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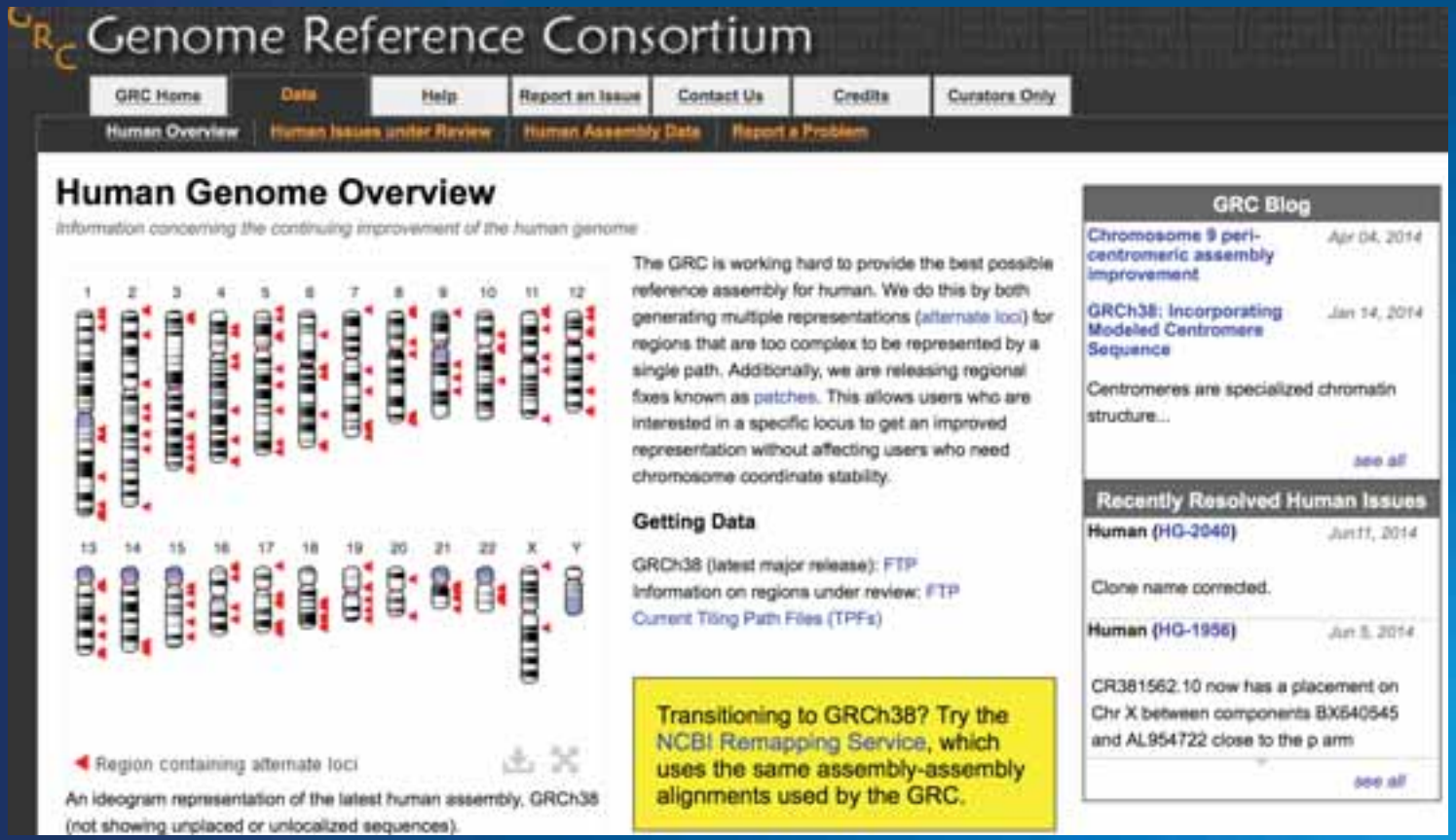
Using GIS for Delivering Molecular Diagnostics

Alice Rathjen, CEO, Founder DNA Guide

The MAP of the Human Genome

State of Art Bioinformatics:

Keyword Search Returns Points on the Map

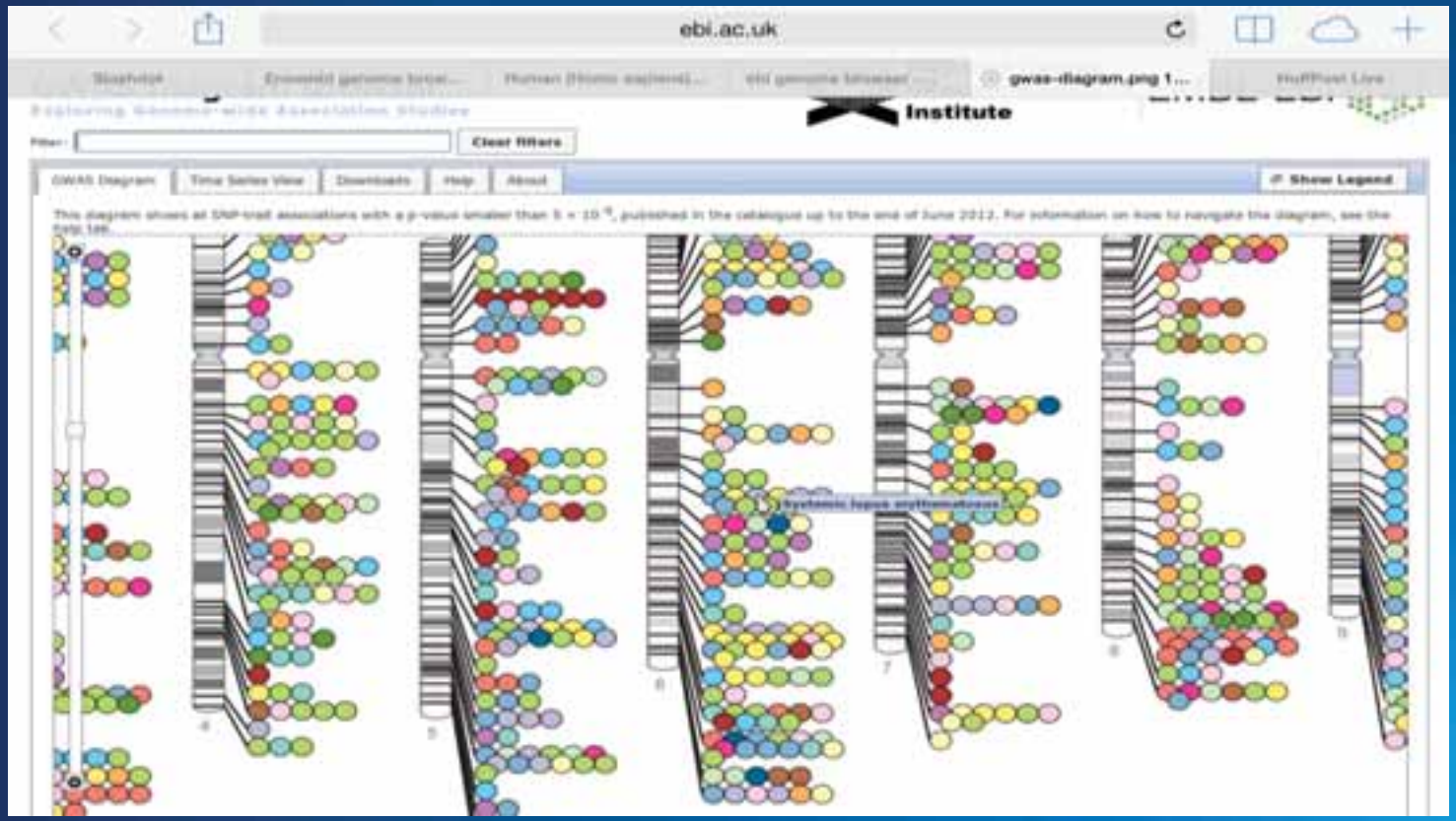


State of Art Bioinformatics:

Thematic Maps – European Bioinformatics Institute

State of Art Bioinformatics:

Pan and Zoom, Annotation on Tooltip



State of Art Bioinformatics:

Maps Finally Getting Crowded

Rapid Scientific Advances
Yet, Current Lab Format…

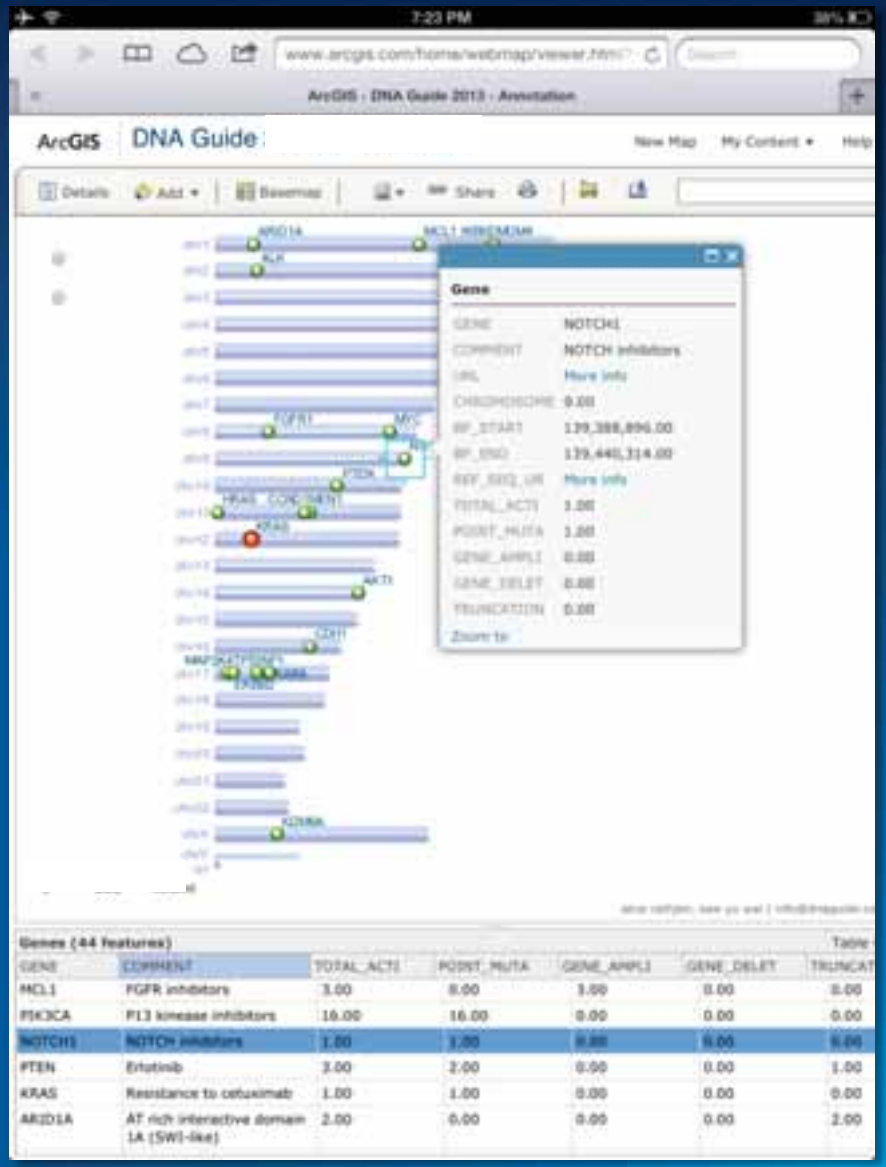
Molecular Diagnostic
Delivered as a Static Text via
PDF Email Attachment

Bringing Genomic Data to the Clinic using GIS

- GIS Already Up and Running for most Large Entities Facilities Mapping Infrastructure
- Proven Technology for distributing location based information to diverse users
- Rapid Deployment
- Easy Adoption
- Scales Indefinitely

Types of Molecular Diagnostics

- Cancer Tumor Analysis (200 genes)
- Carrier Testing (1-50 points of data)
- Drug Response (1-15 points of data)
- Disease Risk (1 to 60 points of data per disease)



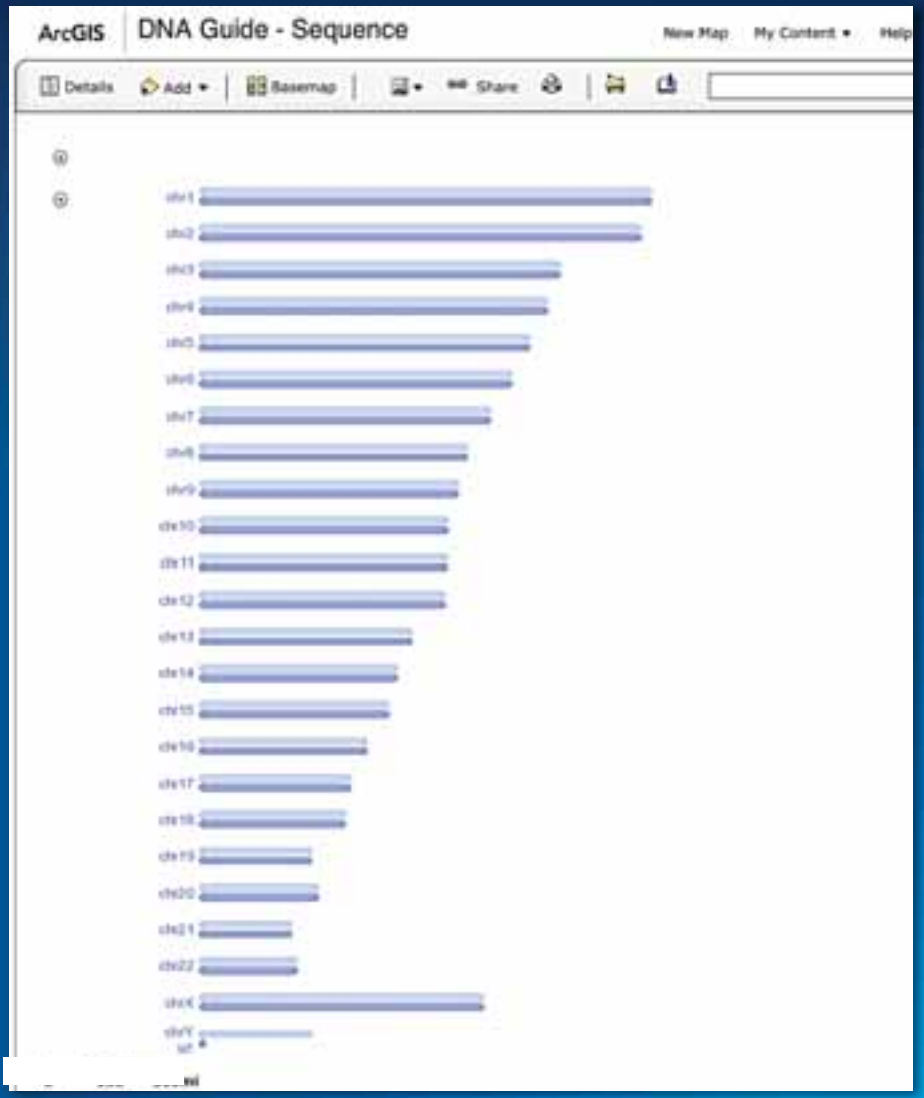
Genomic Coordinate Sources

- Human Reference Genome Builds
  http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human
- Human Genes
  HUGO www.**gene**names.org
- SNPs DBSNP
  www.ncbi.nlm.nih.gov/SNP

Bringing Genomic Data into GIS

Calculating x Coordinate

- Longest Chromosome , Chr 1, has 250,000,000 base pairs
- Using WebMercator,
  1 base pair = .01 map units
- X Coordinate = genetic marker position divided by 100
- Example coordinate/position (1234567 = 12345.67)



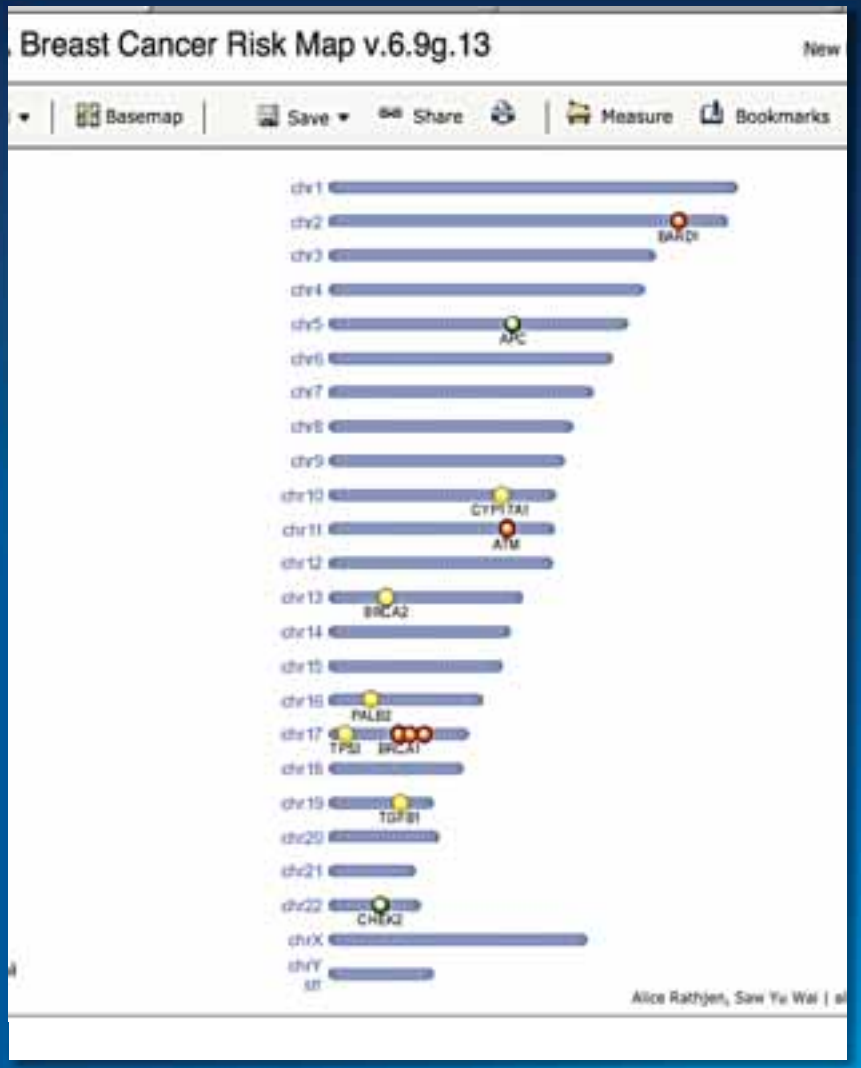
Bringing Genomic Data into GIS

Calculating y Coordinate*

23 Chromosome Pairs

- Chromosome number times -200.000.00 = y coordinate (i.e. chr 22 = -4400000.00)
- Chromosome X coordinate = -4600000.00
- Chromosome Y coordinate = -4800000.00

*midline, for diploid browser add .01 and subtract .01 for each chromosome pair



Custom Genomic Symbols for GIS

Genomes consist of large .txt files full of A, T, C and G's

DNA Bases: A, T, C, G's.

Bringing Genomic Data into GIS

A's and T's bond together, C's and G's bond together.

Double Helix (A's & T's, C's & G's)

T A G C

A T C G

Bringing Genomic Data into GIS

The data is .txt files is for one side of the DNA Strand.

Top Strand is the **Plus (+) Strand**

T+ A+ G+ C+

A- T- C- G-

Bottom Strand is the **Minus(-) Strand**

DNA Mapping

Sample Dataset

- RSID number is the research ID for the marker.
- The Genotype “AG” means the person has an “A” at this location on one of their Chromosome 1s and a G at this location on their second Chromosome 1

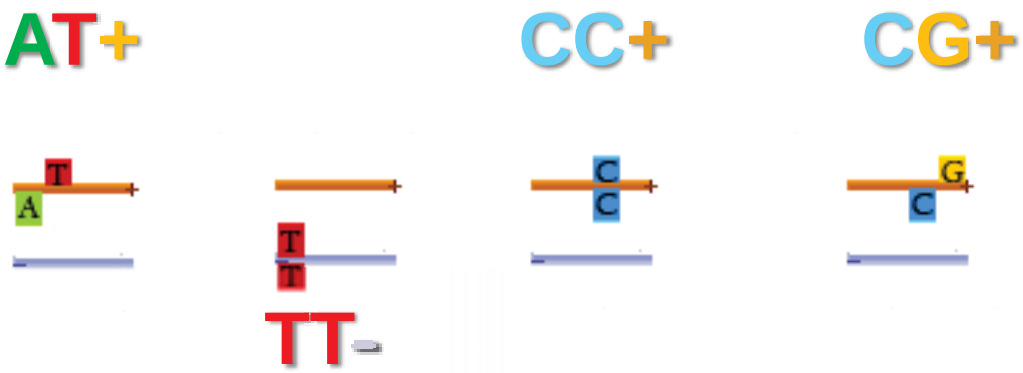
Human Reference Build 36 , + orientation

#rsid	chromosome	position	genotype
rs3094315	1	742429	AG
rs3131972	1	742584	AG
rs11240777	1	788822	AG
rs6681049	1	789870	CC
rs4970383	1	828418	CC
rs4475691	1	836671	CC
rs7537756	1	844113	AA
rs13302982	1	851671	GG
rs1110052	1	863421	GT
rs2272756	1	871896	AG
rs3748597	1	878522	CC
rs13303106	1	881808	AG
rs28415373	1	883844	CC
rs13303010	1	884436	AA

Custom Symbols for Spatial Analysis

Points in genome where humans vary are called SNPs

Patient data often shown in pairs of letters



(**One letter for each chromosome pair**)

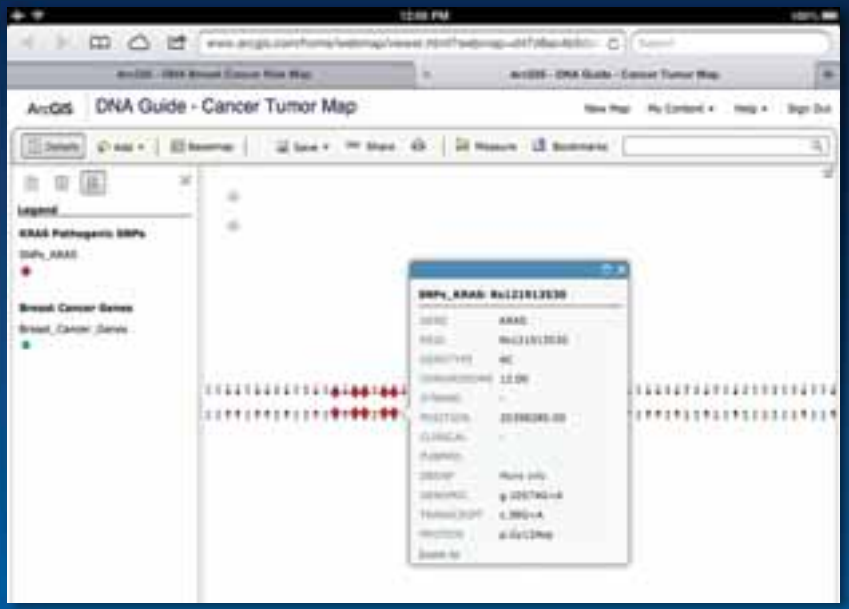
Benefits of GIS

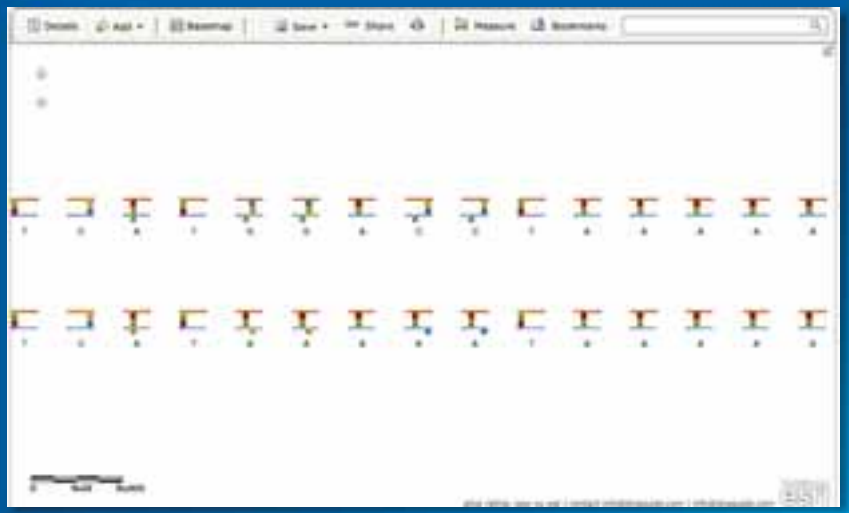
Enabling Information Flow

- Topological Analysis across layers
- Medical Utility
  (4 stars rating - Pathologists, AMA)
- Viewing Risk (E=Everyone, PG = Physician Guidance, R = Restricted –via genetic counselors)

Patient Engagement

- Real Time Consent
  (fine grain control for sharing info)





Thank You ESRI

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