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# **Preparing for the Genomic Data Deluge of Personalized Medicine**

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# Preparing for Personalized Medicine

- Deluge vs Managed Mapping Solutions
- DNA Mapping 101
- Genomic Coordinate Sources
- Recognizing the First Waves
- Addressing Needs of Various Stakeholders

# The Current State of Genetic Data.



# Fear and Hoarding

# Secure Sharing of Genomic Maps

The screenshot displays the ArcGIS interface for a map titled "DNA Breast Cancer Risk Map v.6.9g.13". The map shows a karyotype of human chromosomes with various genes and mutations marked. A popup window titled "Breast Cancer Risk" is open over a specific mutation on chromosome 2. The popup contains the following information:

Breast Cancer Risk	
Name:	C557S
Gene:	BARD1
Chr:	2
Position:	215617178
Mutation:	CC
Carrier:	CG
Common:	GG
RSID:	rs28997576
Strand:	-
In Honor Of:	
Sponsor:	
Zoom to	

Mapping Software:

- Proven Technology
- Rapid Deployment
- Easy Adoption
- Dynamic Reports/Maps
- Secure
- Scales Indefinitely

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# DNA Mapping 101 ... #1

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

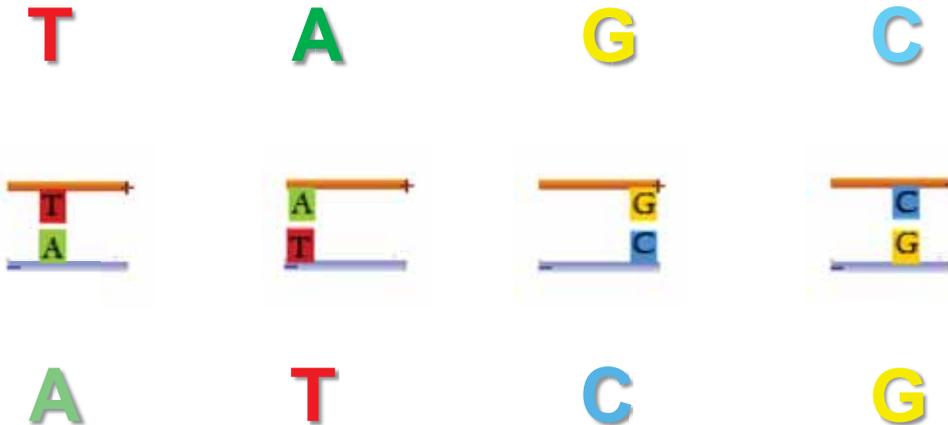
DNA Bases: A, T, C, G's.



# DNA Mapping 101 ... #2

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

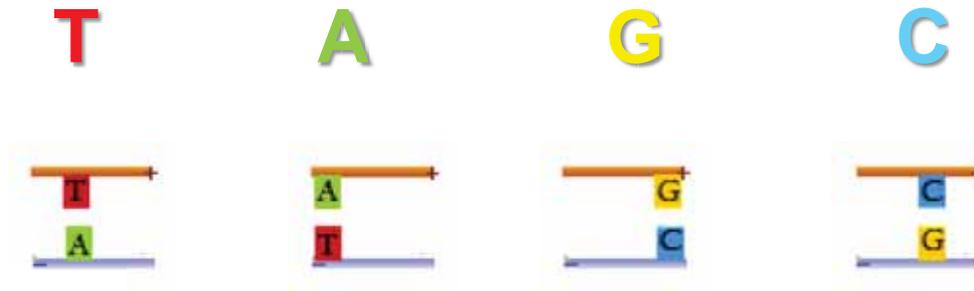
DNA bases come in pairs



# DNA Mapping 101 ... #3

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

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



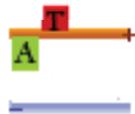
Bottom Strand is the **Minus(-) Strand**

# DNA Mapping 101 ... #4

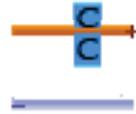
Points in genome where humans vary are called SNPs

Patient data often shown in pairs of letters

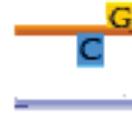
**AT+**



**CC+**



**CG+**



**TT-**

(One letter for each chromosome pair)

# DNA Mapping 101 ... #5

## Sample Dataset

- RSID number is the researchers 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

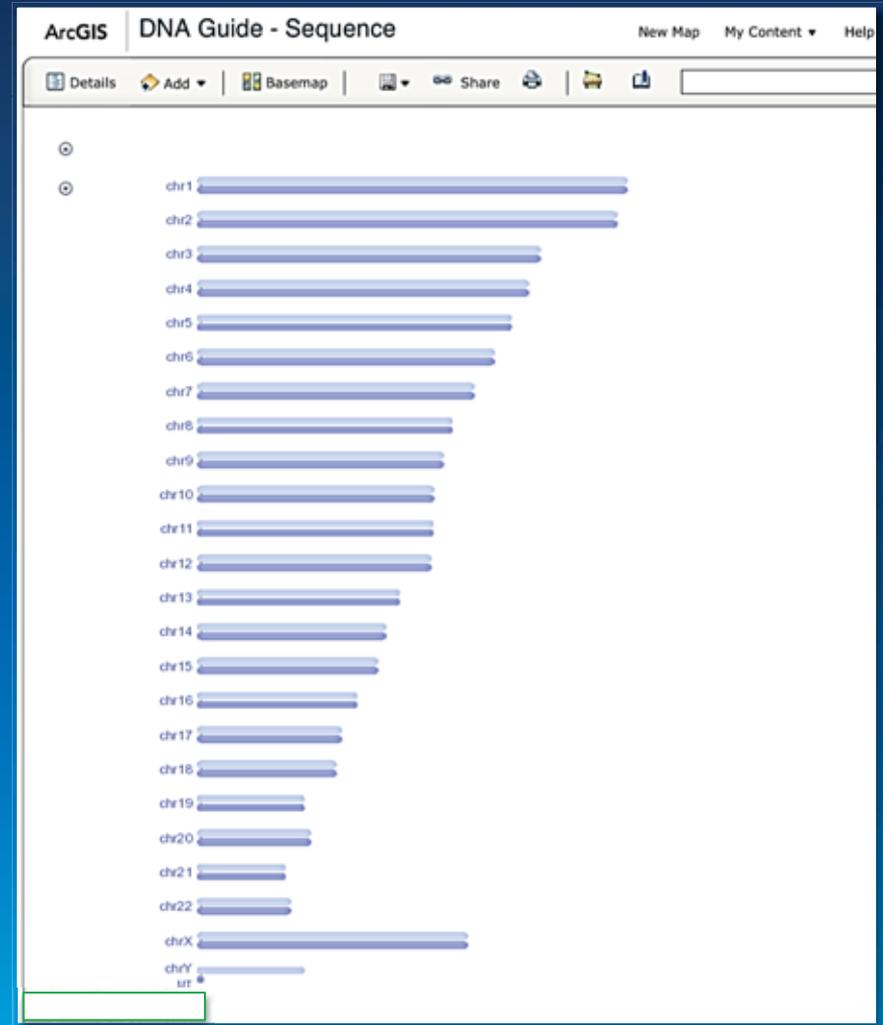
# Human Reference Build 36 , + orientation

#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

# DNA Mapping 101... #6

## Calculating X Coordinate

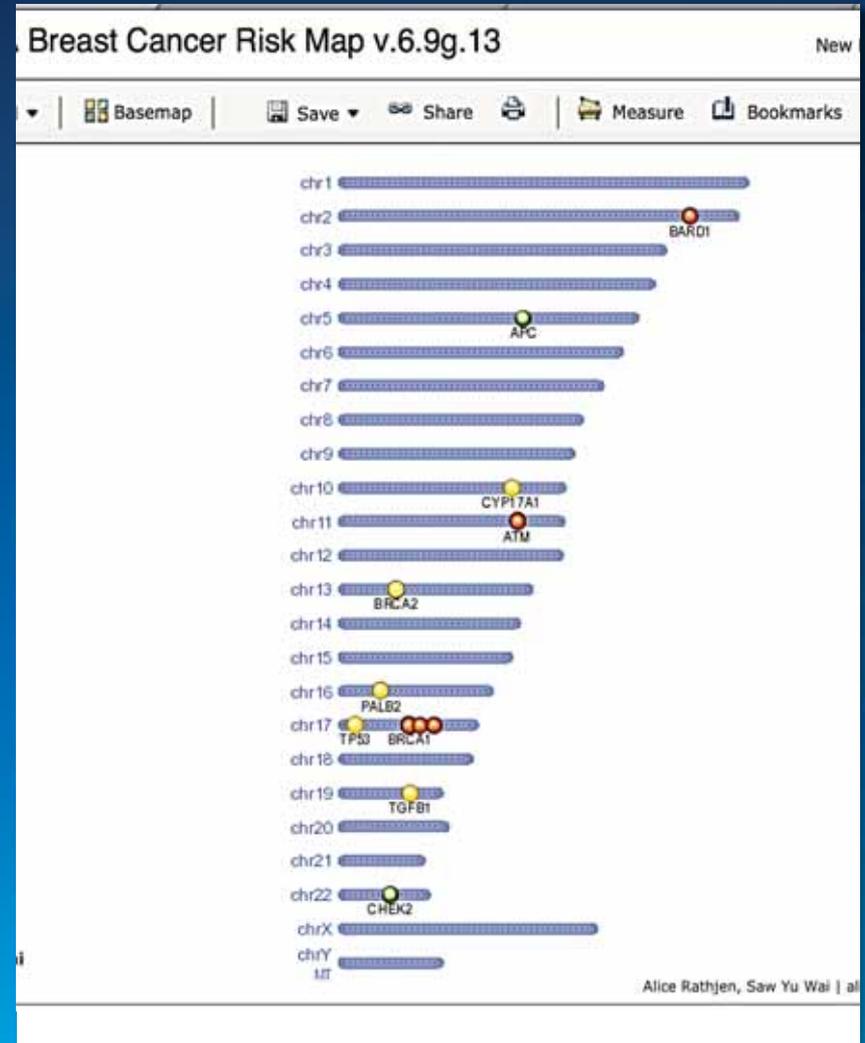
- Longest Chromosome , Chr 1, has 250,000,000 base pairs
- Using WebMercator, 1 base pair = .01 map units
- X Coordinate = genetic marker position / 100
- Example position (1234567 = 12345.67)



# DNA Mapping 101 ... #7

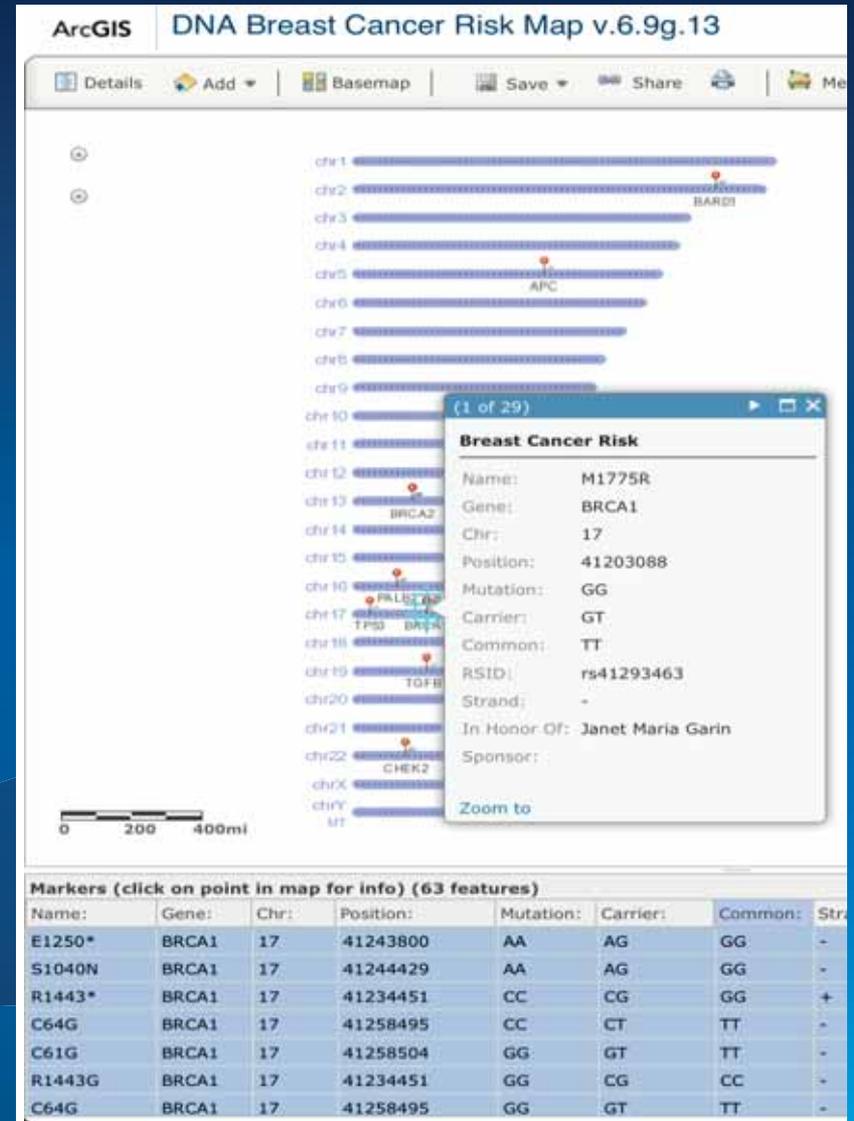
## Calculating Y Coordinate\*

- 23 Chromosome pairs
- Chromosome number times -200,000.00 = Y Coordinate.
- Chr X coordinate = -4,600,000.00
- Chr Y coordinate = -4,800,000.00
- \*midline, diploid browser add .01 and subtract .01 for each pair



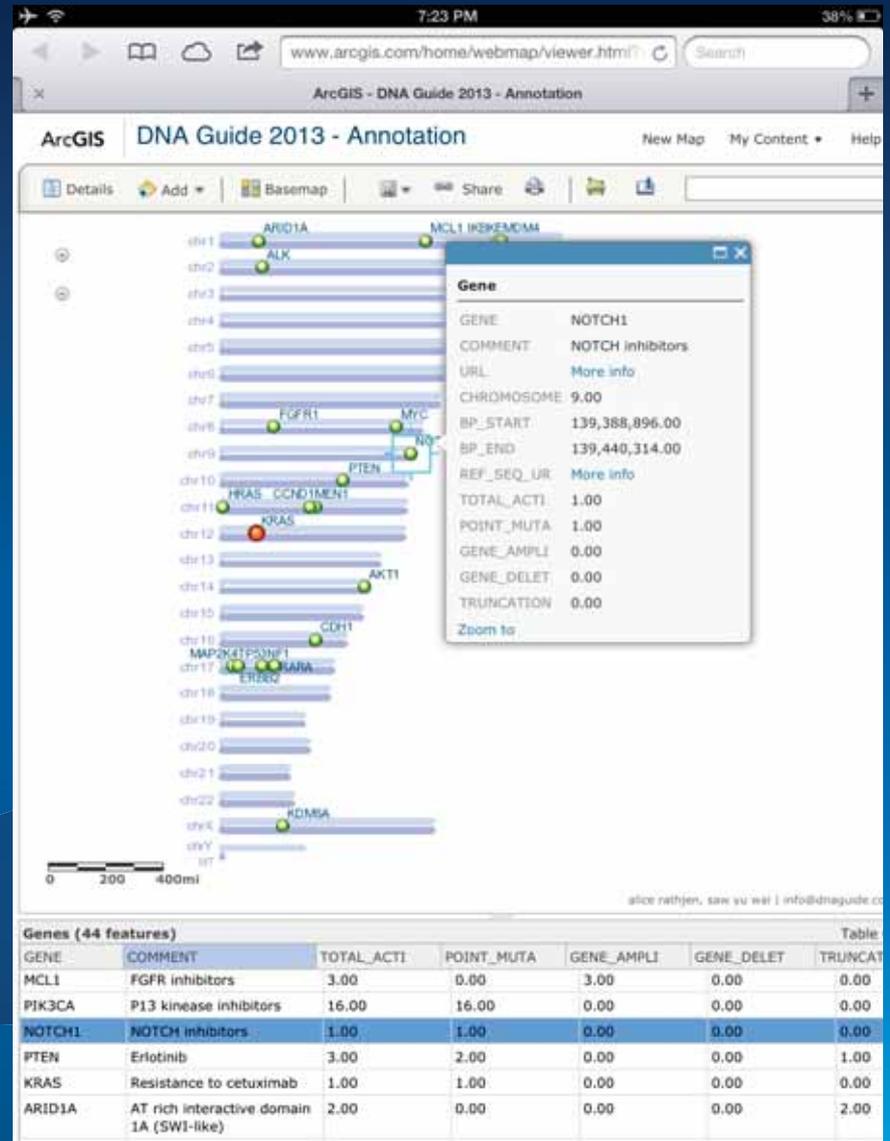
# Genomic Coordinate Sources

- Human Reference Genome Builds  
<http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human>
- Human Genes  
 HUGO [www.genenames.org](http://www.genenames.org)
- SNPs DBSNP  
[www.ncbi.nlm.nih.gov/SNP](http://www.ncbi.nlm.nih.gov/SNP)



# Waves of Patient Data

- 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)



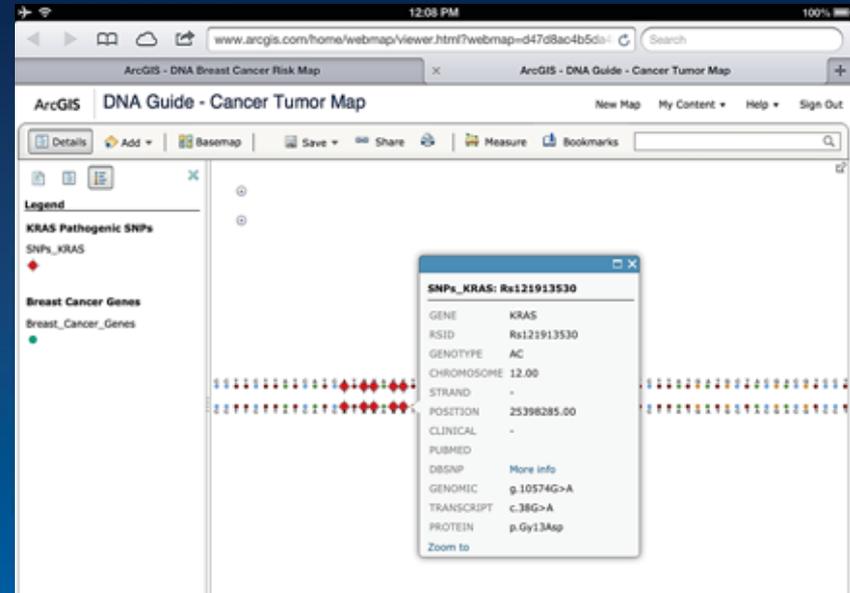
# Needs of Stakeholders

## Annotation Turf Wars

- Quality of Science (A-F, Scientific Peer Review)
- Medical Utility (5 stars, 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

Alice Rathjen

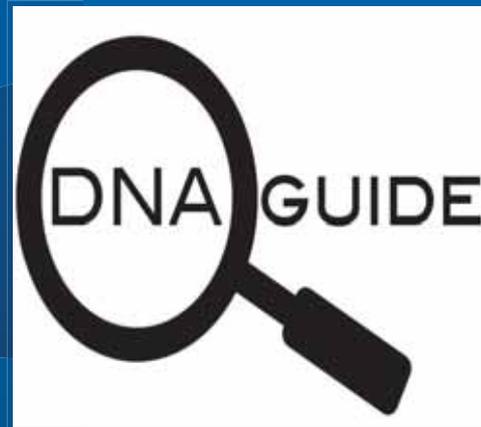
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