530</p> <p>Send</p> <p>530: A/T</p>	<p>← → ↻ localhost:9000</p> <p>SNP. 21 chromosome.</p> <p>530, 716, 1441, 2217, 6370, 2180, 1299, 1141, 7337, 684, 3153, 3403, 1693, 6378</p> <p>Send</p> <p>530: A/T 684: A/G 716: C/T 1141: A/G 1299: A/G 1693: A/G 2180: A/G 2217: C/T 3153: C/T 3403: C/T 6370: C/T 6378: A/G 7337: C/G</p>
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Variants of progress

- Optimisation of parsing
- Creating your own system for database
- Adding more information about SNPs



Happy bioinformatic



Yellow team

- Королева Яна
- Прокофьева Наталия
- Карп Татьяна
- Даниил Байзигитов