Genome-Wide Association Studies and Clinical Applications

U.S. Department of Health and Human Services
National Institutes of Health
National Human Genome Research Institute

Teri A. Manolio, M.D., Ph.D.
Director, Office of Population Genomics
Senior Advisor to the Director, NHGRI,
for Population Genomics
March 8, 2011

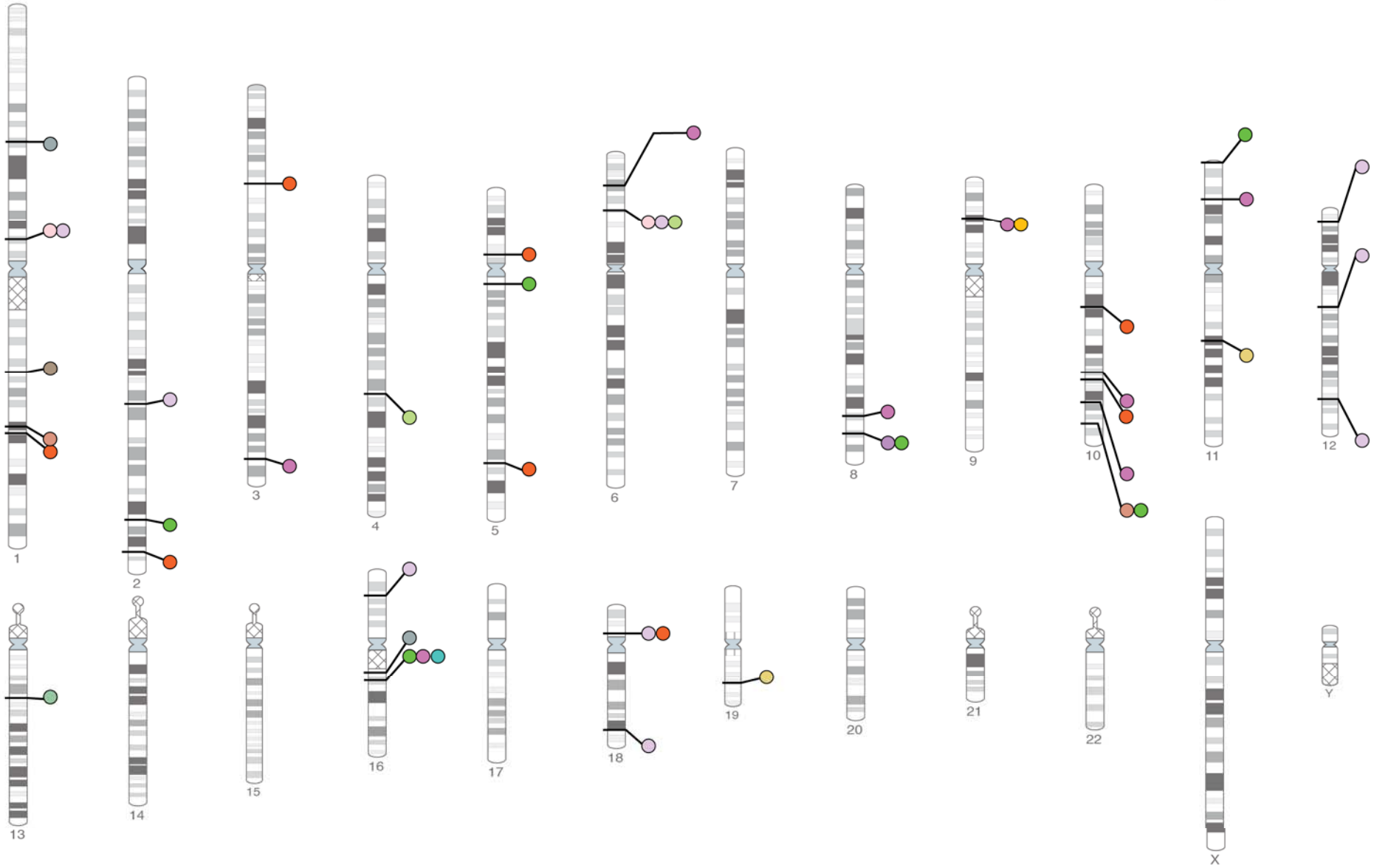
What do GWA Studies Have To Do with Direct to Consumer Testing?

- Among first studies identifying several easily measured variants associated with risk of common, complex diseases
  - Common, not Mendelian
  - Followed long barren period
  - This actually works!
- Results more rigorously validated than some prior genetic findings
- Findings more rapidly incorporated into multi-gene or genome-wide panels
- Accelerating pace of discovery

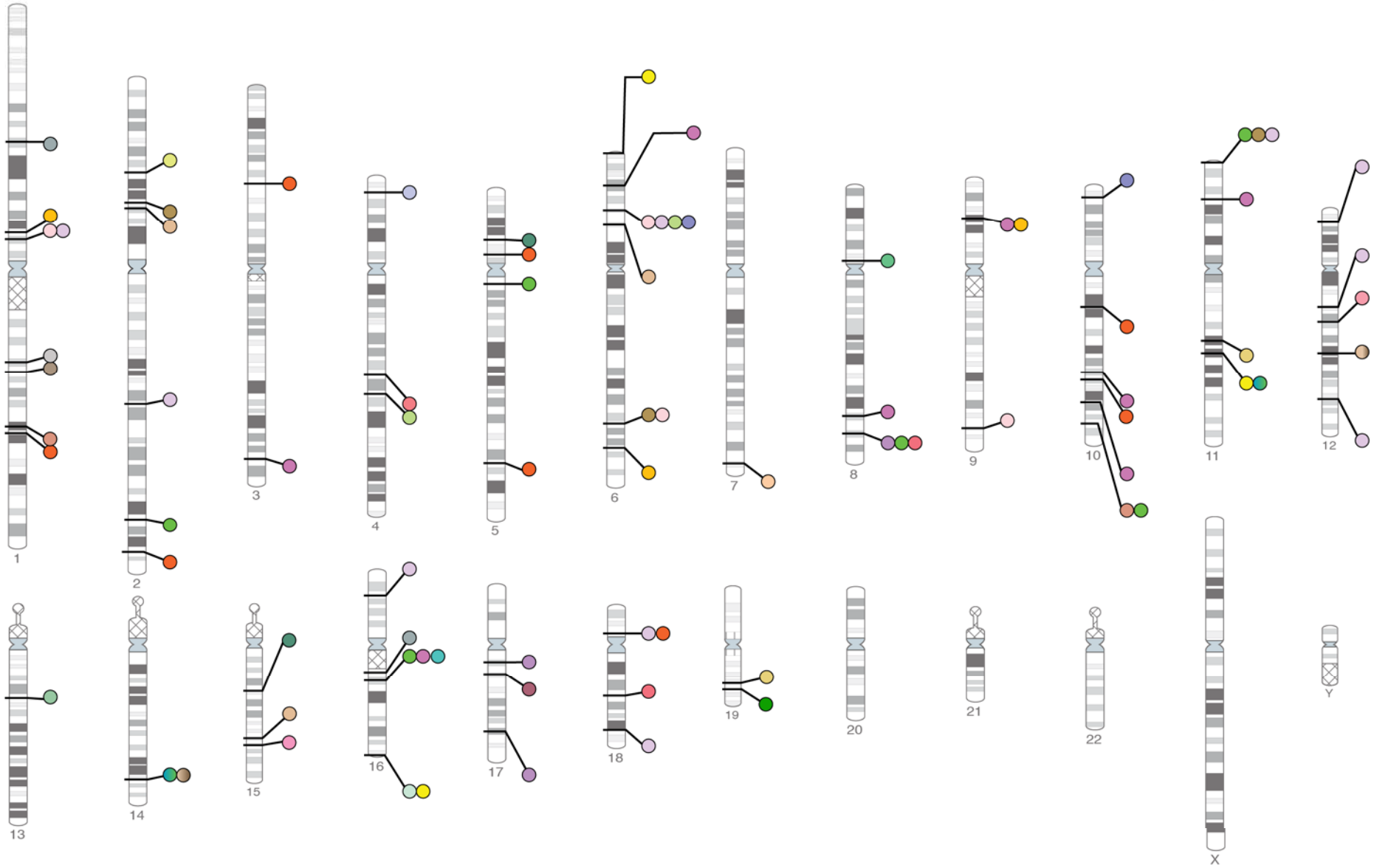
2005

2006

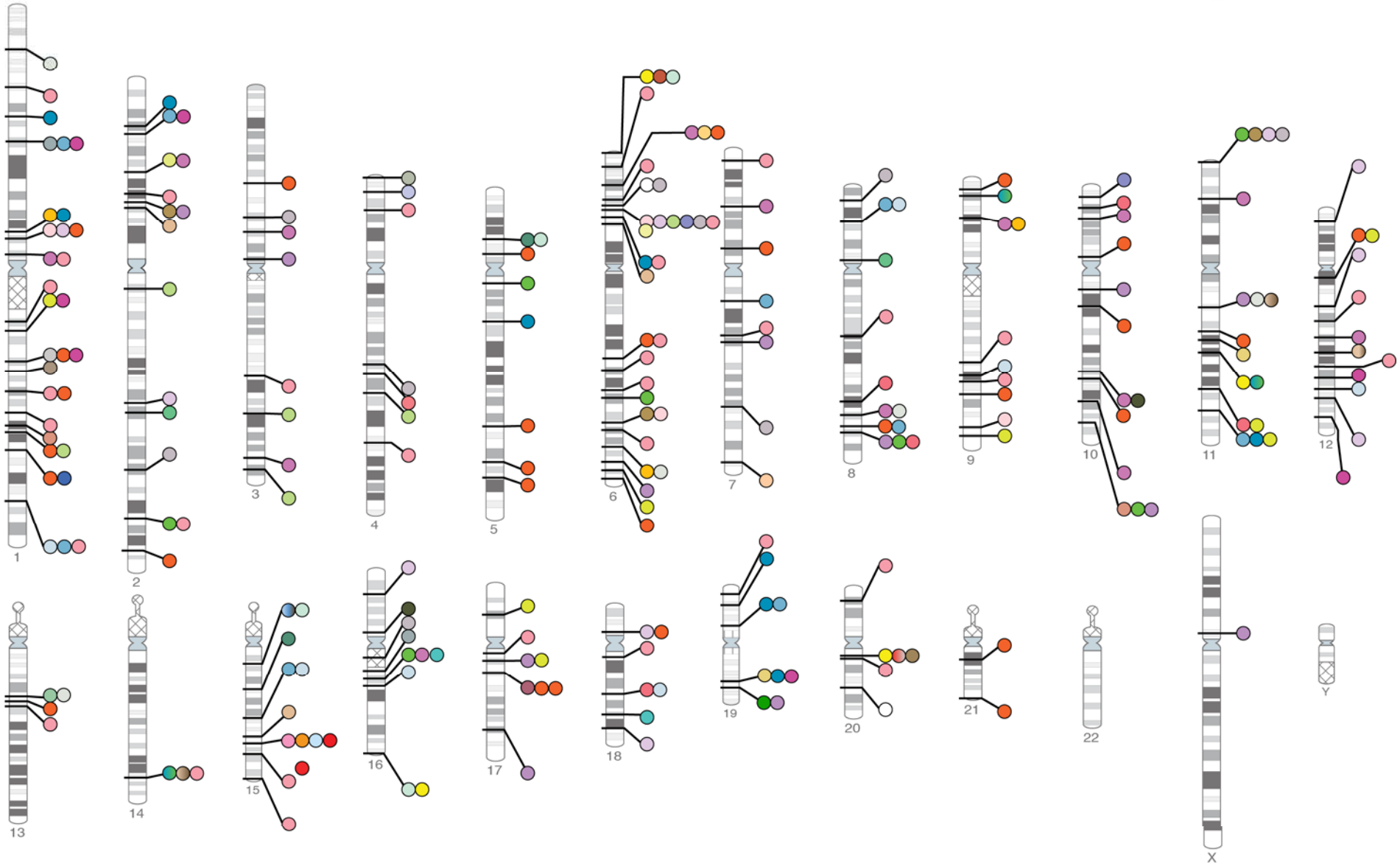
2007 2nd quarter



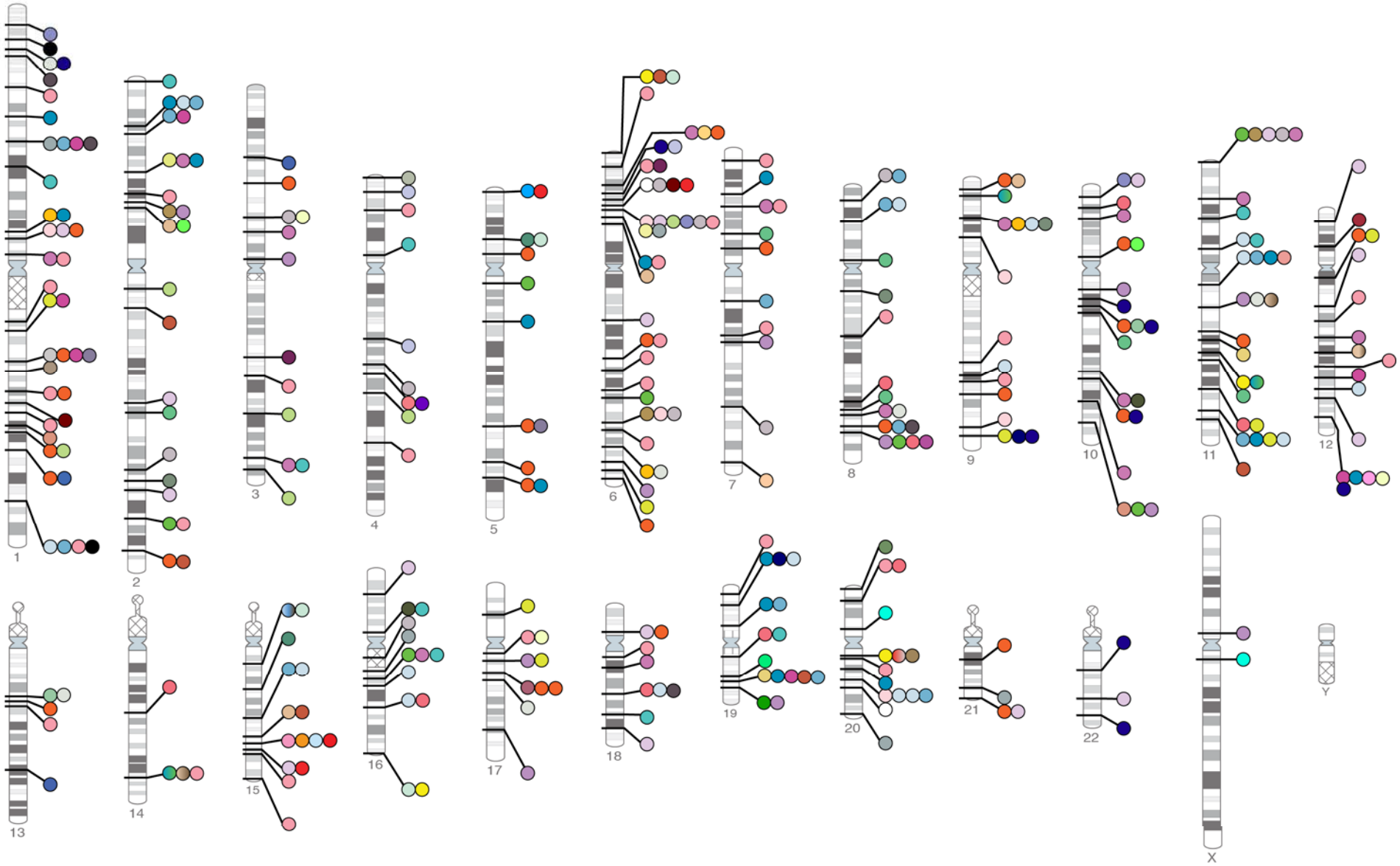
2007 4th quarter



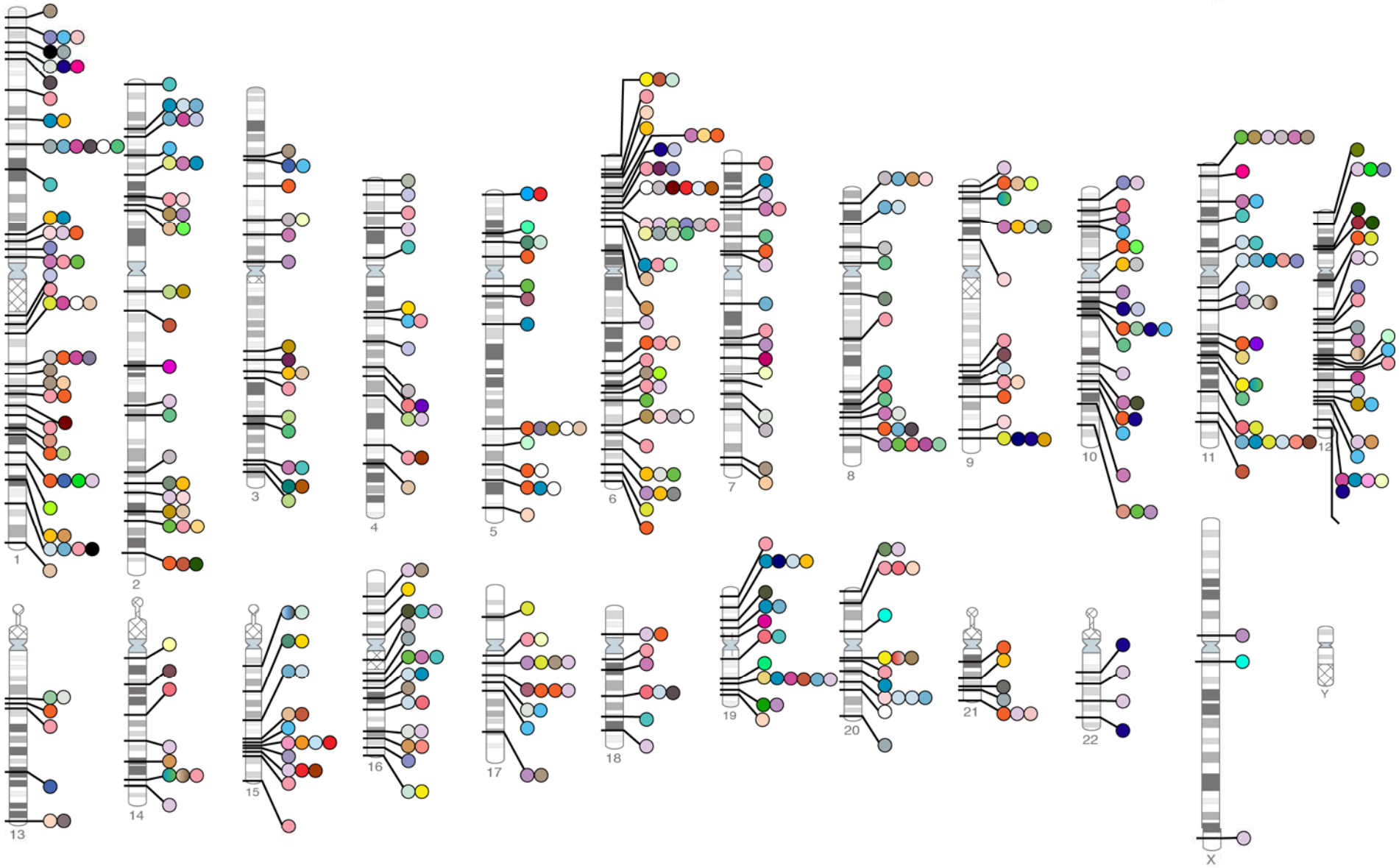
2008 2nd quarter



2008 4th quarter



2009 2nd quarter



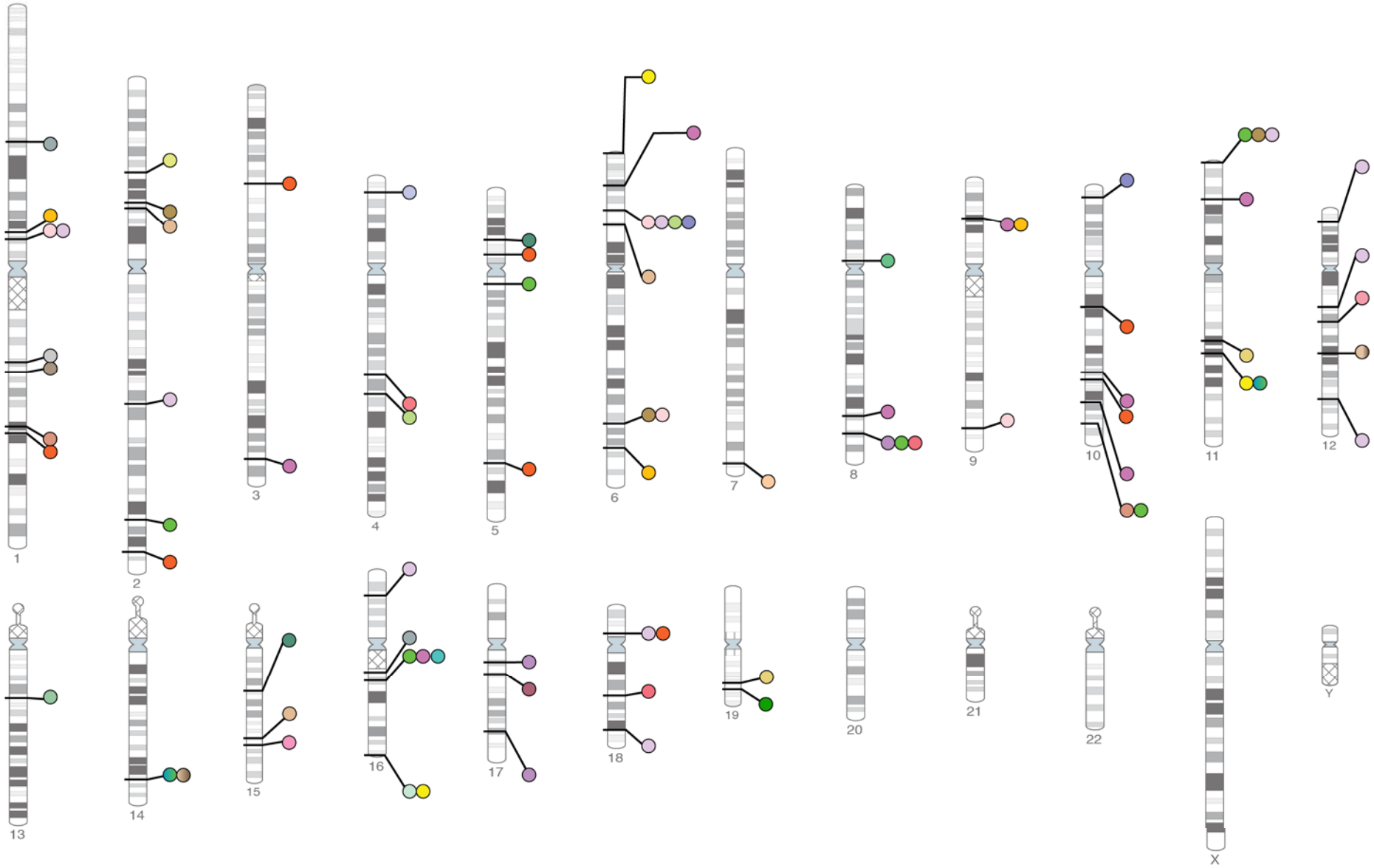
2009 4th quarter

904 Associations at $p < 5 \times 10^{-8}$

2010 2nd quarter

NHGRI GWA Catalog
www.genome.gov/GWAStudies

2007 4th quarter



deCODE ME

deCODE genetics
the pioneers in gene discovery

23andMe genetics just got personal.

sign in
have a claim

Navigenics

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Your genes offer a road map to optimal health

The Navigenics advantage

We're in the midst of the genomics revolution, discovering new connections between your DNA, your environment and your personal health and wellness. You can feel confident choosing Navigenics to help you harness this information, because:

- Our team of leading scientific and clinical advisers provides expert review of our service.
- Our lab is certified under CLIA, the law covering accuracy and timeliness of test results.
- Our genetic counselors, available by phone help you interpret your results.

What is a Genome-Wide Association Study?

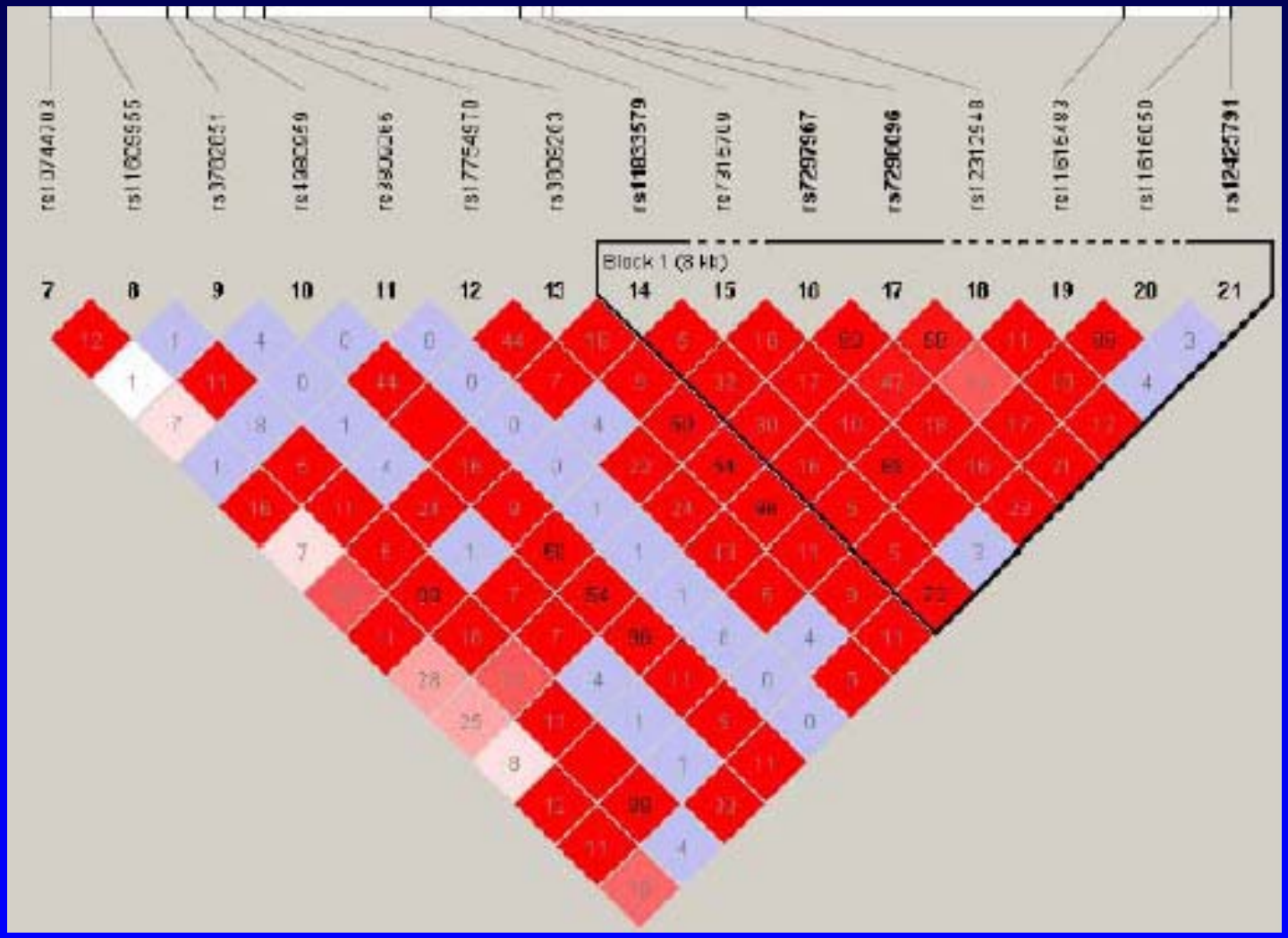
- Method for interrogating all 10 million common (MAF > 5%) variable points in human genome
- Variation inherited in groups, or blocks, so not all 10 million points have to be tested
- Blocks are shorter (so need to test more points) the less closely people are related
- Allows studies in unrelated persons, assuming 5,000 – 10,000 base pair lengths in common (500,000 – 1,000,000 markers @ $450)

DNA on Chromosome 7

GAAATAATTAATGTTTTCCTTCCTTCTCCTATTTTGTCCTTTACTTCAATTTATTTATTTATTATTAATATTATTATTTTTTG
AGACGGAGTTTC/ACTCTTGTTGCCAACCTGGAGTGCAGTGGCGTGATCTCAGCTCACTGCACACTCCGCTTTCCTG
GTTTCAAGCGATTCTCCTGCCTCAGCCTCCTGAGTAGCTGGGACTACAGTCACACACCACCACGCCCGGCTAATTTTT
GTATTTTTAGTAGAGTTGGGGTTTCACCATGTTGGCCAGACTGGTCTCGAACTCCTGACCTTGTGATCCGCCAGCCTC
TGCCTCCCAAAGAGCTGGGATTACAGGCCTGAGCCACCGCGCTCGGCCCTTTGCATCAATTTCTACAGCTTGTTTTCT
TTGCCTGGACTTTACAAGTCTTACCTTGTTCTGCC/TTCAGATATTTGTGTGGTCTCATTCTGGTGTGCCAGTAGCTAA
AAATCCATGATTTGCTCTCATCCCACTCCTGTTGTTCATCTCCTCTTATCTGCGGTCAC/ATATCTCTTCGTGATTGC
ATTCTGATCCCCAGTACTTAGCATGTGCGTAACAACTCTGCCTCTGCTTTCCCAGGCTGTTGATGGGGTGCTGTTCAT
GCCTCAGAAAAATGCATTGTAAGTTAAATTATTAAAGATTTTAAATATAGGAAAAAAGTAAGCAAACATAAGGAACAA
AAAGGAAAGAACATGTATTCTAATCCATTATTTATTATACAATTAAGAAATTTGGAAACTTTAGATTACACTGCTTTTA
GAGATGGAGATGTAGTAAGTCTTTTACTCTTTACAAAATACATGTGTTAGCAATTTTGGGAAGAATAGTAACTCACCC
GAACAGTG/TAATGTGAATATGTCACTTACTAGAGGAAAGAAGGCACTTGAAAAACATCTCTAAACCGTATAAAAAC
AATTACATCATAATGATGAAAACCCAAGGAATTTTTTTAGAAAACATTACCAGGGCTAATAACAAAGTAGAGCCACAT
GTCATTTATCTTCCCTTTGTGTCTGTGTGAGAATTCTAGAGTTATATTTGTACATAGCATGGAAAAATGAGAGGCTAGT
TTATCAACTAGTTCATTTTTAAAAGTCTAACACATCCTAGGTATAGGTGAACTGTCCTCCTGCCAATGTATTGCACATT
TGTGCCCAGATCCAGCATAGCCTATGTTTGCCATTTACAAACGTTTATGTCTTAAGAGAGGAAATATGAAGAGCAAAA
CAGTGCATGCTGGAGAGAGAAAGCTGATACAAATATAAAT/GAAACAATAATTGGAAAAATTGAGAAACTACTCATT
TTCTAAATTACTCATGTATTTTCCTAGAATTTAAGTCTTTTAATTTTTGATAAATCCCAATGTGAGACAAGATAAGTATT
AGTGATGGTATGAGTAATTAATATCTGTTATATAATATTCATTTTCATAGTGGAAGAAATAAAATAAAGGTTGTGATGA
TTGTTGATTATTTTTTCTAGAGGGGTTGTCAGGGAAAGAAATTGCTTTTT

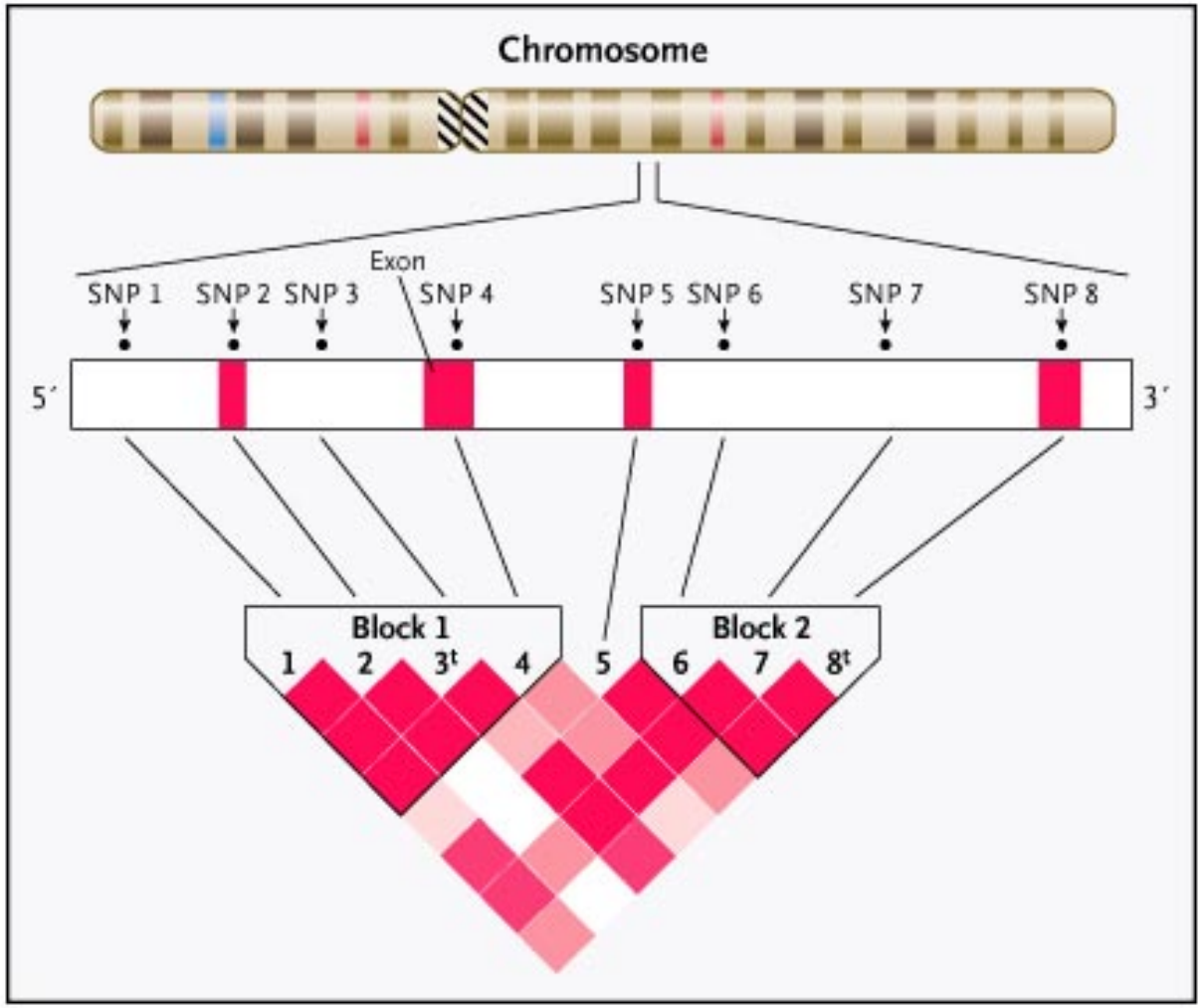
SNPs 1 / 300 bases

Linkage Disequilibrium Plot of *NINJ2* Region Associated with Total and Ischemic Stroke



Ikram MA et al., *N Engl J Med* 2009; 360:1718-28.

Mapping the Relationships Among SNPs



Christensen and Murray, *N Engl J Med* 2007; 356:1094-97.

Distances Among East Coast Cities

	Boston	Provi-dence	New York	Phila-delphia	Balti-more
Providence	59				
New York	210	152			
Philadelphia	320	237	86		
Baltimore	430	325	173	87	
Washington	450	358	206	120	34

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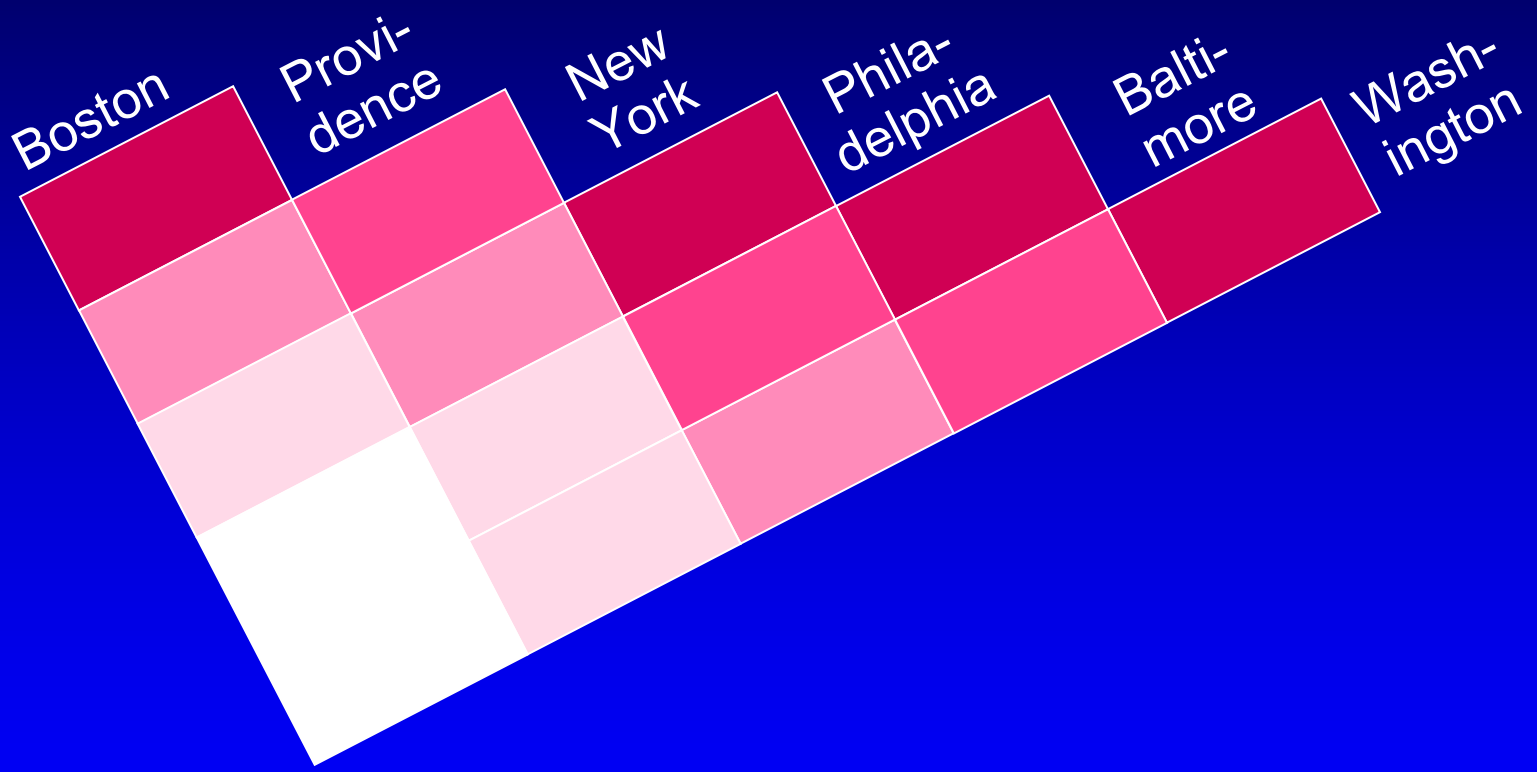

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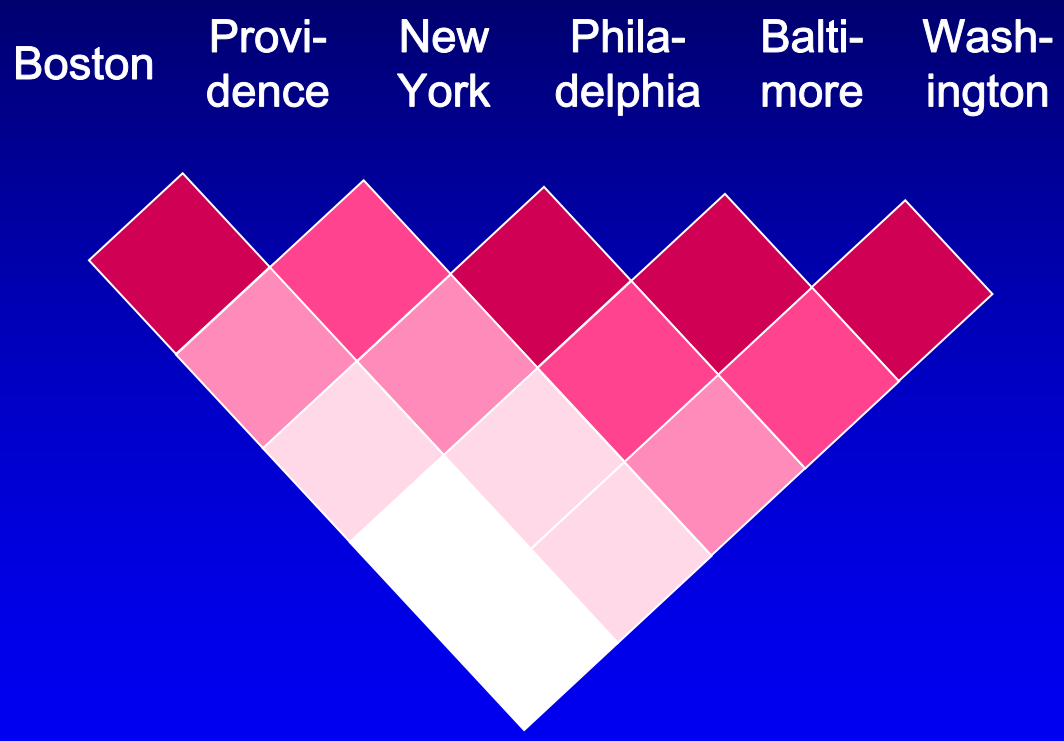
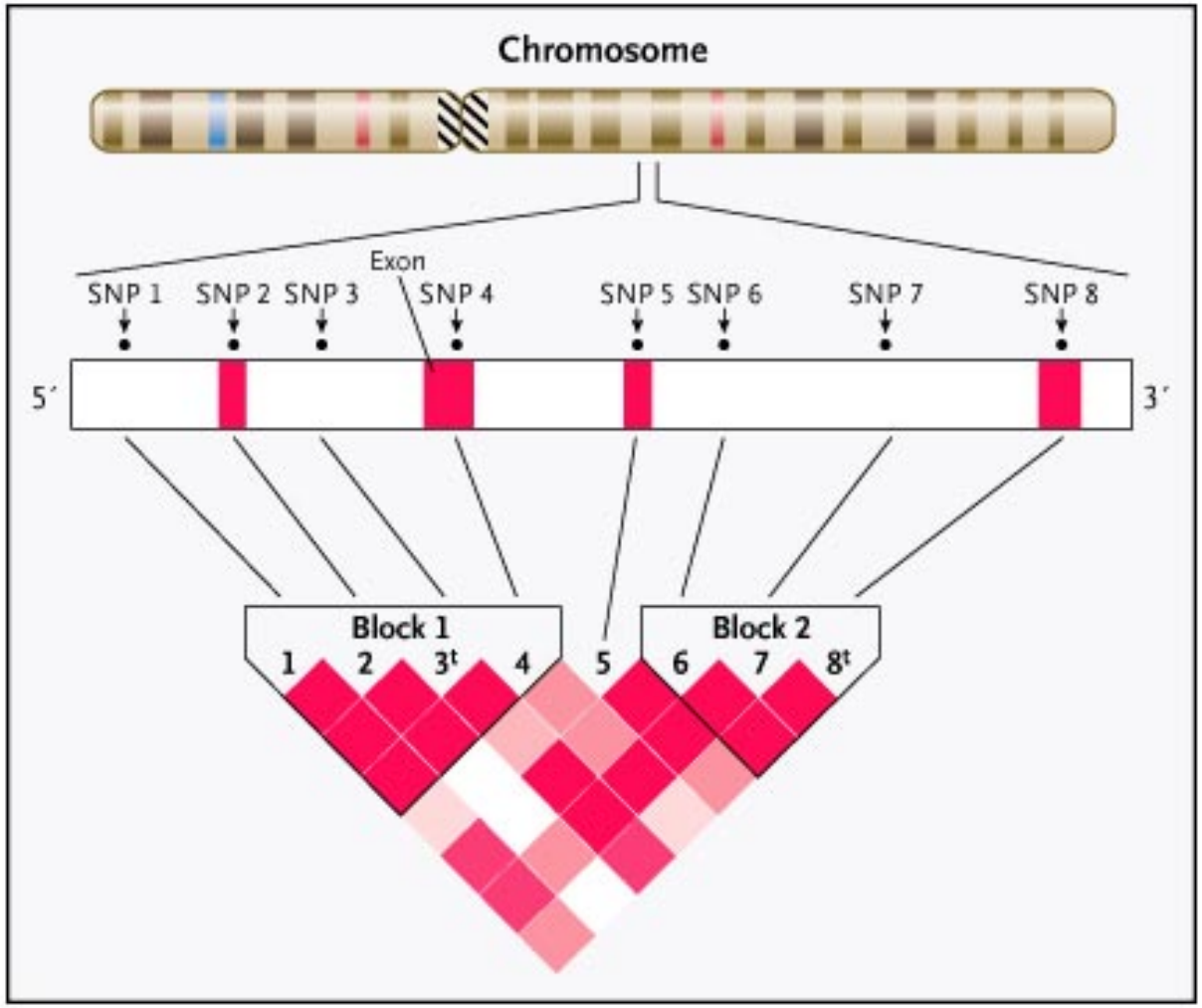


Mapping the Relationships Among SNPs



Christensen and Murray, *N Engl J Med* 2007; 356:1094-97.

One Tag SNP May Serve as Proxy for Many

Block 1 Block 2

SNP1 SNP2 SNP3 SNP4 SNP5 SNP6 SNP7 SNP8

↓ ↓ ↓ ↓ ↓ ↓ ↓ ↓

CAGATCGCTGGATGAATCGCATCTGTAAGCAT

CGGATTGCTGCATGGATCGCATCTGTAAGCAC

CAGATCGCTGGATGAATCGCATCTGTAAGCAT

CAGATCGCTGGATGAATCCCATCAGTACGCAT

CGGATTGCTGCATGGATCCCATCAGTACGCAT

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Block 1 Block 2

SNP1	SNP2	SNP3	SNP4	SNP5	SNP6	SNP7	SNP8
↓	↓	↓	↓	↓	↓	↓	↓
A	C	G	A	G	T	A	T
G	T	C	G	G	T	A	C
A	C	G	A	G	T	A	T
A	C	G	A	C	A	C	T
G	T	C	G	C	A	C	T
G	T	C	G	C	A	C	C

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C	G	T	A	C
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G	C	A	C	T
C	C	A	C	T
C	C	A	C	C

One Tag SNP May Serve as Proxy for Many

Block 1 | Block 2

SNP3	SNP6	SNP8
↓	↓	↓
G	T	T
C	T	C
G	T	T
G	A	T
C	A	T
C	A	C

One Tag SNP May Serve as Proxy for Many

Block 1	Block 2	Singleton	Frequency
G	T	T	35%
C	T	C	30%
G	T	T	10%
G	A	T	8%
C	A	T	7%
C	A	C	6%
other haplotypes			4%

International HapMap Project

www.hapm

1000 Genomes
A Deep Catalog of Human Genetic Variation

www.1000genomes.org

Vol 437|27 October 2005|doi:10.1038/nature04226 nature

Vol 449|18 October 2007|doi:10.1038/nature06258 nature

nature Vol 467|2 September 2010|doi:10.1038/nature09298

ARTICLE

Nature 2010; 467:1061-73.

doi:10.1038/nature09534

A map of human genome variation from population-scale sequencing

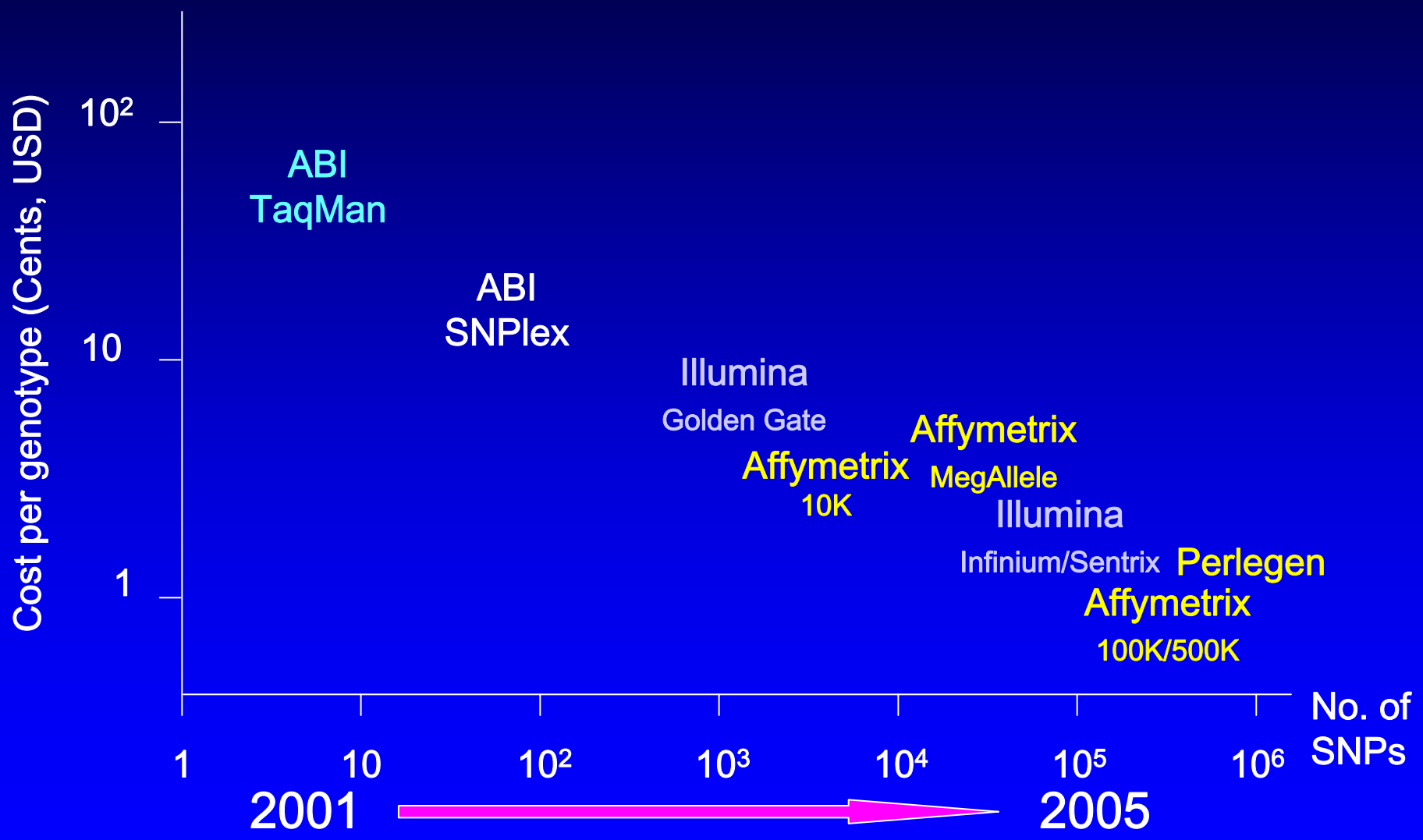
The 1000 Genomes Project Consortium*

The 1000 Genomes Project aims to provide a deep characterization of human genome sequence variation as a foundation for investigating the relationship between genotype and phenotype. Here we present results of the pilot phase of the project, designed to develop and compare different strategies for genome-wide sequencing with high-throughput platforms. We undertook three projects: low-coverage whole-genome sequencing of 179 individuals from four populations; high-coverage sequencing of two mother-father-child trios; and exon-targeted sequencing of 697 individuals from seven populations. We describe the location, allele frequency and local haplotype structure of approximately 15 million single nucleotide polymorphisms, 1 million short insertions and deletions, and 20,000 structural variants, most of which were previously undescribed. We show that, because we have catalogued the vast majority of common variation, over 95% of the currently accessible variants found in any individual are present in this data set. On average, each person is found to carry approximately 250 to 300 loss-of-function variants in annotated genes and 50 to 100 variants previously implicated in inherited disorders. We demonstrate how these results can be used

A HapMap for More Efficient Association Studies: Goals

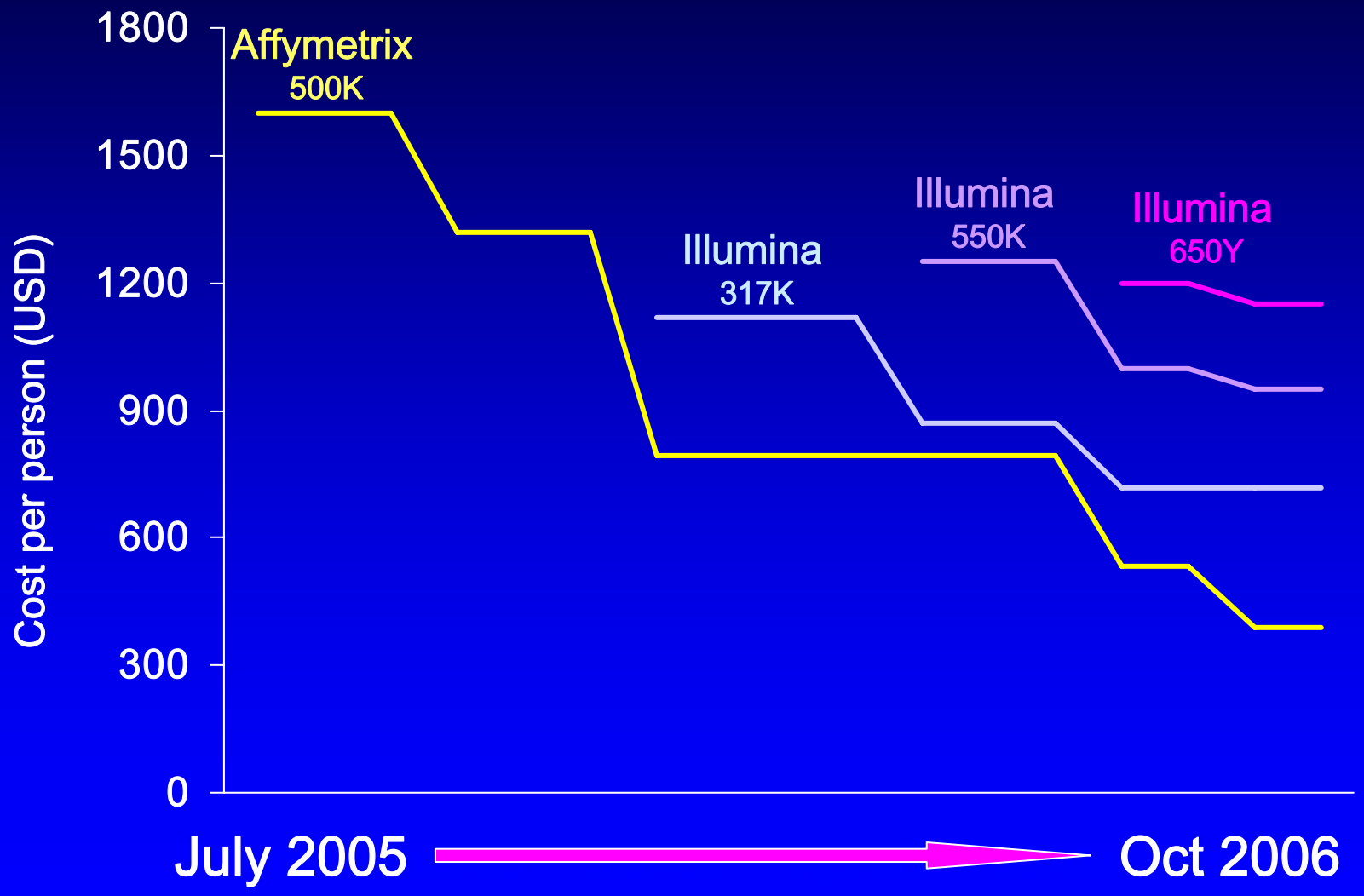
- Use just the density of SNPs needed to find associations between SNPs and diseases
- Do not miss chromosomal regions with disease association
- Produce a tool to assist in finding genes affecting health and disease
- Use more SNPs for complete genome coverage of populations of recent African ancestry populations due to shorter LD

Progress in Genotyping Technology



Courtesy S. Chanock, NCI

Continued Progress in Genotyping Technology



Courtesy S. Gabriel, Broad Institute

Association of Alleles and Genotypes of rs1333049 with Myocardial Infarction

	C N (%)	G N (%)	χ^2 (1df)	P-value
Cases	2,132 (55.4)	1,716 (44.6)	55.1	1.2×10^{-13}
Controls	2,783 (47.4)	3,089 (52.6)		
	Allelic Odds Ratio = 1.38			

Samani N et al, *N Engl J Med* 2007; 357:443-53.

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Controls	2,783 (47.4)	3,089 (52.6)		

Allelic Odds Ratio = 1.38

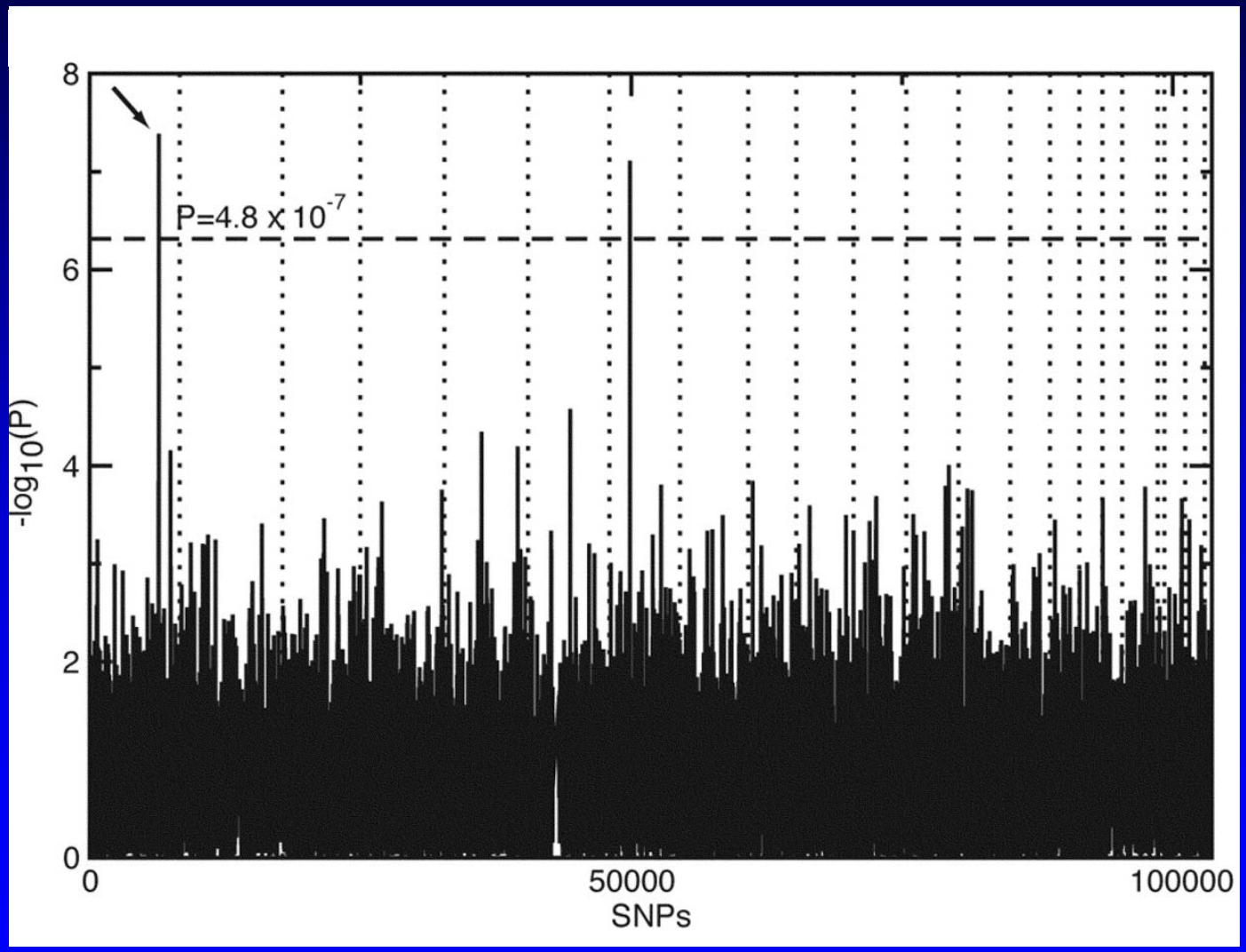
	CC N (%)	CG N (%)	GG N (%)	χ^2 (2df)	P-value
Cases	586 (30.5)	960 (49.9)	378 (19.6)	59.7	1.1×10^{-14}
Controls	676 (23.0)	1,431 (48.7)	829 (28.2)		

Heterozygote Odds Ratio = 1.47

Homozygote Odds Ratio = 1.90

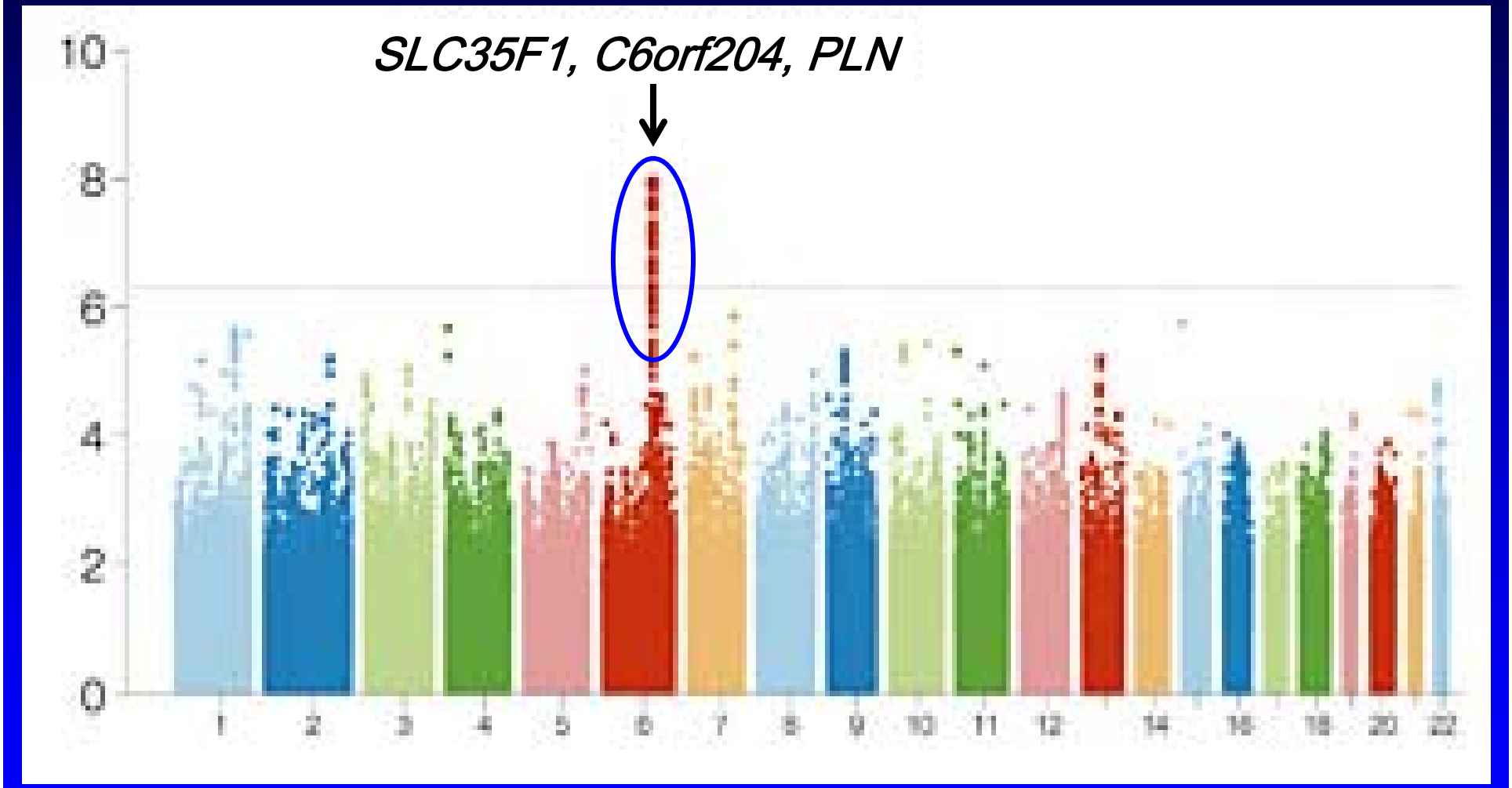
Samani N et al, *N Engl J Med* 2007; 357:443-53.

P Values of GWA Scan for Age-Related Macular Degeneration



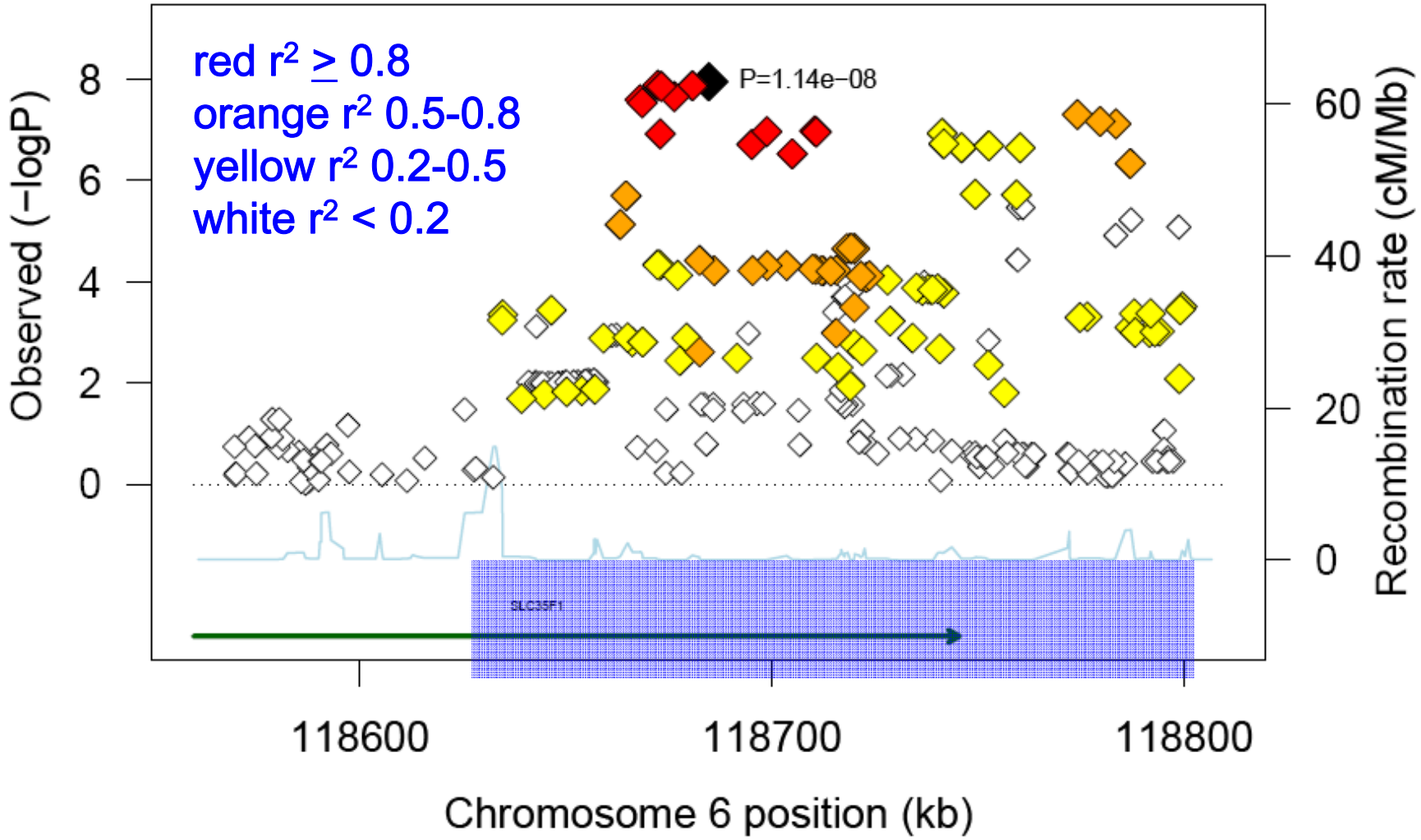
Klein et al, *Science* 2005; 308:385-389.

GWA Scan of LV Internal Dimensions



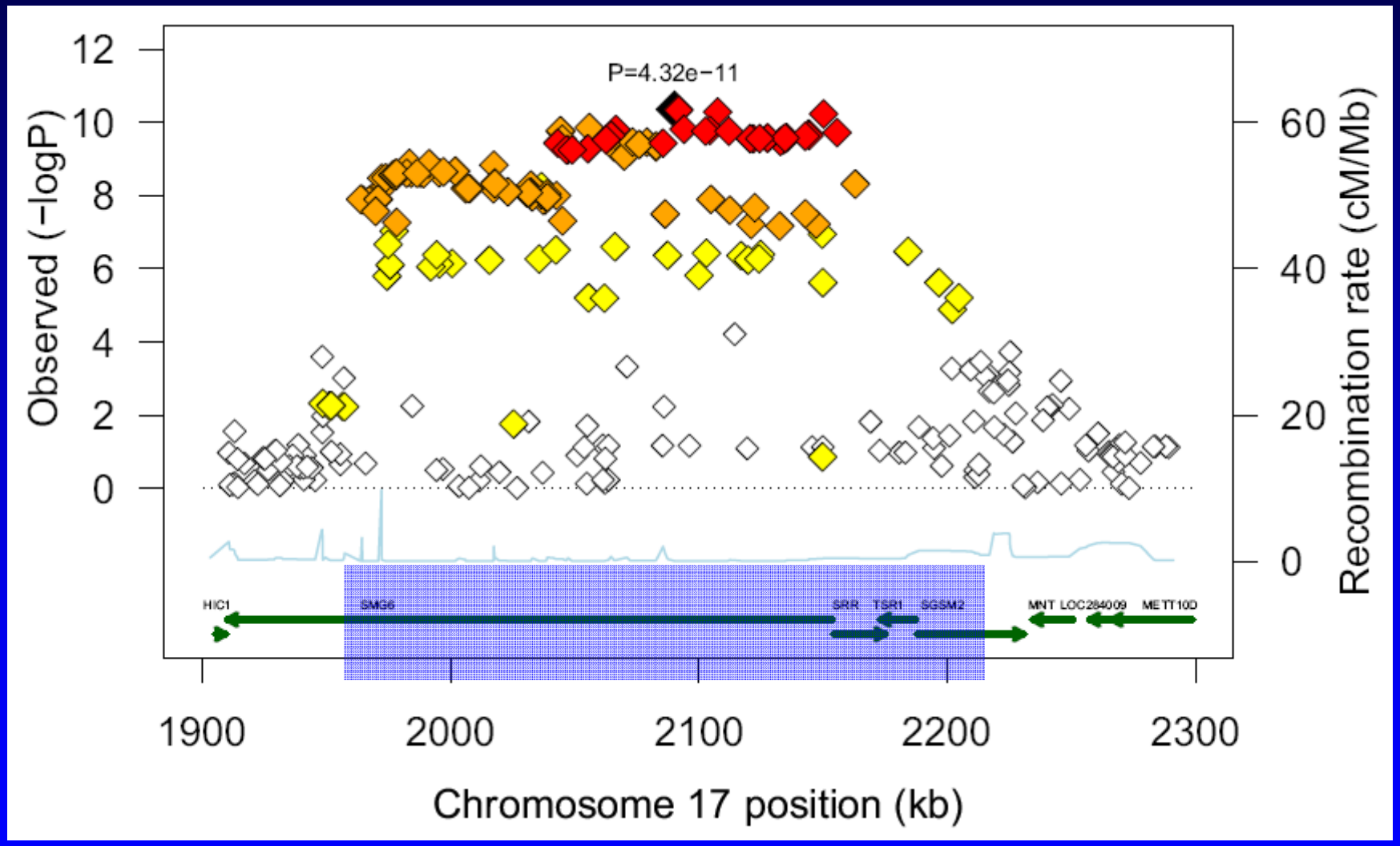
Vasan RS et al., *JAMA* 2009; 302:168-78.

Regional Plot for rs89107 Associated with LV Internal Dimensions



Vasan RS et al., *JAMA* 2009; 302:168-78.

Regional Plot for rs10852932 Associated with Aortic Root Diameter



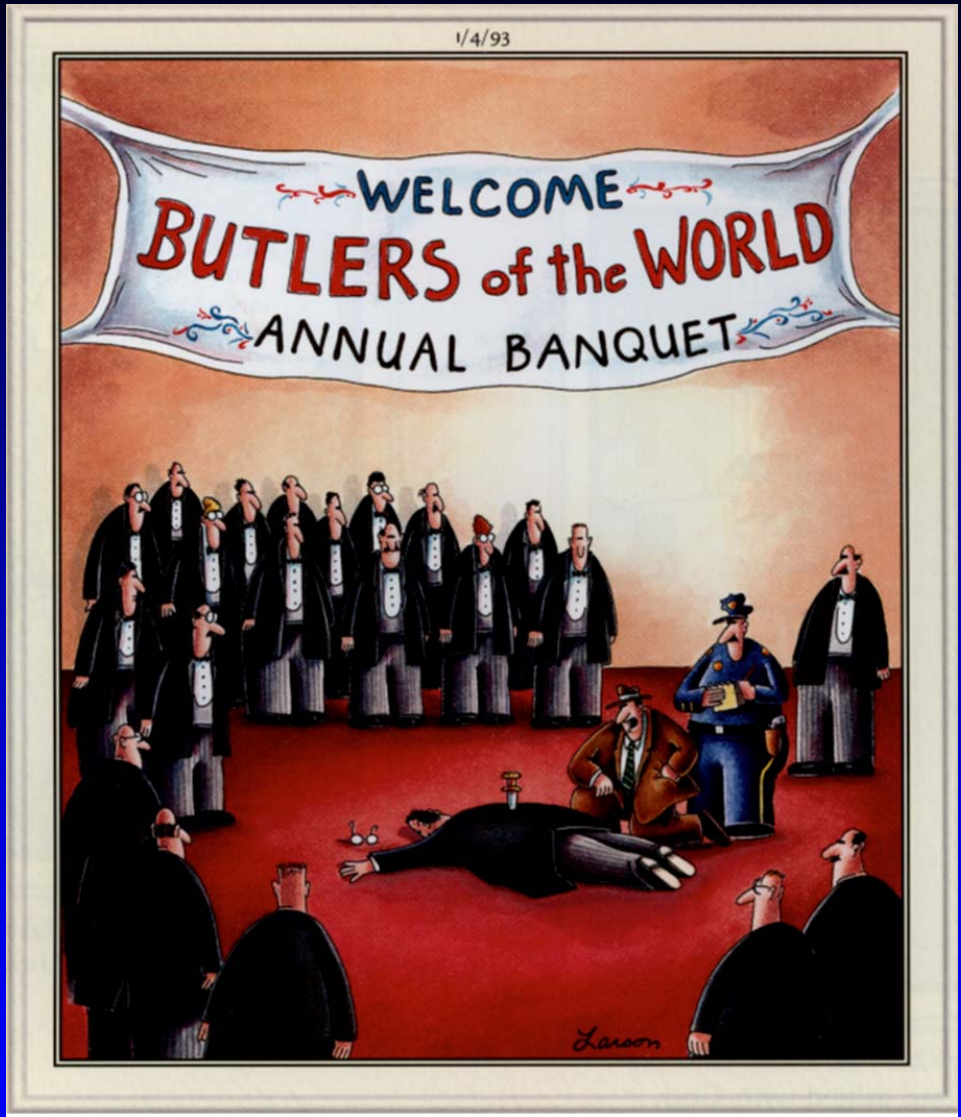
Vasan RS et al., *JAMA* 2009; 302:168-78.

Unique Aspects of GWA Studies

- Permit examination of inherited genetic variability at unprecedented level of resolution
- Permit "agnostic" genome-wide evaluation
- Once genome measured, can be related to any trait
- Most robust associations in GWA studies have not been with genes previously suspected of association with the disease
- Some associations in regions not even known to harbor genes

"The chief strength of the new approach also contains its chief problem: with more than 500,000 comparisons per study, the potential for false positive results is unprecedented."

Hunter DJ and Kraft P, *N Engl J Med* 2007; 357:436-39.



"God, Collings, I hate to start a Monday with a case like this."

Larson, G. *The Complete Far Side*. 2003.

nature genetics
volume 22 no. 1 may 1999

Freely associating

May 1999

Editorial: Once and Again—Issues Surrounding Replication in Genetic Association Studies

PERSPECTIVE

The Future of Association Studies: Gene-Based Analysis and Replication

Editorial

Replication Publication

Statistical false positive or true disease pathway?

John A Todd

Nat Genet July 2006

FEATURE

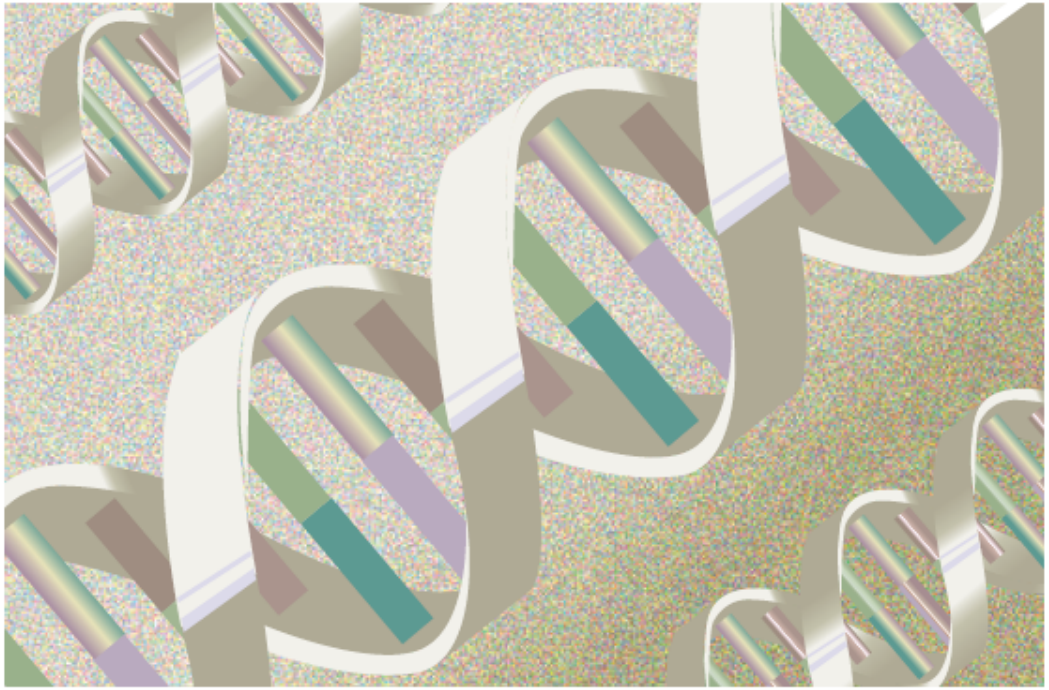
Replicating genotype–phenotype associations

What constitutes replication of a genotype–phenotype association, and how best can it be achieved?

NCI-NHGRI Working Group on Replication in Association Studies

The study of human genetics has recently undergone a dramatic transition with the completion of both the sequencing of the human genome and the mapping of human haplotypes of the most common form of genetic variation, the single nucleotide polymorphism (SNP)[1–3]. In concert with this rapid expansion of detailed genomic information, cost-effective genotyping technologies have been developed that can assay hundreds of thousands of SNPs simultaneously. Together, these advances have allowed a systematic, even 'agnostic', approach to genome-wide interrogation, thereby relaxing the requirement for strong prior hypotheses.

So far, comprehensive reviews of the published literature, most of which reports work based on the candidate-gene approach, have demonstrated a plethora of questionable genotype–phenotype associations, replication of which has often failed in independent studies[4–7]. As the transition to genome-wide association studies occurs, the challenge will be to separate true associations from the blizzard of false positives attained through attempts to rep-



studies because of issues in either the initial study or the attempted replication[4–6,32,33]. Small sample size is a frequent problem and can result

conclusion from the literature because follow-up studies have not consistently analysed the same markers or those in perfect linkage dis-

Chanock S, Manolio T, et al., *Nature* 2007; 447:655-60.

Traits with Published GWA Studies (n = 199, 3/4/11)

- Macular Degen.
- Glaucoma
- Myopia
- Optic Disc Size
- Corneal Thickness
- Corneal Dystrophy
- Retinal Vessel Size

- Lung Cancer
- SCLC Treatment Rsp.
- Prostate Cancer
- Breast Cancer
- Aromatase Inh. Rsp.
- Mammographic Dens.
- Colorectal Cancer
- Bladder Cancer
- Neuroblastoma
- Melanoma
- Basal Cell Cancer
- *TP53* Cancer Pred'n
- Ac/Ch Lymph. Leuk.
- Asparaginase Hypersen.
- Follicular Lymphoma
- Thyroid Cancer
- Myeloprolif. Syn.
- Testic. Germ Cell Ca.
- Glioma
- Ovarian Cancer
- Pancreatic Cancer
- Esophageal Cancer
- Nasopharyngeal Ca
- Hepatocellular Ca
- Renal Cell Ca
- Bleomycin Sens.
- MTX Pharmacokin.

- Cleft Palate
- Periodontitis
- Tooth Development
- Quinine Taste Sens.
- Eosinophilic Esoph.
- Infl. Bowel Disease

- Celiac Disease
- Hirschsprung Dis.
- Ileal Carcinoid
- Biliiubin Levels
- Gal
- 1° S
- Bili
- Nor
- Cir
- Dru
- Ace
- Hep
- Hep
- Chr

- ECG
- Cor
- Cor
- Hea
- Atri
- Ver
- Res
- Str
- Intr
- Car
- Kay
- Mo
- Hyp
- Ant
- Aor
- Lip
- Wa
- Xim
- Clo

- Par
- Amyotrophic Lat.Scler.
- Multiple Sclerosis
- MS Interferon-β Rsp.
- Prog. Supra. Palsy
- Neuromyelitis Optica

- CSF Protein Levels
- Tauopathies
- Alzheimer's Disease
- Var. Creutzfeldt-Jakob

- ADHD Treatment Rsp.
- Personality Traits
- Carbamazepine Rsp.

- Rheum. Arthritis

- RA Anti-TNF Rsp.
- Syst. Lupus Eryth.
- Juv. Idiop. Arthritis
- Ankylosing Spond.

- Obesity, BMI, Waist
- IR, MetabolicTraits
- Adipokine Levels
- Anorexia nervosa
- Exercise Behavior

- Fetal Growth
- Height
- Digit Length Ratio
- Thyroid Function
- Menarche
- Menopause/Ov. Failure
- Polycystic Ovary Syn
- Endometriosis
- Alopecia
- Male Infertility
- Erectile Dysfunction
- Hypospadias

- High Altitude Adapt.
- Fetal Hemoglobin
- Iron Status
- Hem/Thromb Levels
- C-Reactive Protein
- Adhesion Molecules
- Eosinophil Numbers
- Total IgE Levels
- Urate Levels, Gout
- Protein Levels
- N-Glycan Levels
- PSA Levels
- Folate Path. Vitamins
- β-Carotene Levels
- Vitamin D Levels
- Phosphorus Levels
- Sphyngolipid Levels
- Recombination Rate
- Telomere Length
- Longevity
- Radiation Response
- Self-Rated Health
- Constitut. Med. Type
- Hair Color/Morphol.
- Pigmentation
- Vitiligo
- Keloid
- Recessive Diseases

Neuroblastoma
Glioma
Ovarian Cancer
Primary Sclerosing Cholangitis
Amyotrophic Lateral Sclerosis
Progressive Supranuclear Palsy
Idiopathic Pulmonary Fibrosis
Height
Alopecia
Hair Color
Urate Levels
Clopidogrel Platelet Response
Stevens-Johnson Syndrome

NHGRI Catalog of GWA Studies:

http://www.genome.gov/gwastudies/

genome.gov

Google™ Search SEARCH

As of 04/15/10, this table includes 544 publications and 2664 SNPs.

This table is best viewed with a screen resolution of 1280 x 1024 or higher.

Search By:

PNAS

Potential etiologic and functional implications of genome-wide association loci for human diseases and traits

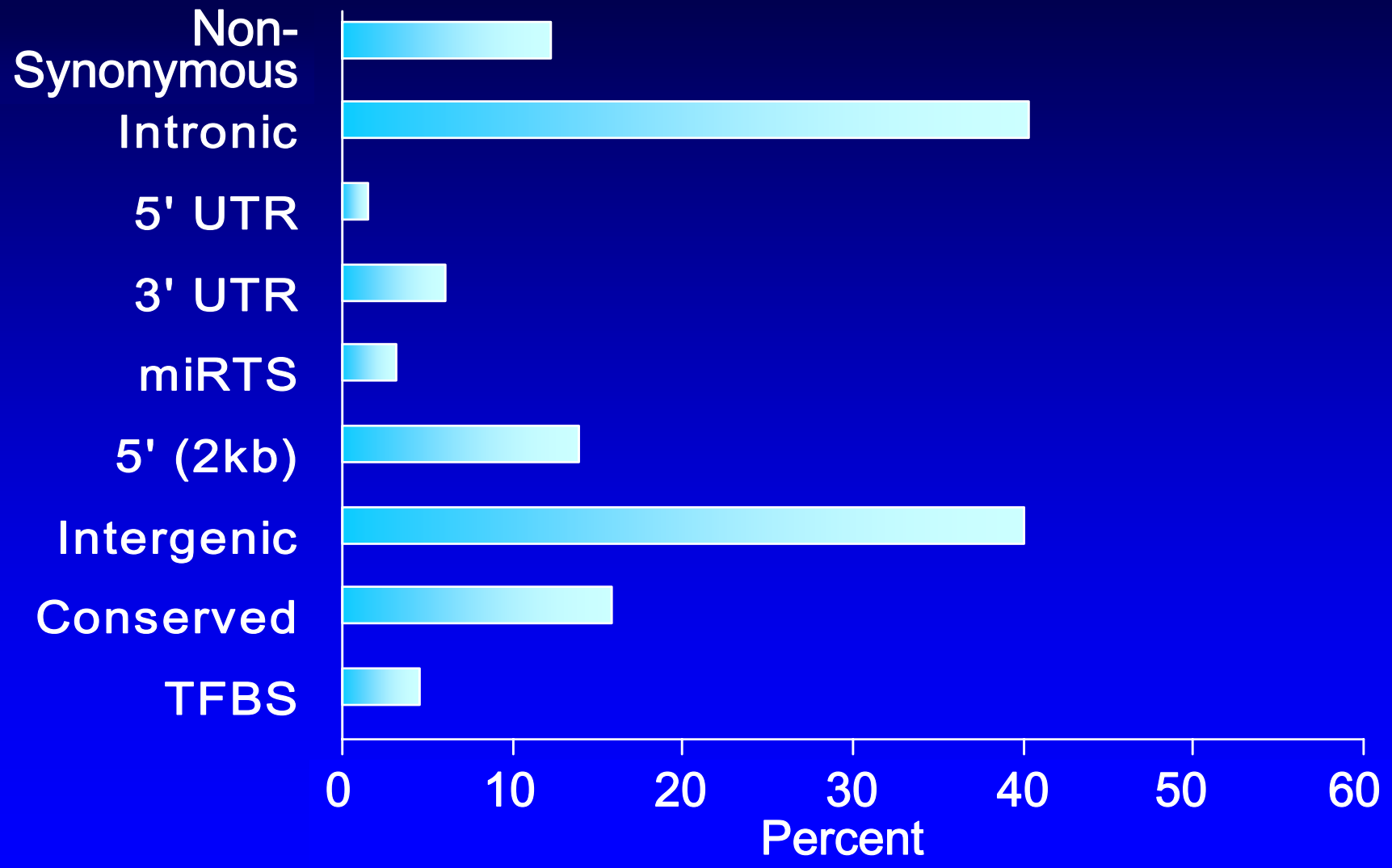
Lucia A. Hindorff[a,1], Praveen Sethupathy[b,1], Heather A. Junkins[a], Erin M. Ramos[a], Jayashri P. Mehta[c], Francis S. Collins[b,2], and Teri A. Manolio[a,2]

[a]Office of P… Information… anch, National Human G… 20892 … for Biotechnology

Contributed… r review February 19, 200…

We have … rait associations from publi… for use in investigating g… associated SNPs (TASs). Re… allele frequency 36%, inte… were associated with mod… 1.33, IQR 1.20–1.61]. Am… orted TASs were significant… nous sites [OR = 3.9 (2.2–7… egions [OR = 2.3 (1.5–3.6), … nly selected from genotypin… ntronic (45%) or intergenic… d in introns and were sign… ons [OR = 0.44 (0.34–0.58… Ss than expected by chance … positive selection [OR = 1.3 (0.8–2.1), p = 0.2]. This new online resource, together … f Published Genome-… nome.gov/gwastudies), … SNP-trait associations … e, we (i) describe the … ods we have used to … descriptive character-… risk allele frequencies … rlying functionality of … nomic annotation sets … onte Carlo simulations … etween recent human … es. There are several … s within the present … iant is often unknown. … variant may be: (i) the TAS itself, (ii) a known common SNP in strong linkage disequi-

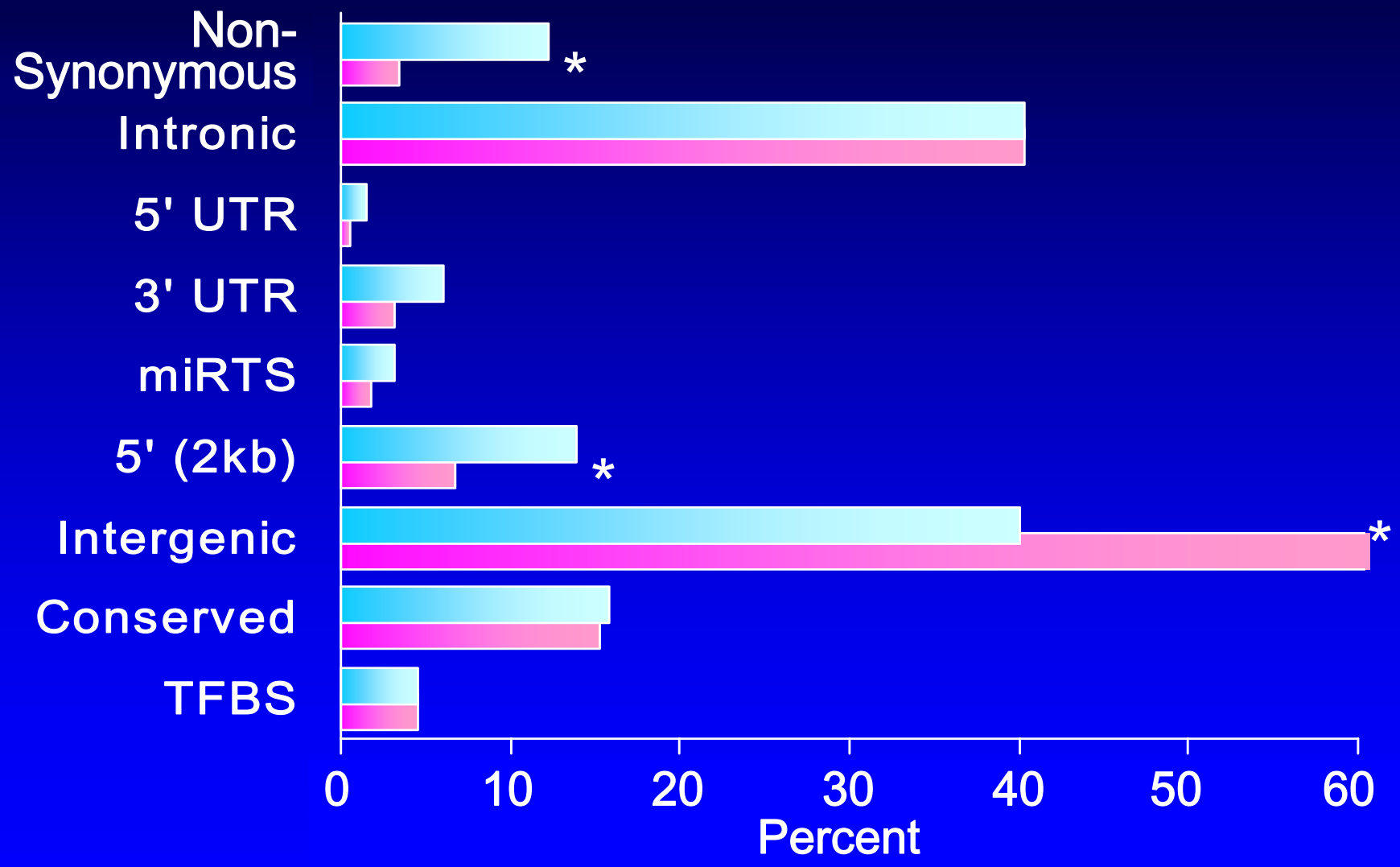
Functional Classification of 465 Trait - Associated SNP (TAS) Blocks ($r^2 > 0.9$)



Hindorff L et al, *PNAS* 2009; 106:9362-67.

TAS Block

Functional Classification of 465 TAS Blocks and Random LD Blocks

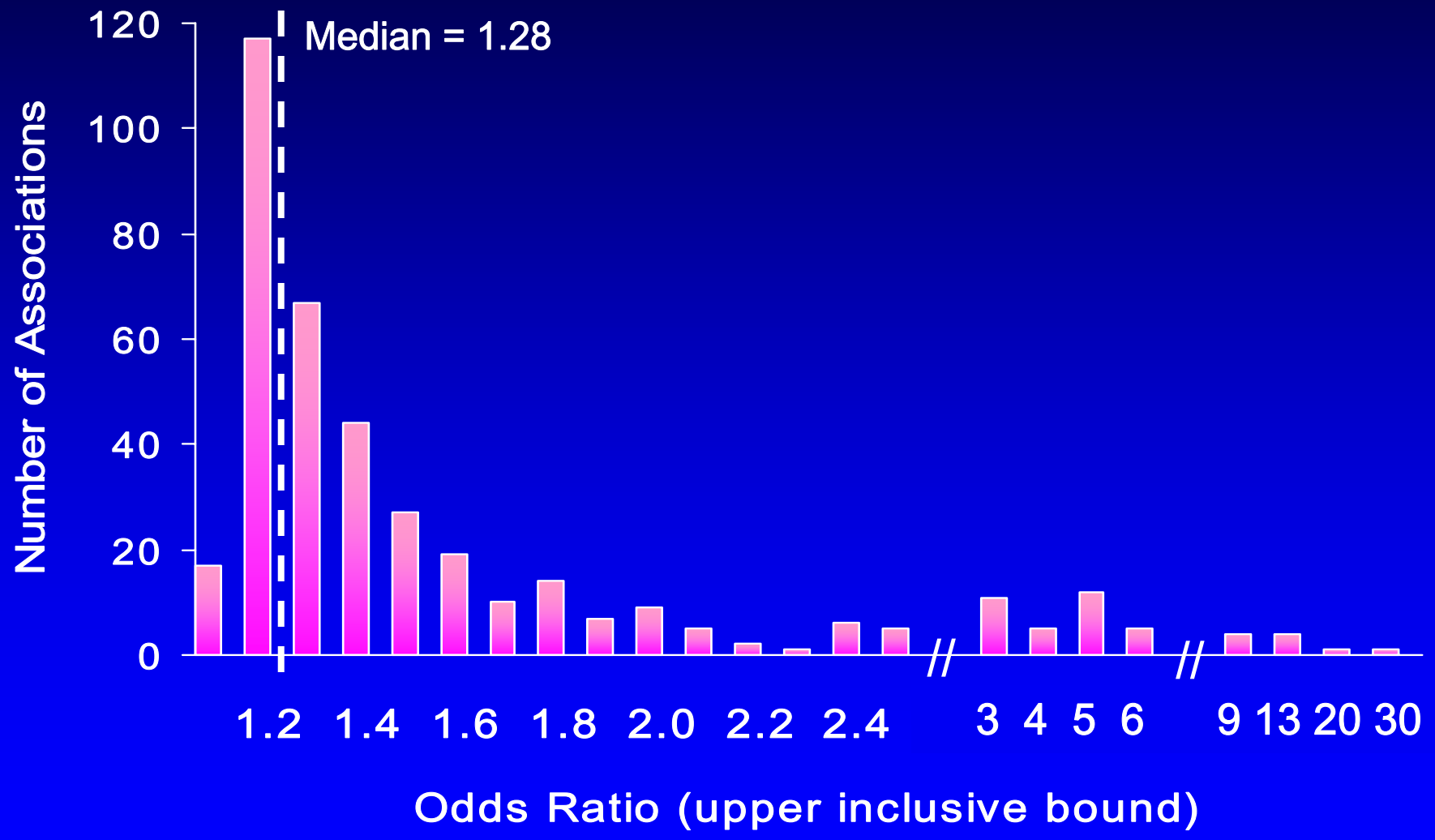


Hindorff L et al, *PNAS* 2009; 106:9362-67.

TAS Block

Random Block

Odds Ratios of Discrete Associations



Discoveries to Date

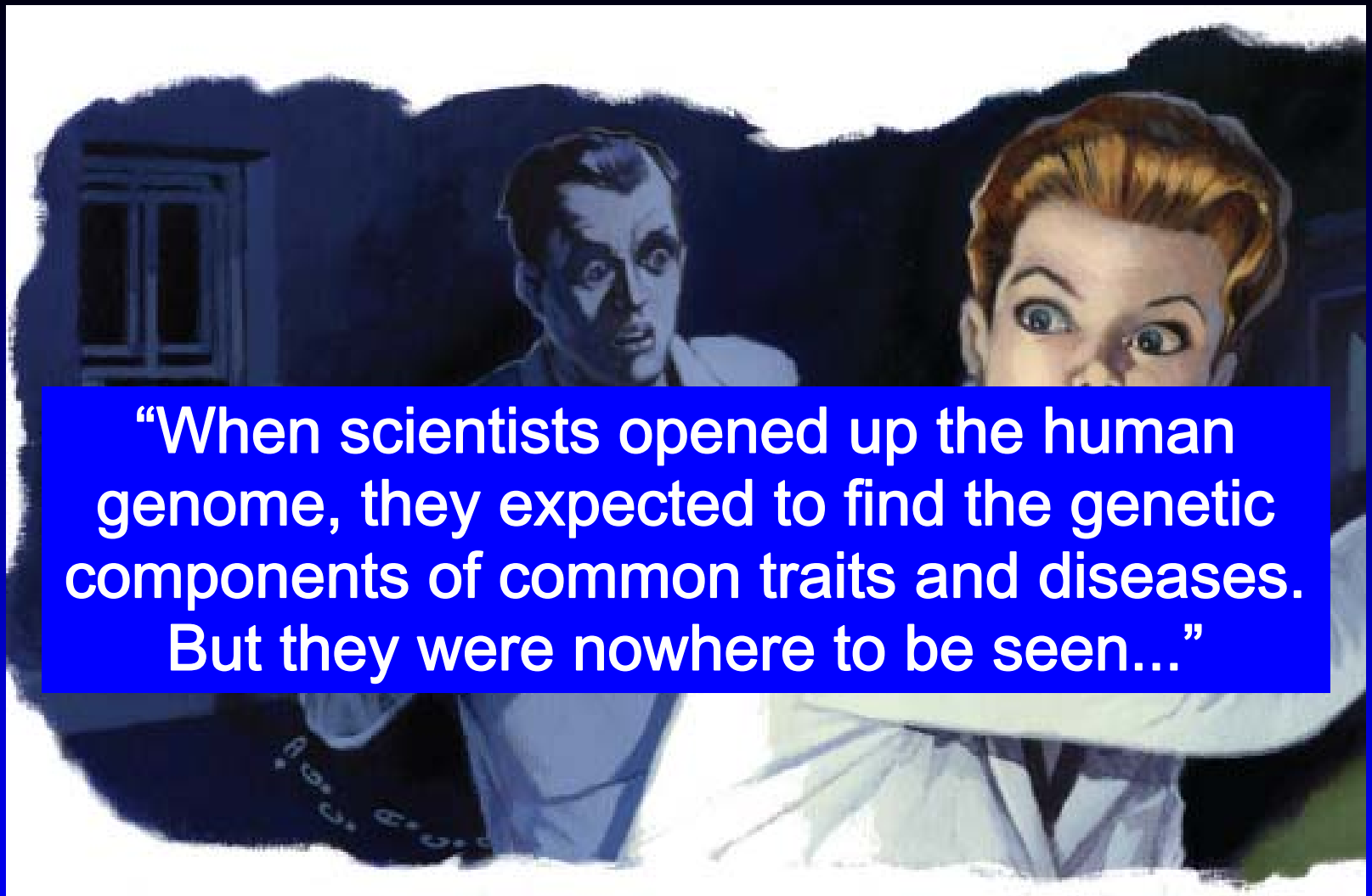
- Over 80% of GWA-identified SNPs and LD SNPs ($r^2 > 0.9$) are intronic or intergenic
- Very few GWA studies have been narrowed to true functional (presumed causal) variants
- Selection bias in genotyped SNPs for excess of missense, common, and European variants
- Most associated odds ratios are < 1.5
- Some traits have dozens or even hundred(s) of associated loci
- GWA-defined SNPs explain very little heritability

Familial Resemblance?

http://en.wikipedia.org/wiki/Image:Kennedy_bros.jpg#file

Proportion of Familial Risk Explained by GWA Studies to Date

Condition	Number of Loci	Percent (%) Explained	Measure
AMD	5	50	Sibling Recurrence Risk
Crohn's Disease	71	23	Liability
SLE	6	15	Sibling Recurrence Risk
T2DM	40	10	Sibling Recurrence Risk
Height	180	10	Phenotypic Variance
Lipid Levels	95	10 - 12	Phenotypic Variance



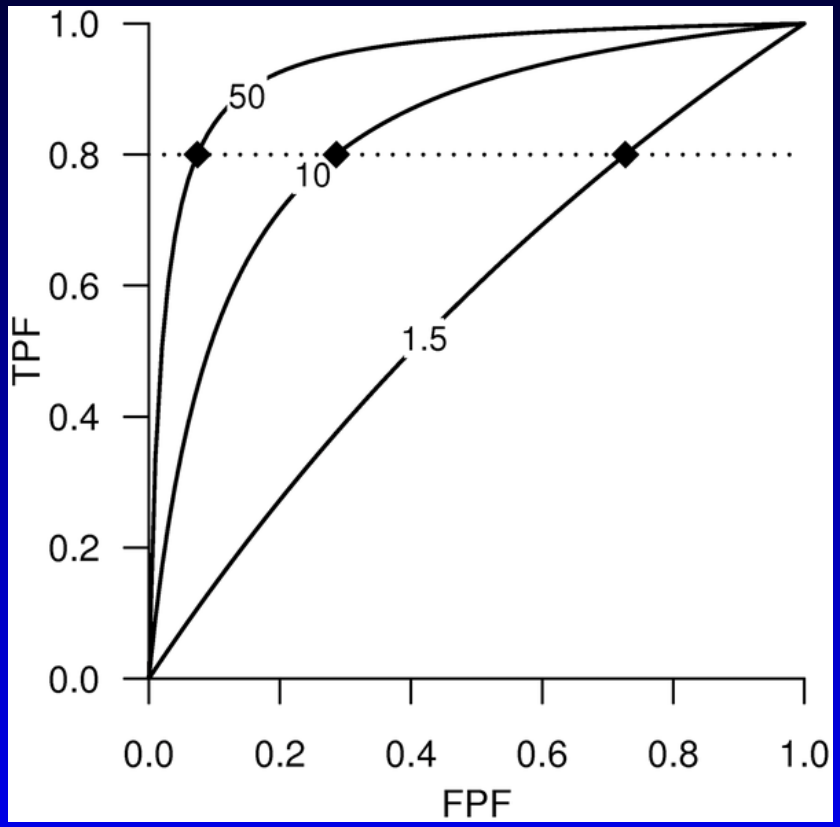
"When scientists opened up the human genome, they expected to find the genetic components of common traits and diseases. But they were nowhere to be seen..."

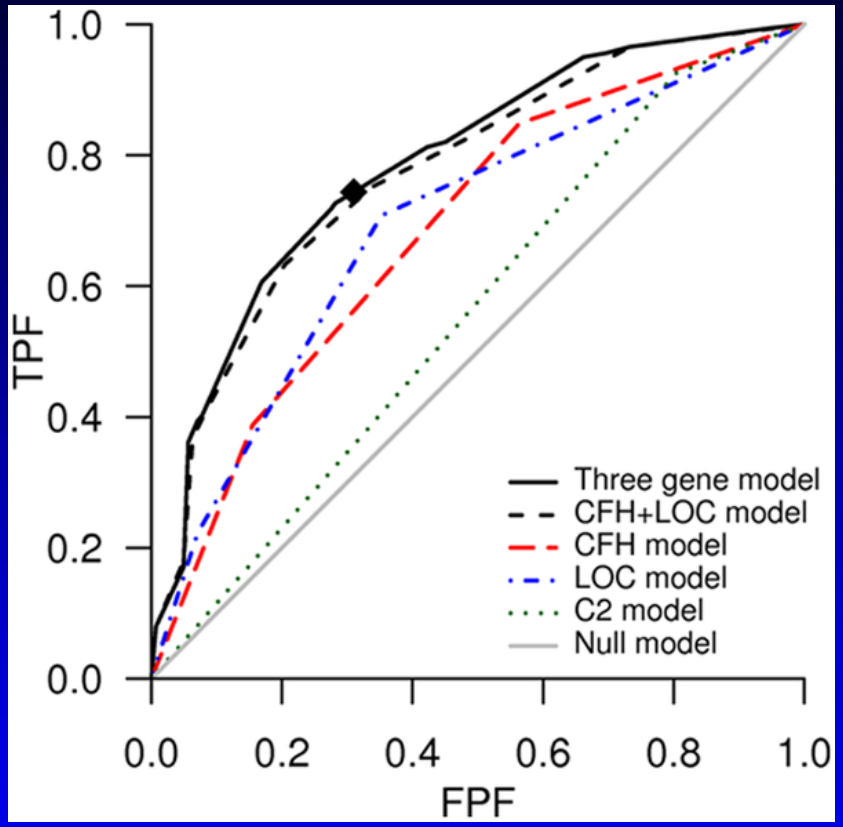
The case of the missing heritability

When scientists opened up the human genome, they expected to find the genetic components of common traits and diseases. But they were nowhere to be seen. **Brendan Maher** shines a light on six places where the missing loot could be stashed away.

Maher, B. *Nature* 2008; 456:18-21.

Area Under ROC Curve for Predicting AMD





- AUC with 3 variants = 0.79
- To correctly classify 74% of cases would misclassify 31% of controls
- At 80% sensitivity, would misclassify > 40% controls

Jakobsdottir J et al, *PLoS Genet* 2009; e1000337.

What are the limitations of genetic markers in risk assessment for disease?

- Most markers are not deterministic– many people without the markers will develop disease, and many people with the markers will not
- Most of the genetic risk remains unexplained
- Little or no evidence to date that interventions based on genotype improve outcome
- Genetic markers may provide additional risk information for more aggressive risk management in carriers, but again little evidence
- And yet…

Stevens-Johnson Syndrome

http://www.avimedi.net/en/stevens-johnson-syndrome-photos.html#photos

Stevens-Johnson Syndrome

http://www.avimedi.net/en/stevens-johnson-syndrome-photos.html#photos

Stevens- Johnson Syndrome, HLA-B*1502, and Carbamazepine

Association between prostaglandin E receptor 3 polymorphisms and Stevens-Johnson syndrome identified b

Genome-wide association study of serious

Genetic susceptibility to carbamazepine-induced cutaneous adverse drug reactions

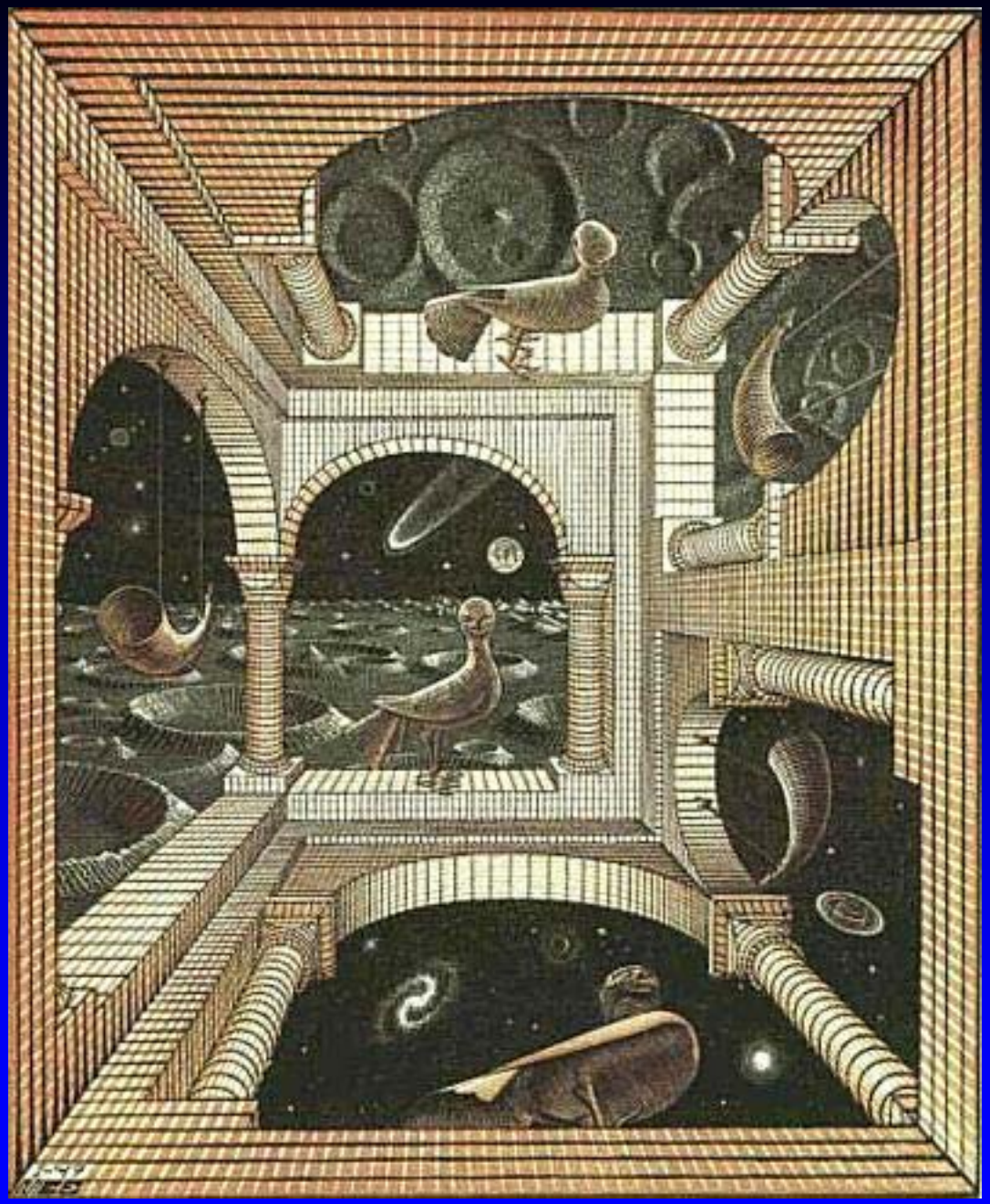
Shuen-Iu Hung[a,1], Wen-Hung Chung[b,c,1], Shiou-Hwa Jee[d], Wen-Chieh Chen[e], Yun-Ting Chang[f], Woan-Ruoh Lee[g], Shu-Ling Hu[h], Meng-Tse Wu[i],

CAUTION: This patient carries the HLA-B*1502 allele, a known risk factor for carbamazepine-induced SJS in Han Chinese individuals…

The anticonvulsant carbamazepine (CBZ) frequently causes cutaneous adverse drug reactions (cADRs), including maculopapular eruption (MPE), hypersensitivity syndrome (HSS), Stevens–Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN). We reported that SJS/TEN caused by CBZ is strongly associated with the *HLA-B*1502* gene in Han Chinese. Here, we extended our genetic study to different types of CBZ-cADRs (91 patients, including 60 patients with SJS/TEN, 13 patients with genetic susceptibility to CBZ-induced cADRs is phenotype-specific. *Pharmacogenetics and Genomics* 16:297–306
© 2006 Lippincott Williams & Wilkins.

Pharmacogenetics and Genomics 2006, 16:297–306

Keywords: carbamazepine, cutaneous adverse drug reactions, genetic polymorphisms, hypersensitivity syndrome, major histocompatibility complex, pharmacogenomics, Stevens–Johnson syndrome, toxic epidermal necrolysis



MC Escher, at http://web.ncf.ca/ek867

Clinical Questions that GWA-Defined Associations May Help Answer

- Can newly-discovered loci teach us about physiology or classification of disease?
- Can GWA-defined loci be used to develop specific, targeted treatments?
- Can GWA loci help direct treatment selection, dosing, or monitoring for adverse effects?
- Can GWA loci be used to predict disease or indicate more intensive screening?
- Can GWA loci identify those in whom more intensive environmental modification is appropriate?

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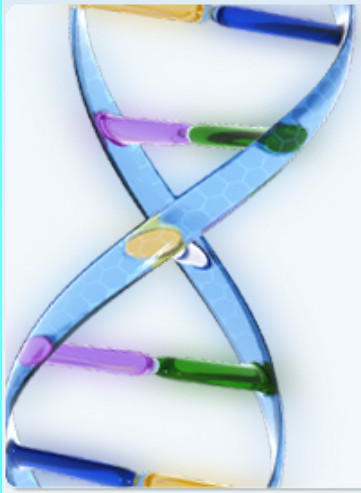

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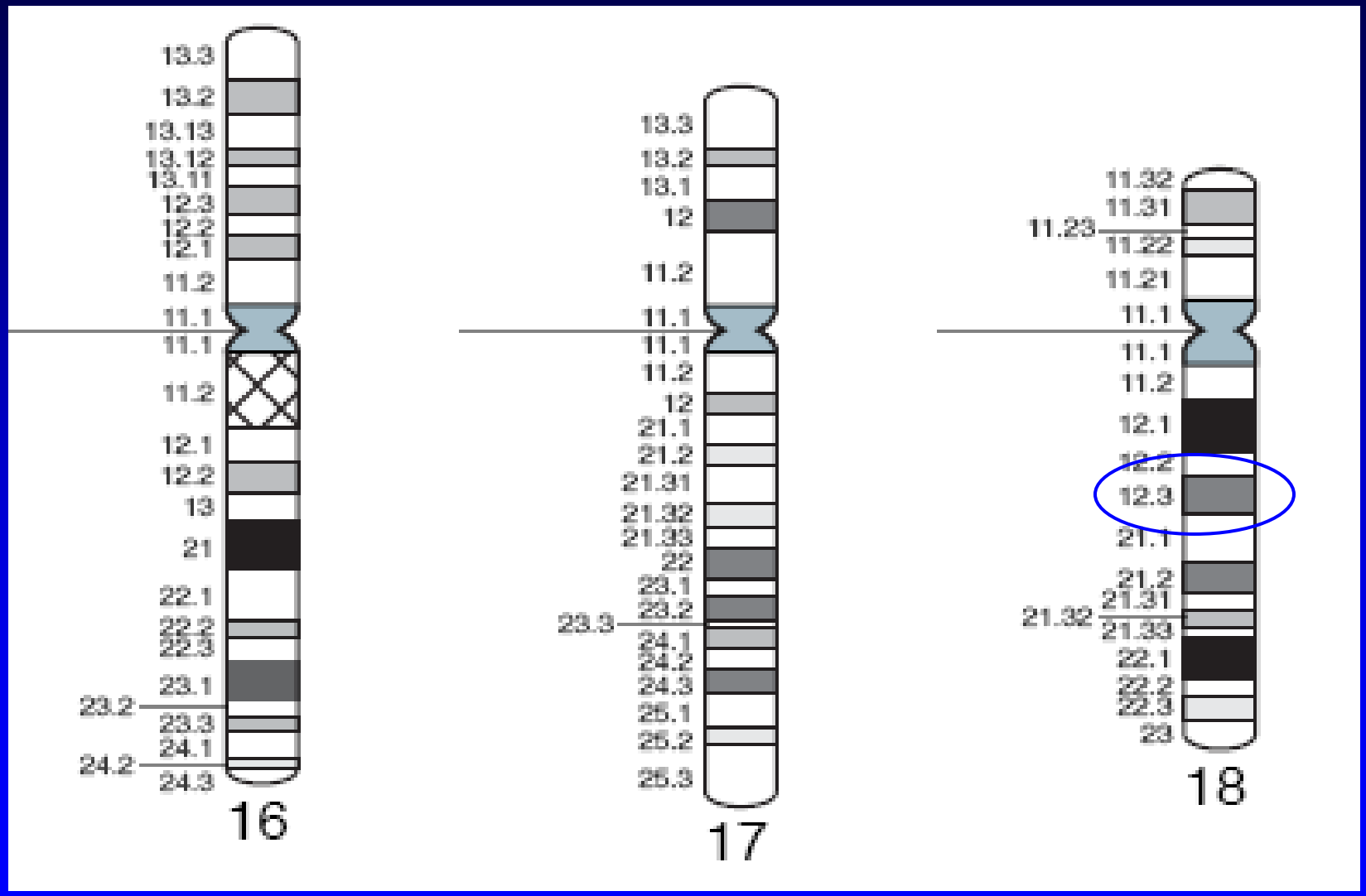
Predicting Disease Risk in the Genomic Era

- Pretty much the same as it was before
- May be useful in initiating counseling about non-genetic risk factors
- Possibly useful for screening for very high risk of many conditions at once
- Identifying genetic variants influencing response to drugs may be more immediately useful
- Clinical decision tools currently under development

Genomic Jargon Break