

PERSONAL GENOME PROFILE 3:

Colin Smith, a genomics professor from England, was the first participant of the UK arm of the Personal Genome Project. Colin is an advocate for “open consent” and making genomic and health data publicly available. He donated his information to contribute to understanding of the human genome.

Colin’s information is on the personalgenomes.org.uk site as **uk4CA868**. His public profile can be found at <https://my.personalgenomes.org.uk/profile/uk4CA868>.

His profile includes raw data from online genetic testing company 23&me, a report from whole genome sequencing, and a report from analysis of DNA methylation.

You can read Colin’s thoughts on donating his genome in this article from *The Conversation* <https://theconversation.com/why-i-donated-my-entire-genome-sequence-to-the-public-83741>

or hear him talk about it in this 2.5min clip <https://www.youtube.com/watch?v=6gLa6I04hTc>



- Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer
3	rs1800460	(A;G)	(TPMT*3B) impaired drug metabolism
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes
3.0	rs1142345	(A;G)	TPMT*3C . impaired drug metabolism
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis
2.5	rs1121980	(C;T)	1.67x risk for obesity
2.5	rs1154155	(G;G)	2.5x increased risk for narcolepsy
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes
2.5	rs1421085	(C;T)	~1.3x increased obesity risk
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer’s
2.5	rs339331	(T;T)	Prostate cancer risk
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...
2.4	rs1143679	(A;G)	1.78x increased risk for SLE
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke
2.2	rs944289	(T;T)	2.6x increased thyroid cancer risk
2.1	rs1219648	(G;G)	1.64x risk for breast cancer
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...
2.1	rs241448	(C;C)	2.14x increased risk for Alzheimer’s
2.1	rs2420946	(T;T)	1.64x risk for breast cancer

Connect with the Garvan Institute’s Kinghorn Centre for Clinical Genomics:

Visit us <http://www.garvan.org.au/kccg>

Follow our newsfeed on Twitter @kinghorngenomes

Email us at kccgenquiries@garvan.org.au

KINGHORN
CENTRE
FOR CLINICAL
GENOMICS

