

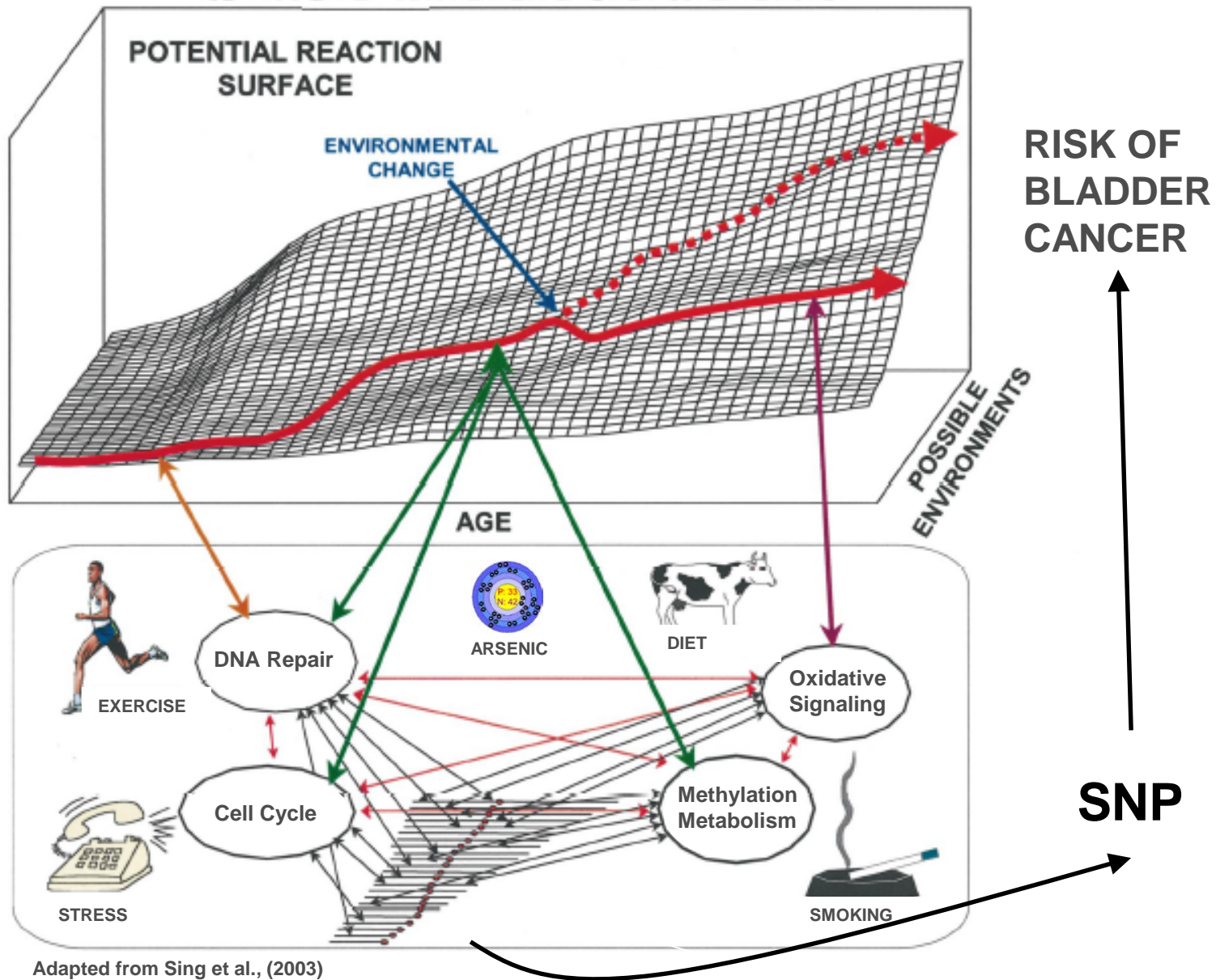
Human Phenotype Networks

Jason H. Moore, Ph.D.

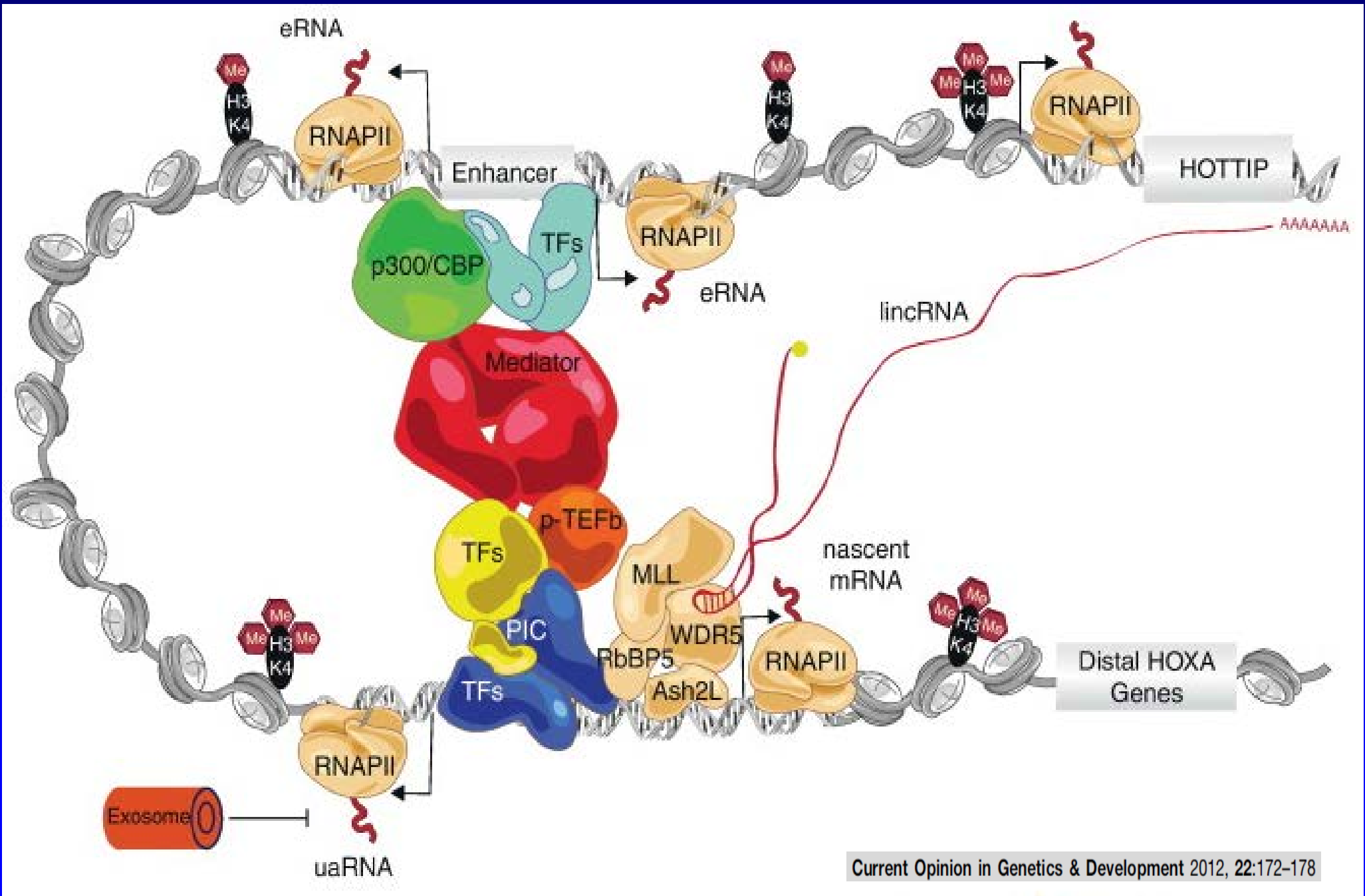
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Adapted from Sing et al., (2003)



“One SNP at a time univariate approach”

20%

Main Effects

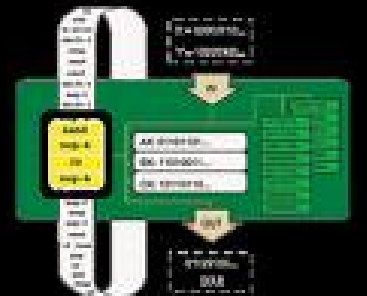
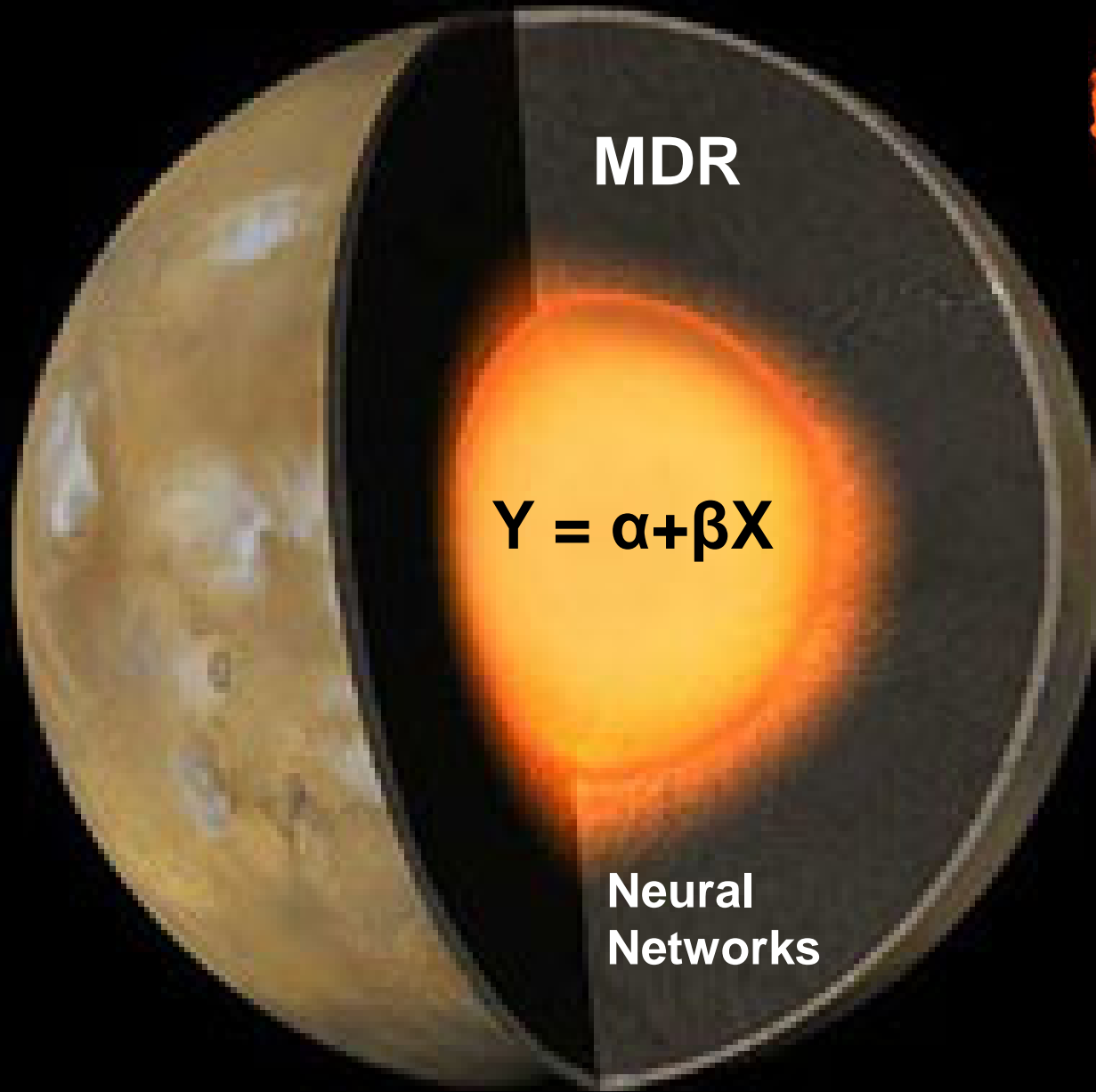
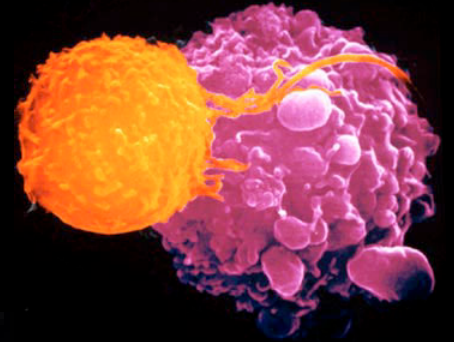
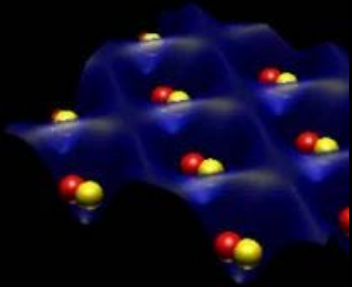


80%

Epistasis
Gene-Environment Interaction
Epigenetics
Heterogeneity

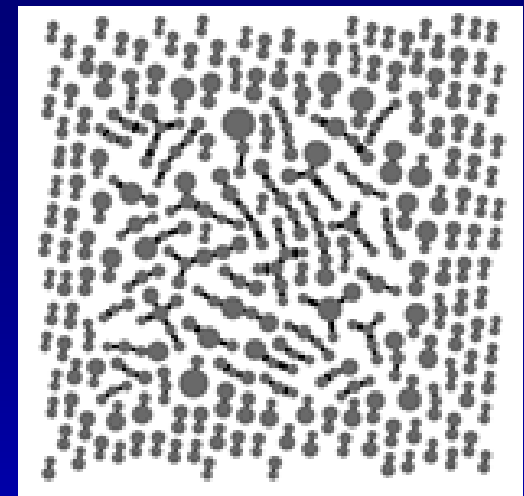
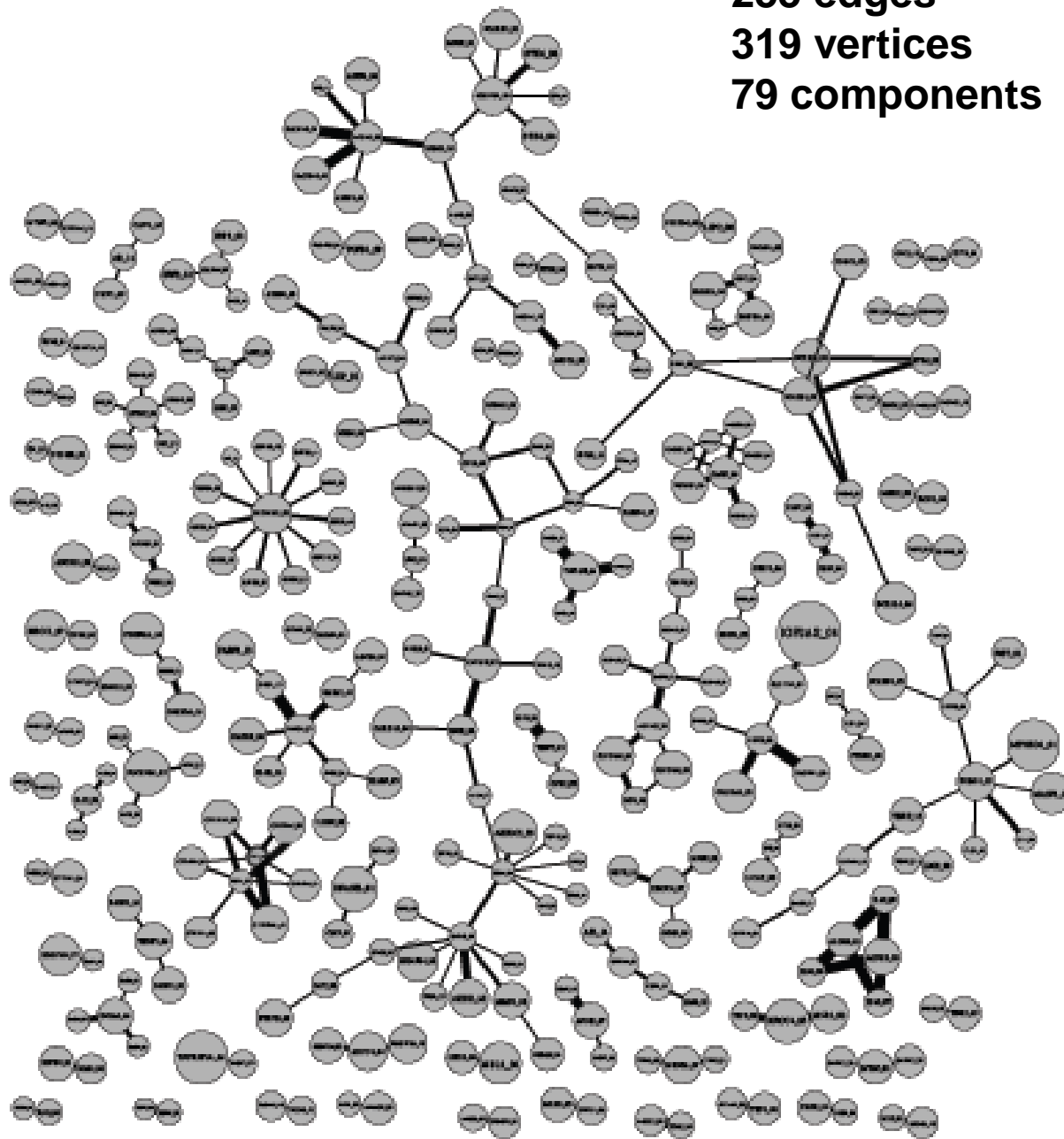
“Multivariate bioinformatics approach”

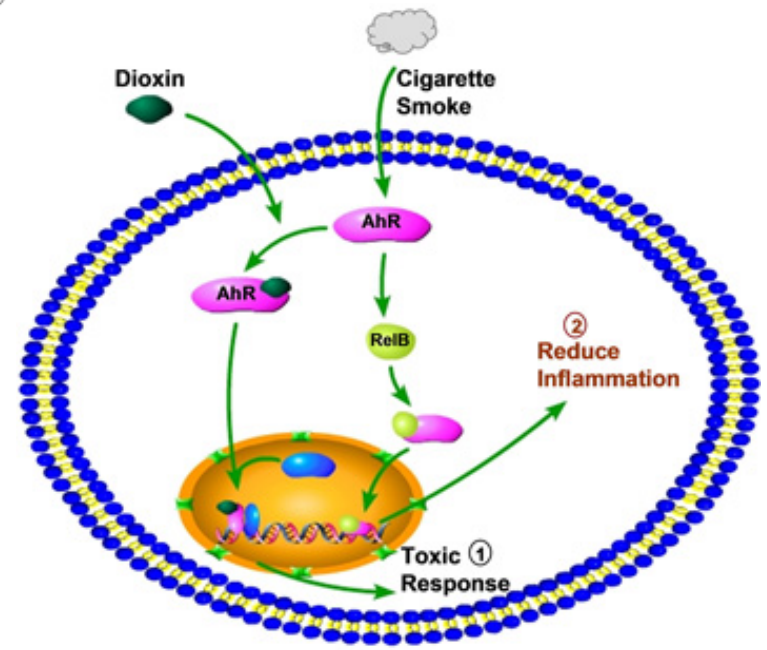
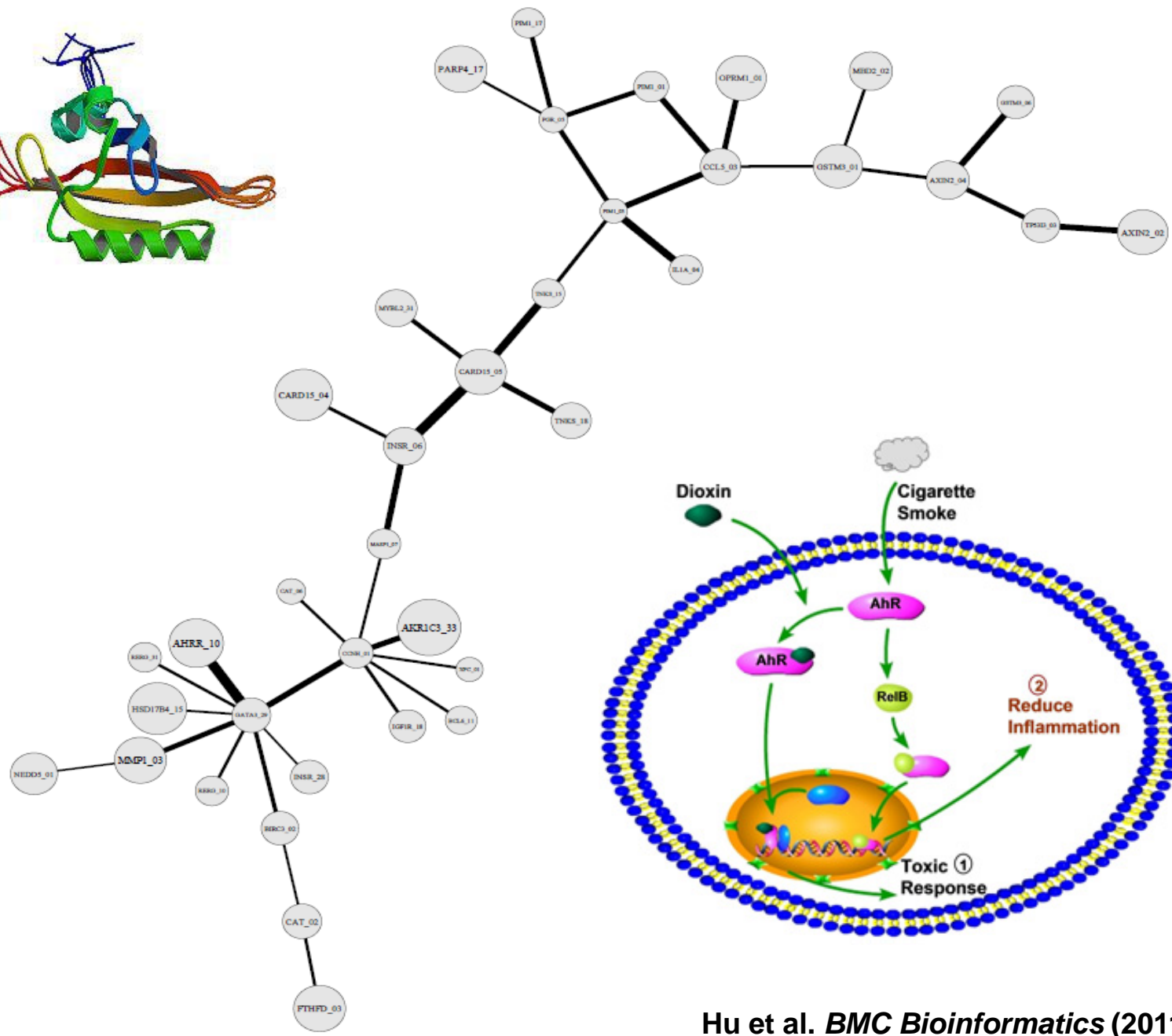
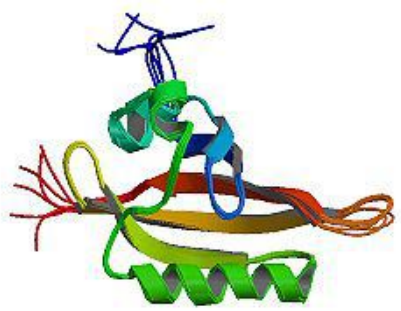




Epistasis Networks

255 edges
319 vertices
79 components

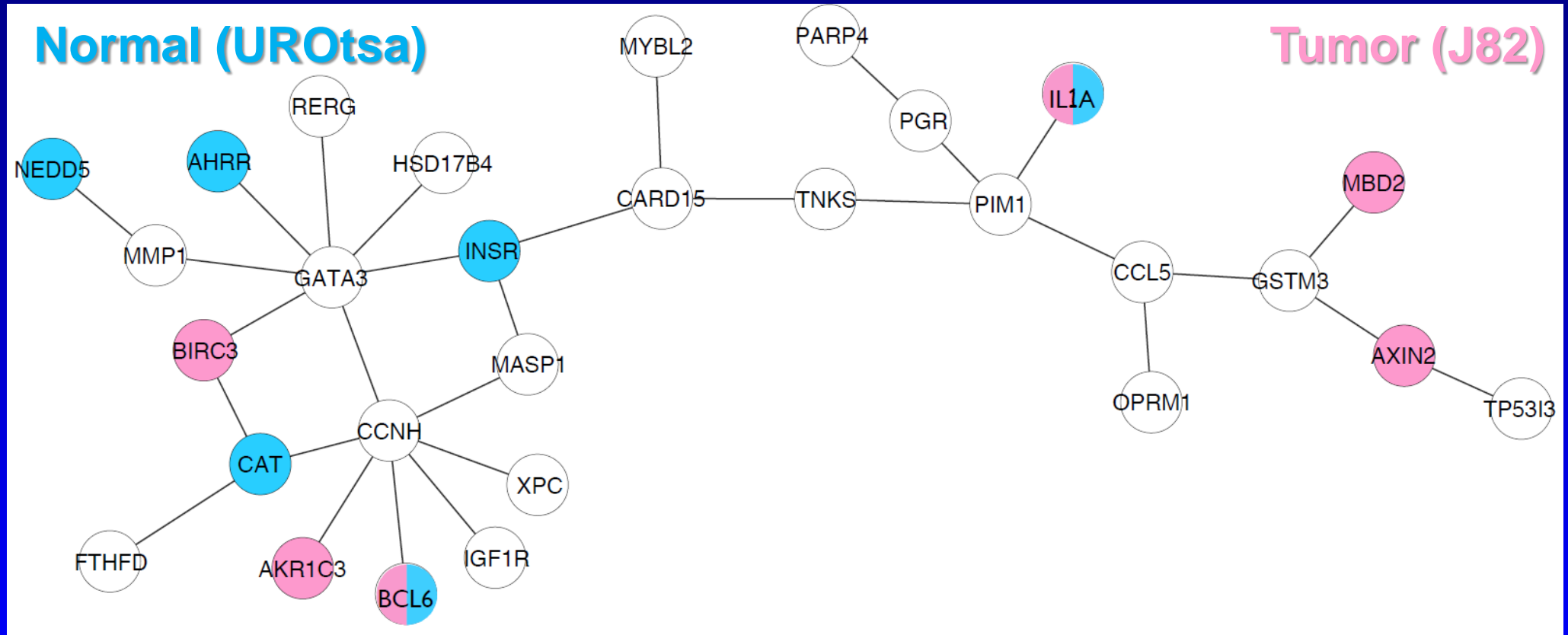




Hu et al. *BMC Bioinformatics* (2011)

Functional Genomics Annotation of a Statistical Epistasis Network

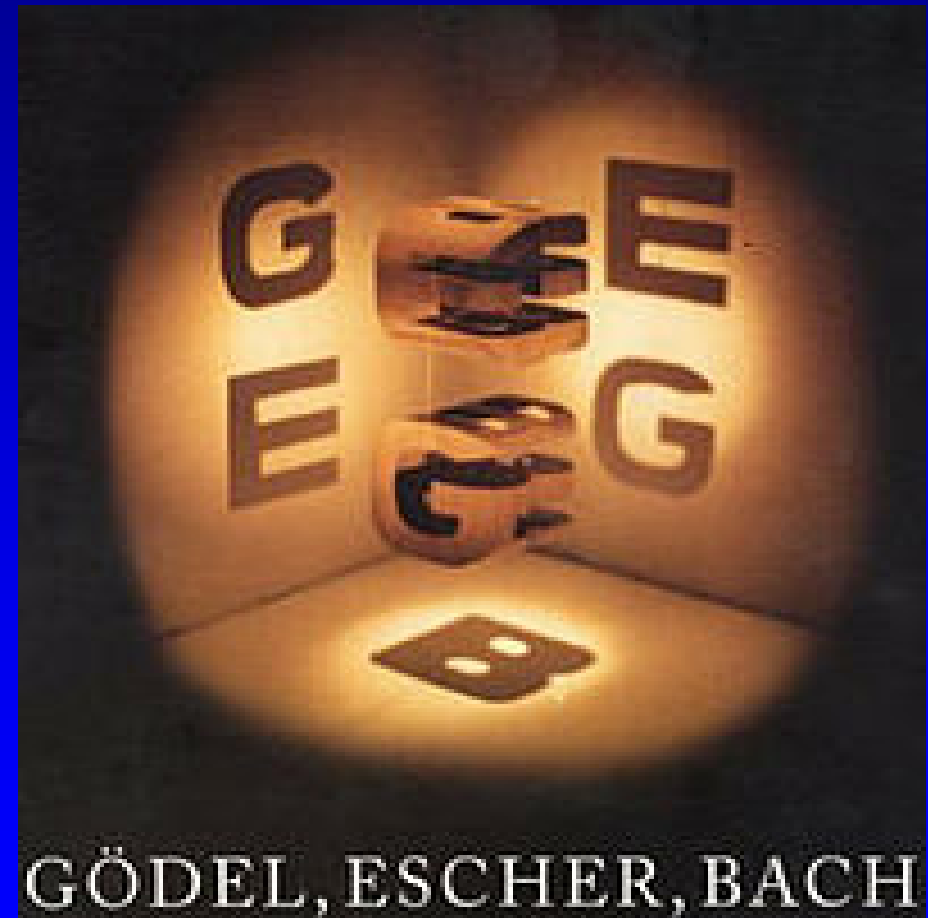
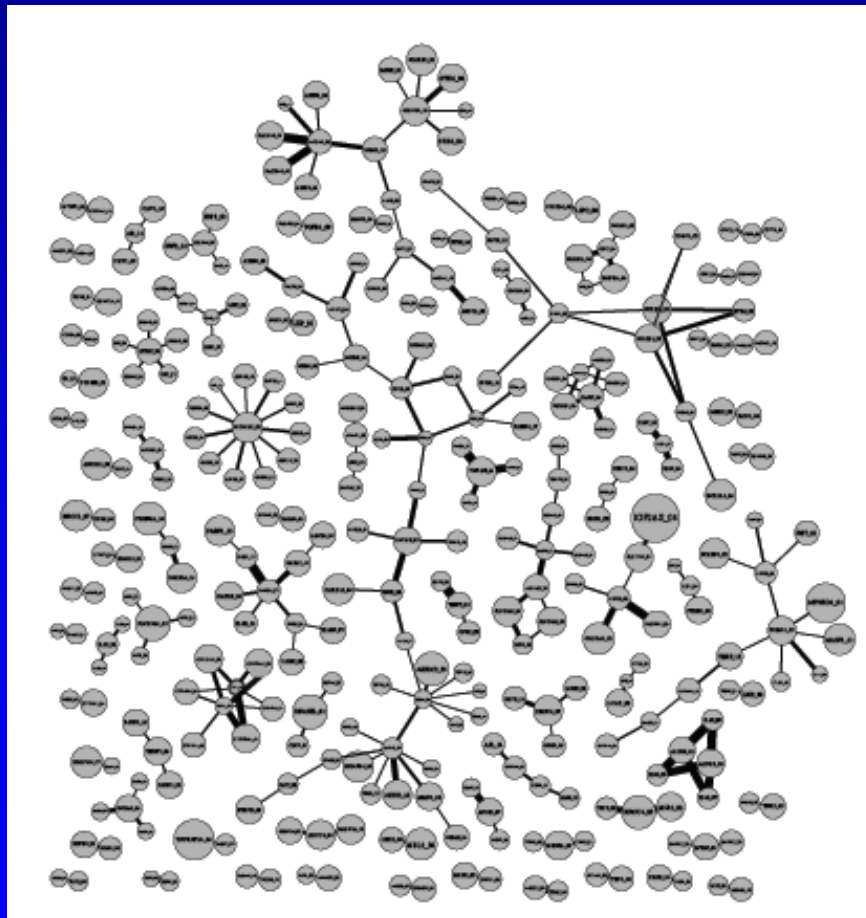
Hu et al., *BioData Mining* (2014)



Shadows of complexity: what biological networks reveal about epistasis and pleiotropy

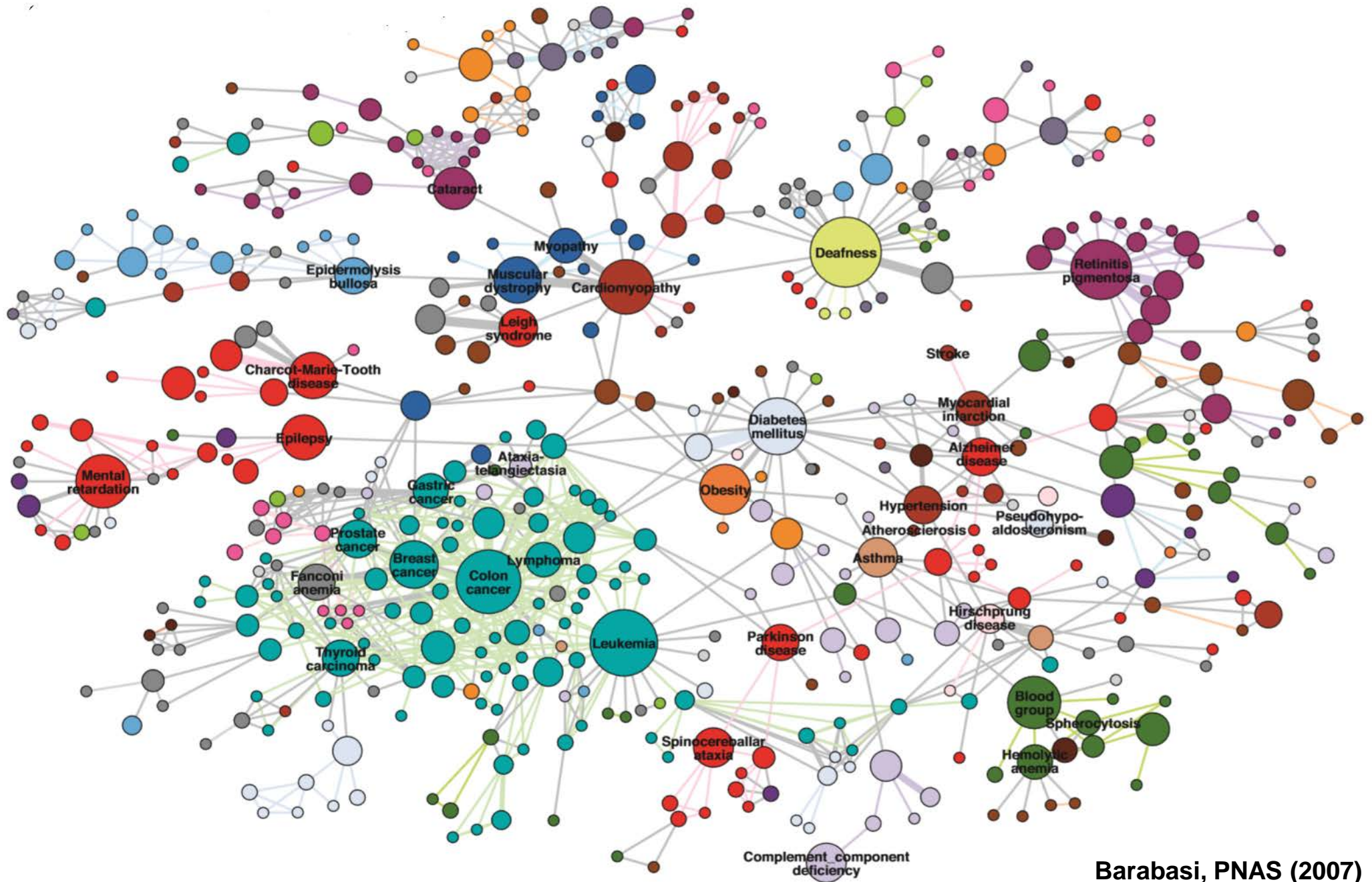
Anna L. Tyler,¹ Folkert W. Asselbergs,² Scott M. Williams,³ and Jason H. Moore Ph.D.^{1,4,5,6,7*}

BioEssays 31:220-227, 2009



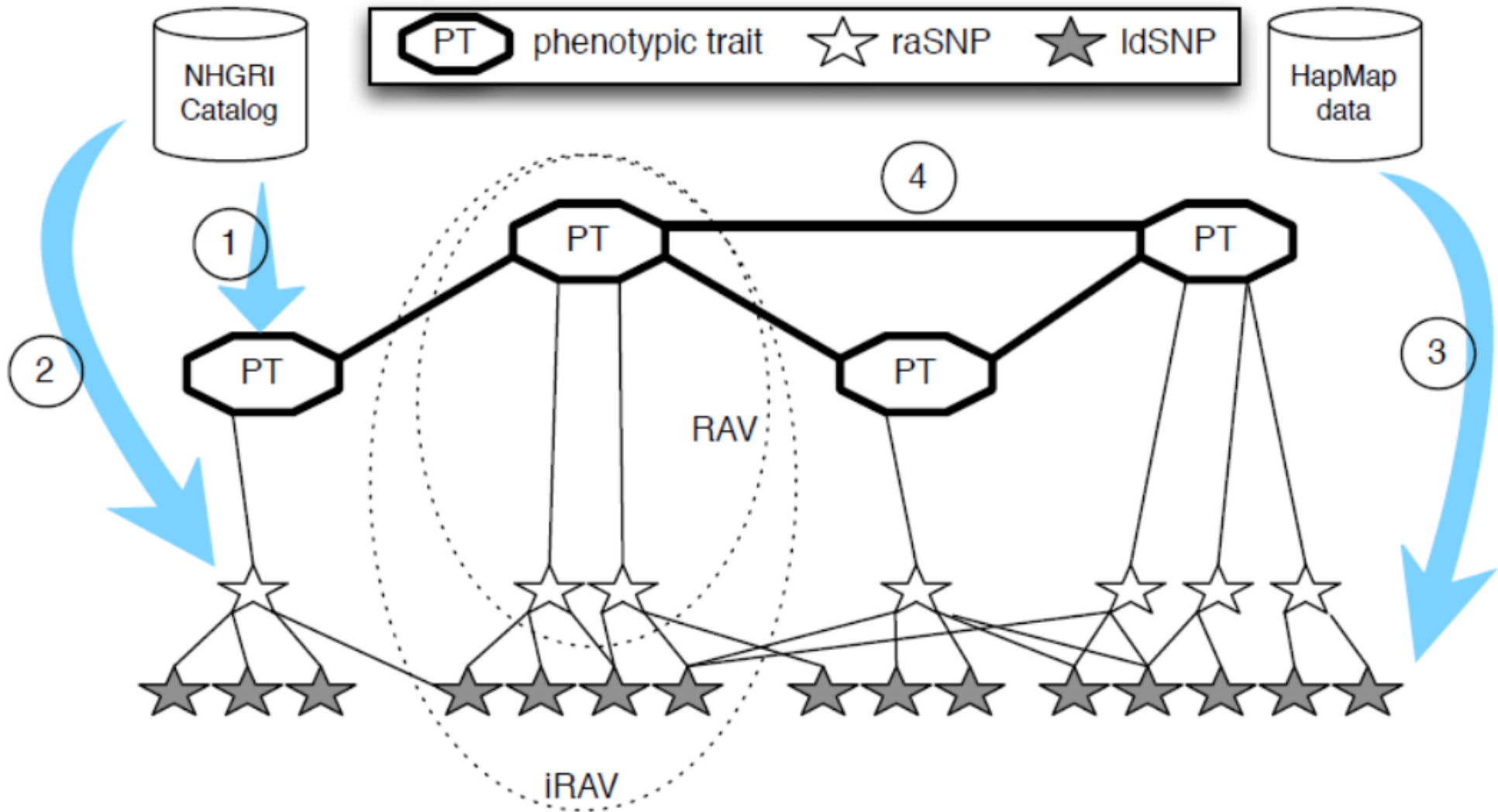
GÖDEL, ESCHER, BACH

Phenotype Networks



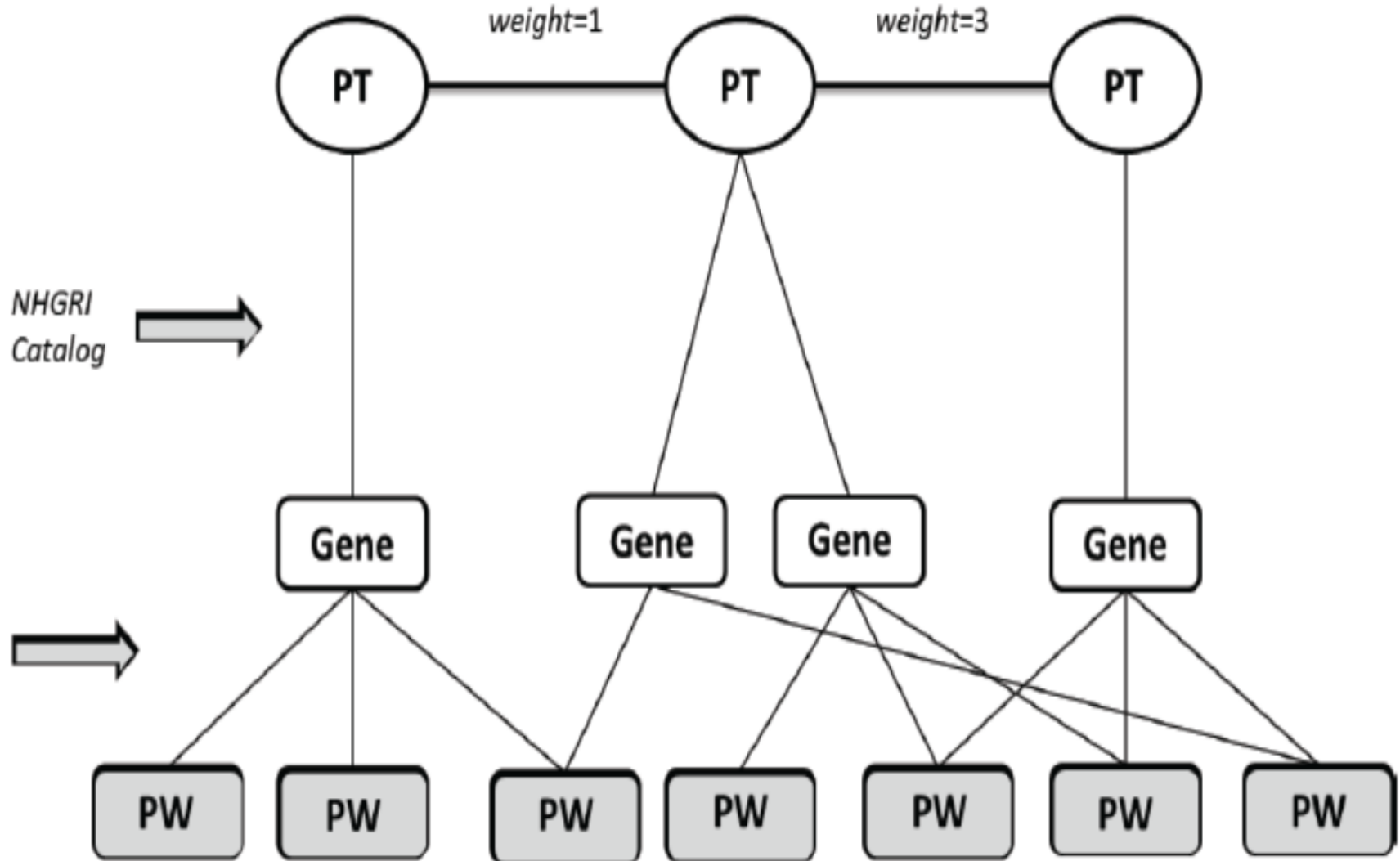
Human Phenotype Networks

Darabos et al., *Lecture Notes in Computer Science* (2013)



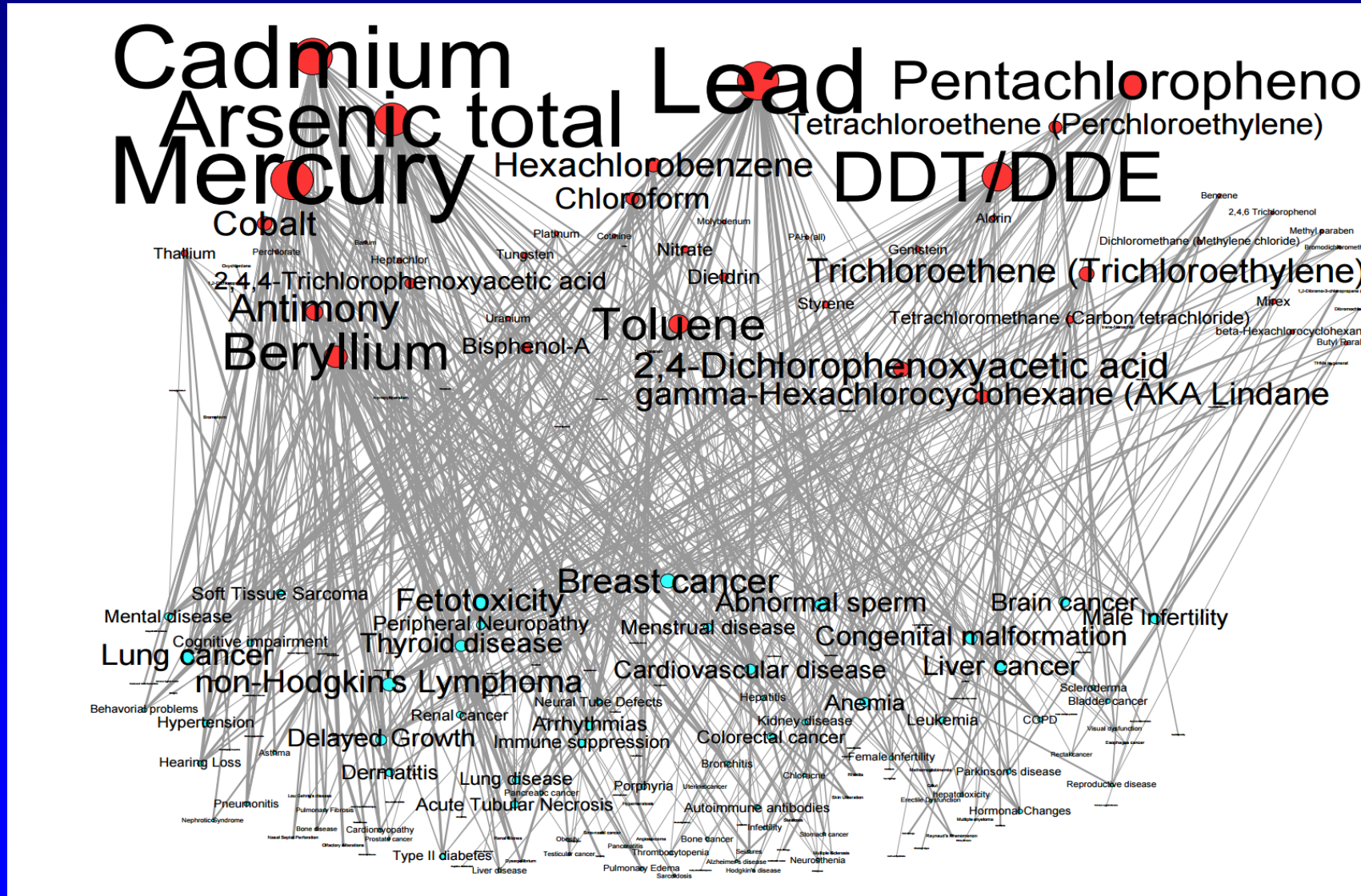
Human Phenotype Networks

Darabos et al., *BioData Mining* (2014)



Exposure-Based Human Phenotype Networks

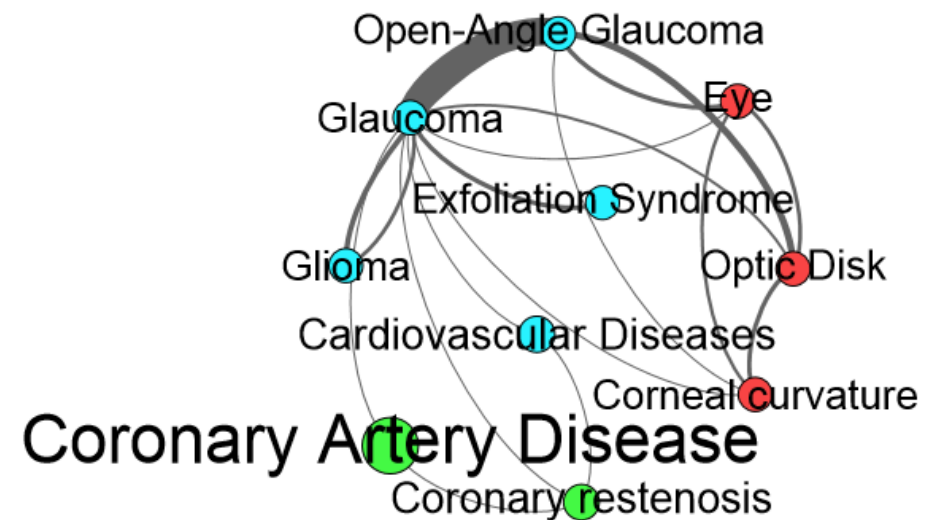
Darabos et al., *Pacific Symposium on Biocomputing* (2015)

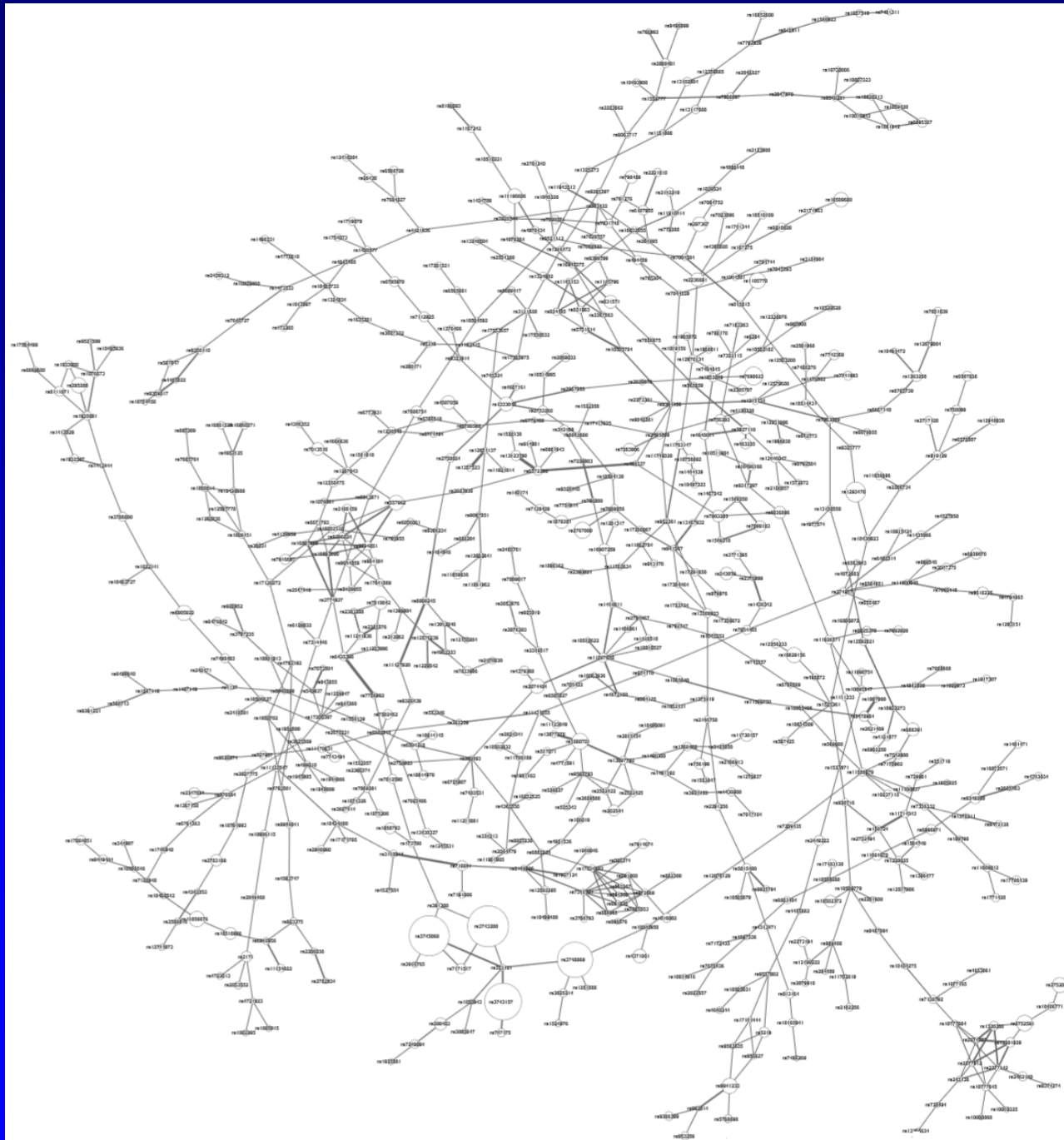


Genome-Wide Genetic Interaction Analysis of Glaucoma using Expert Knowledge Derived from Human Phenotype Networks

Hu et al., *Pacific Symposium on Biocomputing* (2015)

Phenotype	#SNPs	Degree
Exfoliation Syndrome	1	1
Coronary Artery Disease	639	2
Cardiovascular Diseases	70	2
Corneal curvature	13	4
Optic Disk	17	4
Open-Angle Glaucoma	6	5
Glioma	12	2
Eye	13	4
Glaucoma	18	9
Coronary restenosis	56	3





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