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## The genetics of common diseases

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At deCODE genetics in Iceland we have sequenced the whole genomes of 50.000 Icelanders or 12.5% of the nation and genotyped 180.000 or 60% of the nation. In addition we have the genealogy of the entire nation going centuries back in time on a computer database. This allows us to impute sequence variants with allelic frequency down to about 0.01% into all genotyped individuals and their first and second degree relatives. We are in the privileged position of being able to phase the entire genomes of all Icelanders. This has allowed us to determine whether there is a difference in the impact of sequence variants depending on the parent it is inherited from. Taking advantage of this we have found sequence variants that increase the risk of disease when it is inherited from one sex and protect against the same when it is inherited from the other sex. Furthermore, we have discovered a number of sequence variants where there is a difference in the size of the effect depending on the parent of origin. Hence, a parent of origin analysis of associations between variants in the sequence and diversity in phenotypes is a part of our daily routine. Furthermore, we have combined whole genome oxidative bisulfite sequencing of 285 individuals and allele specific RNA sequencing of 11,617 blood samples with parnet-of-orgin phased haplotyes, to produce a new map of imprinted methylation and gene expression pattern across the human genome. Through this we have, for example, gained new insights into parent of origin specific effects on phenotypes.

### Biography

Kári Stefánsson, M.D., Dr. Med. has served as President, Chief Executive Officer and a Director since he founded deCODE genetics in August 1996. Dr. Stefánsson was appointed the Chairman of the Board of Directors of deCODE genetics in December 1999. From 1993 until April 1997, Dr. Stefánsson was a professor of Neurology, Neuropathology and Neuroscience at Harvard University. From 1983 to 1993, he held faculty positions in Neurology, Neuropathology and Neurosciences at the University of Chicago. Dr. Stefánsson received his M.D. and Dr. Med. from the University of Iceland and is board-certified in neurology and neuropathology in the United States.

He has published numerous articles on the genetics of common/complex diseases and has been among the leaders of the world in the discovery of variants in the sequence of the human genome that associate with the risk of common/complex traits. Dr. Stefánsson was chosen by Time magazine as one of the 100 most influential men of the year for 2007 and by Newsweek as one of the 10 most important biologists of the 21 century.

He was the recipient of the Jakobus Award 2007, The World Glaucoma Association Award for present scientific impact 2007, The European Society of Human Genetics Award 2009, and The Andre Jahre Award 2009.