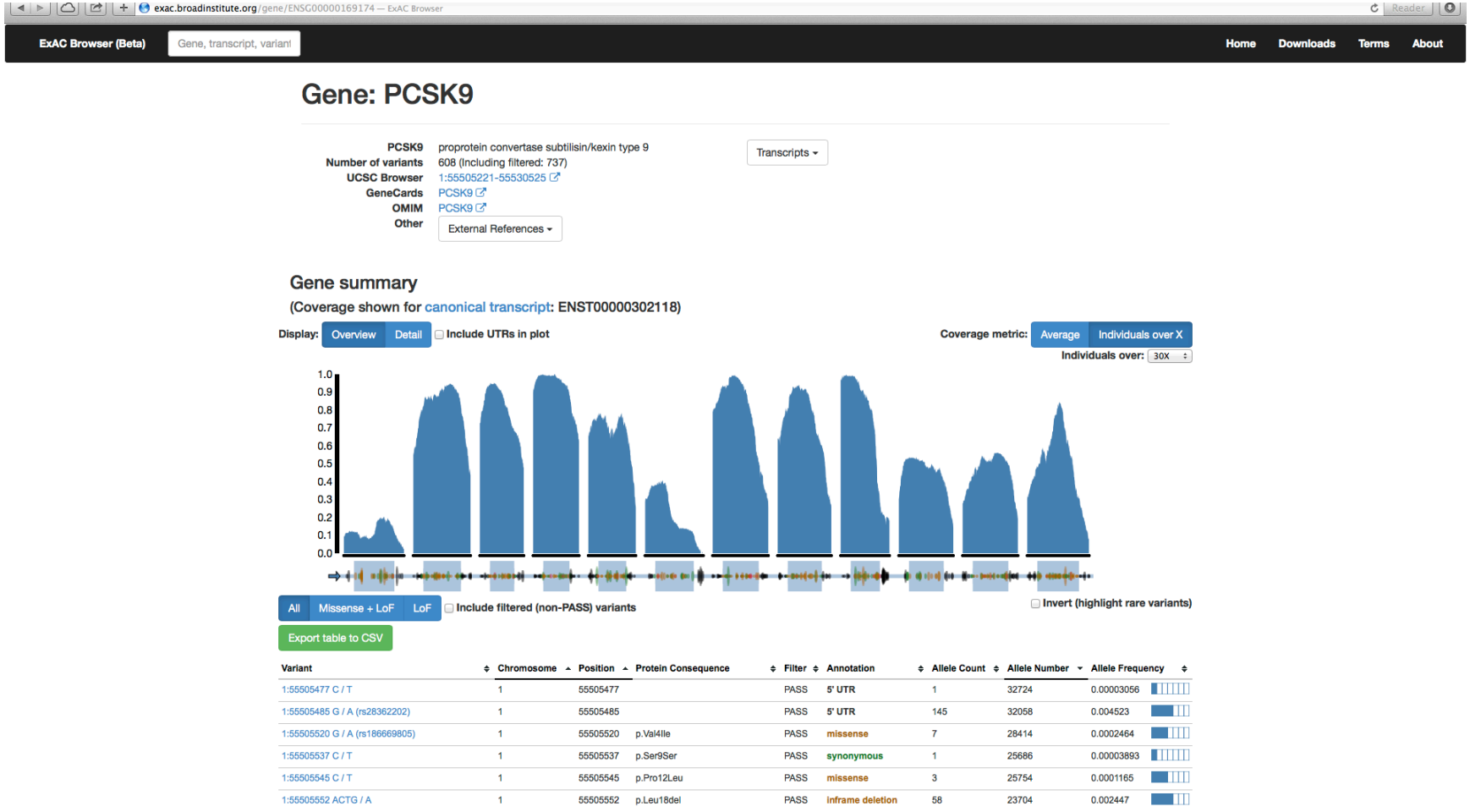
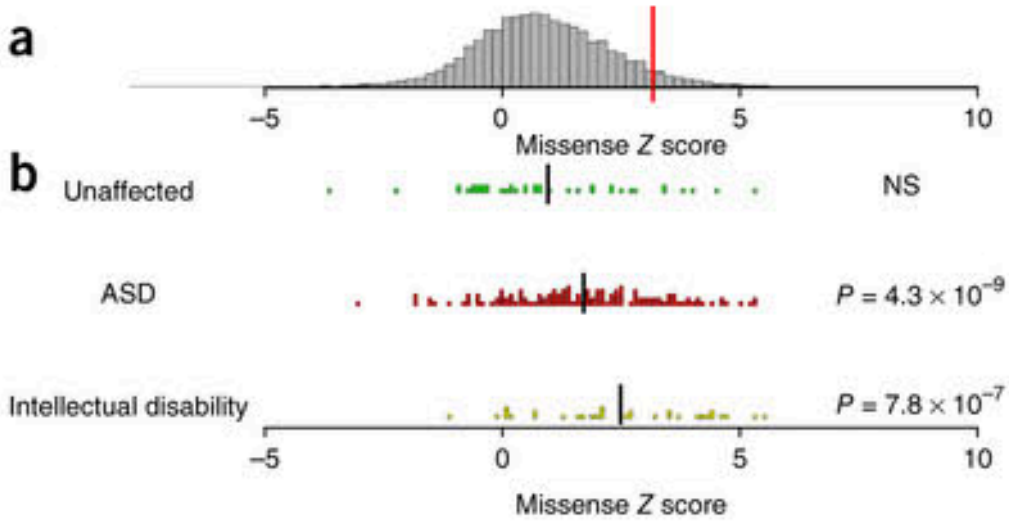
Loss of Function



Variant	Chromosome	Position	Protein Consequence	Filter	Annotation	Allele Count	Allele Number	Allele Frequency
1:55505477 C / T	1	55505477		PASS	5' UTR	1	32724	0.00003056
1:55505485 G / A (rs28362202)	1	55505485		PASS	5' UTR	145	32058	0.004523
1:55505520 G / A (rs186669805)	1	55505520	p.Val4Ile	PASS	missense	7	28414	0.0002464
1:55505537 C / T	1	55505537	p.Ser9Ser	PASS	synonymous	1	25686	0.00003893
1:55505545 C / T	1	55505545	p.Pro12Leu	PASS	missense	3	25754	0.0001165
1:55505552 ACTG / A	1	55505552	p.Leu18del	PASS	inframe deletion	58	23704	0.002447

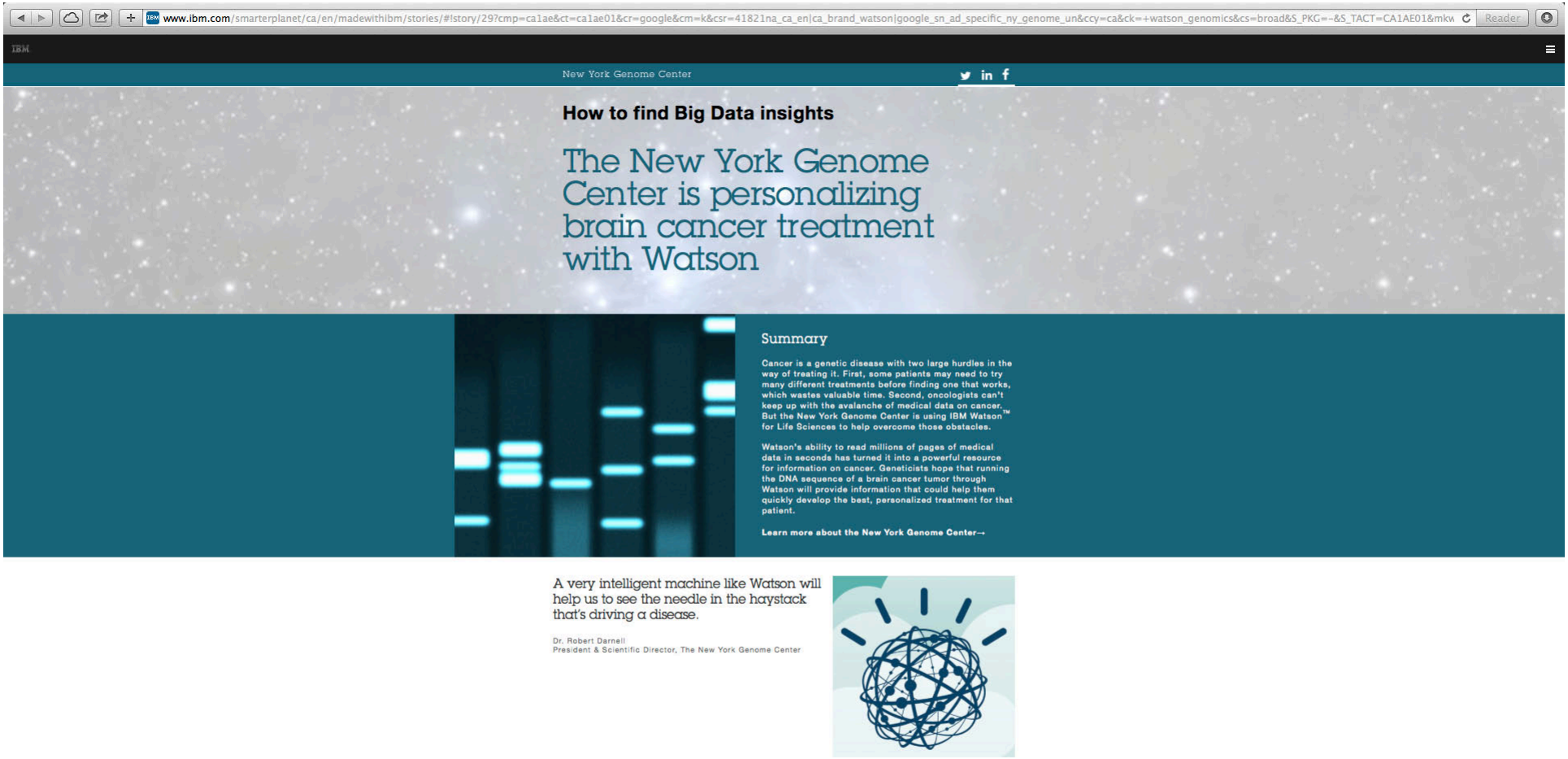
Loss of Function

Figure 2: Distributions of missense *Z* scores and *Z* scores for genes containing *de novo* loss-of-function mutations identified in unaffected individuals, ASD cases and intellectual disability cases.



(**a**) Distribution of missense *Z* scores. The red bar indicates a *Z* score of 3.09, or the threshold for inclusion in the set of 1,003 constrained genes. (**b**) Missense *Z* scores for genes containing *de novo* loss-of-function mutations in unaf…

Watson


www.ibm.com/smarterplanet/ca/en/madewithibm/stories/#!story/29?cmp=ca1ae&ct=ca1ae01&cr=google&cm=k&csr=41821na_ca_en|ca_brand_watson|google_sn_ad_specific_ny_genome_un&ccy=ca&ck=+watson_genomics&cs=broad&S_PKG=-&S_TACT=CA1AE01&mkw
Reader
IBM
New York Genome Center
How to find Big Data insights
The New York Genome Center is personalizing brain cancer treatment with Watson
Summary
Cancer is a genetic disease with two large hurdles in the way of treating it. First, some patients may need to try many different treatments before finding one that works, which wastes valuable time. Second, oncologists can't keep up with the avalanche of medical data on cancer. But the New York Genome Center is using IBM Watson™ for Life Sciences to help overcome those obstacles.
Watson's ability to read millions of pages of medical data in seconds has turned it into a powerful resource for information on cancer. Geneticists hope that running the DNA sequence of a brain cancer tumor through Watson will provide information that could help them quickly develop the best, personalized treatment for that patient.
Learn more about the New York Genome Center→
A very intelligent machine like Watson will help us to see the needle in the haystack that's driving a disease.
Dr. Robert Darnell
President & Scientific Director, The New York Genome Center

Depict

Figure 2: DEPICT analysis.

From

Genome-wide association analyses identify variants in developmental genes associated with hypospadias

Frank Geller, Bjarke Feenstra, Lisbeth Carstensen, Tune H Pers, Iris A L M van Rooij, Izabella Baranowska Körberg, Shweta Choudhry, Juha M Karjalainen, Tine H Schnack, Mads V Hollegaard, Wout F J Feitz, Nel Roeleveld, David M Hougaard, Joel N Hirschhorn, Lude Franke, Laurence S Baskin, Agneta Nordenskjöld, Loes F M van der Zanden & Mads Melbye

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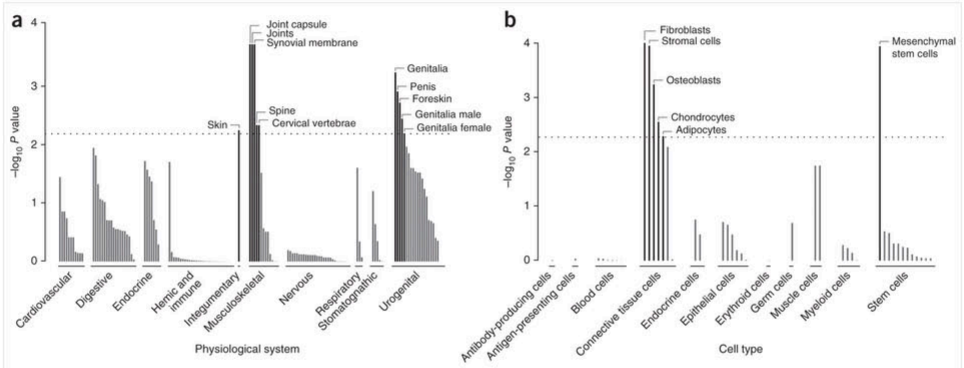

(**a**,**b**) Plots showing the enrichment of loci associated with hypospadias ($P < 1 \times 10^{-5}$ in the GWAS) in specific physiological systems (**a**) and cell types (**b**). On the basis of expression data from 37,427 human microarray samples, DEPICT identified statistically significant enrichment for 11 tissue annotations (**a**) and 6 cell type categories (**b**). Entries from the clade 'tissue' (including three significant tissues) are not displayed. Enrichments are grouped according to system or cell type and significance; annotations above the dotted line have FDR ≤ 5%.

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