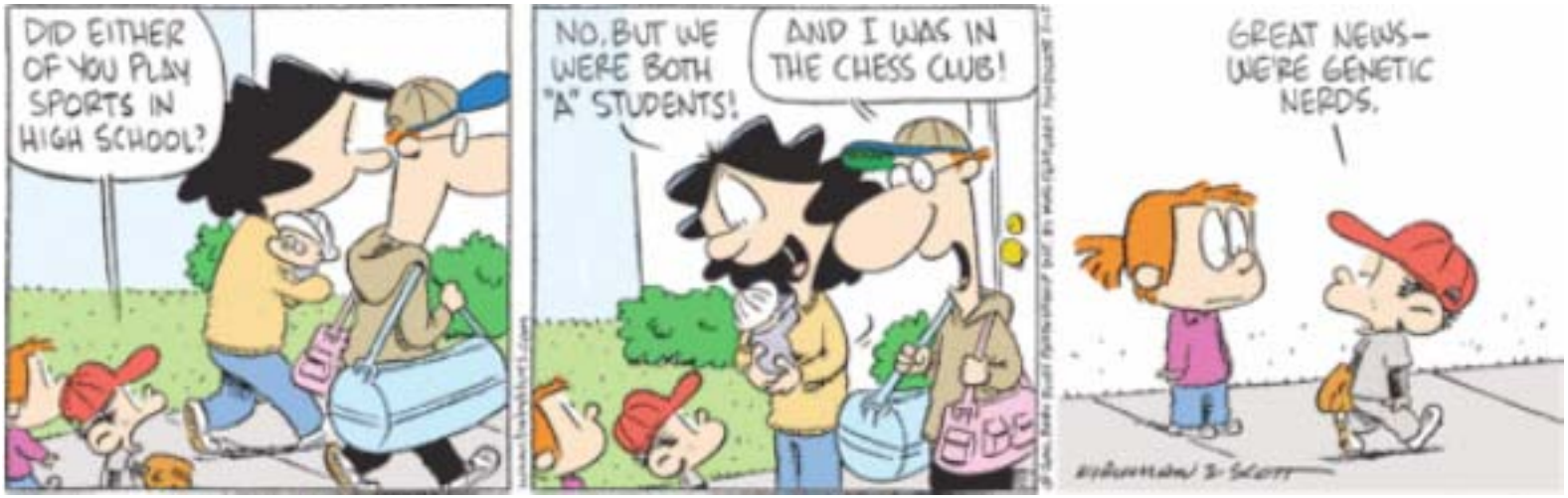

DID EITHER OF YOU PLAY SPORTS IN HIGH SCHOOL?
NO, BUT WE WERE BOTH "A" STUDENTS!
AND I WAS IN THE CHESS CLUB!
GREAT NEWS— WE'RE GENETIC NERDS.

Chromosomes and Microarrays Deciphering the Technology

Angela Scheuerle, M.D.

Tesserae Genetics

Texas BDES

ISCN 2009

An International System for Human Cytogenetic Nomenclature (2009)

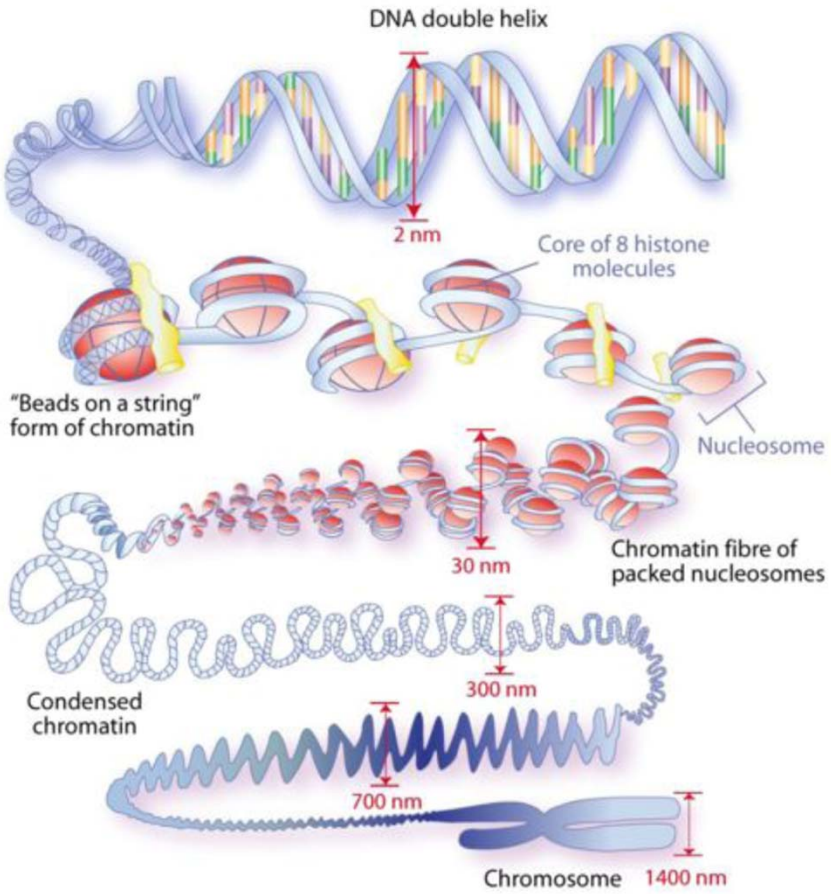
Editors
Lisa G. Shaffer
Marilyn L. Slovak
Lynda J. Campbell

Recommendations of the
International Standing Committee on
Human Cytogenetic Nomenclature

KARGER

Published in collaboration with Cytogenetic and Genome Research

Molecule of DNA



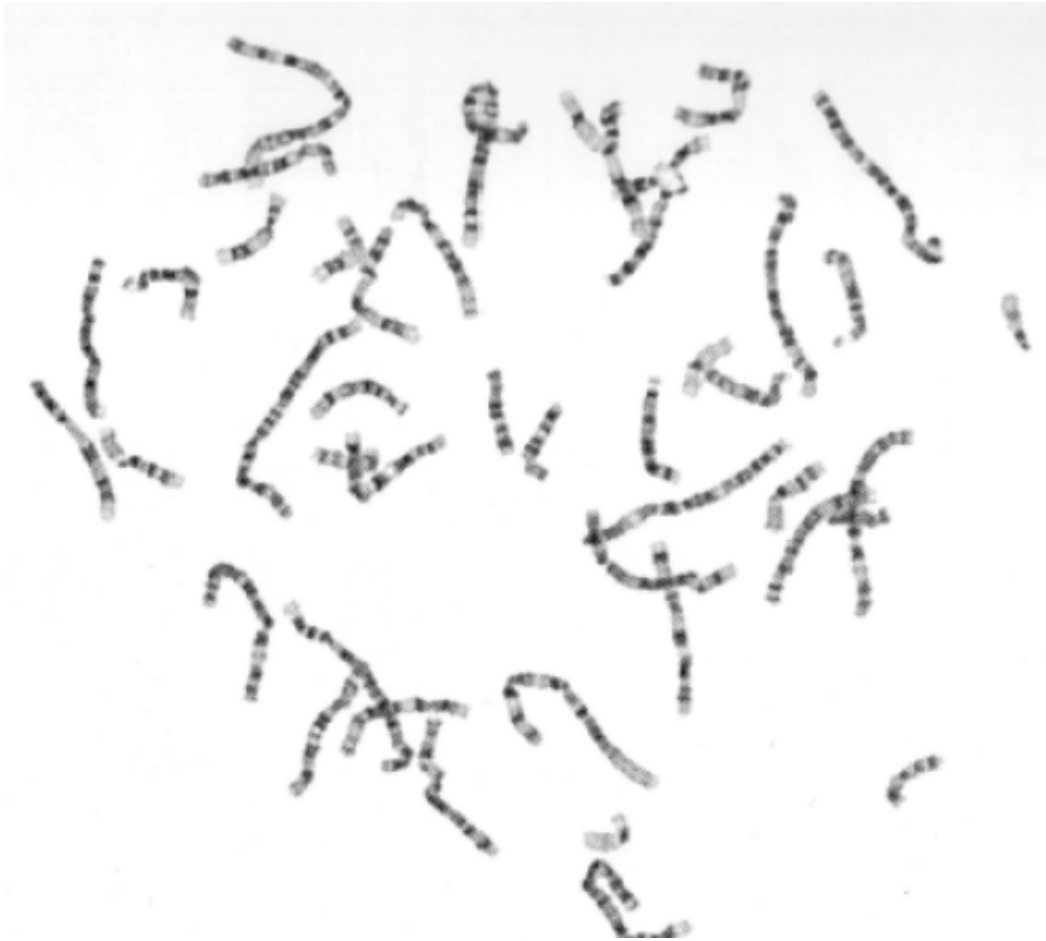
6 Feet of DNA in each Cell

ohyeahdevelopmentalbiology.tumblr.com

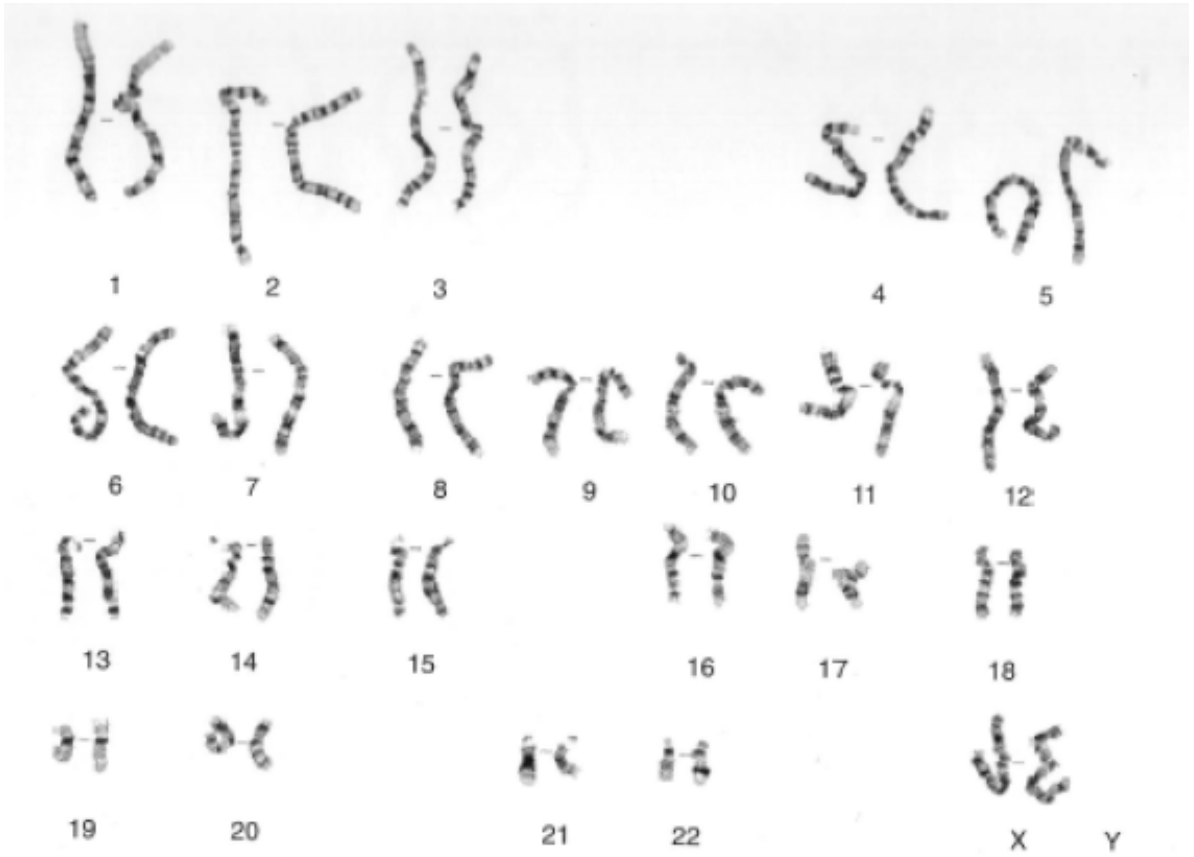
Chromosome

- One huge molecule of DNA
- 100s – 1000s of genes on each
- Visible under a regular microscope
- To be big enough to see, a change must be 4 million bases in size or greater (≥4Mb)
  - Roughly the size of a band on the chromosome

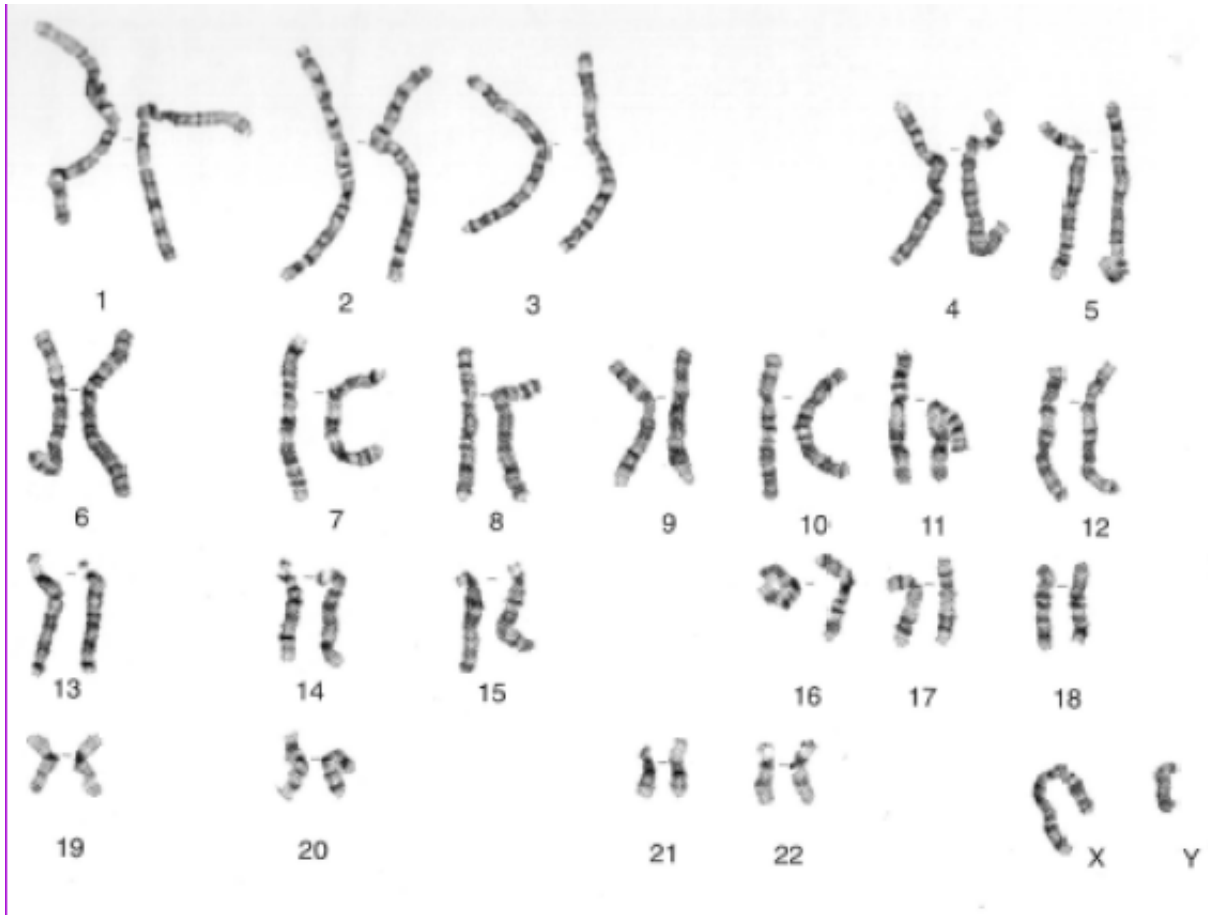
Chromosomes



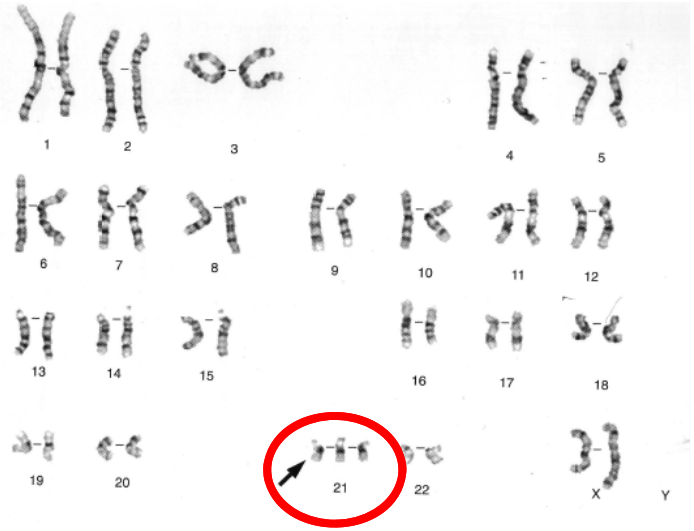
46,XX



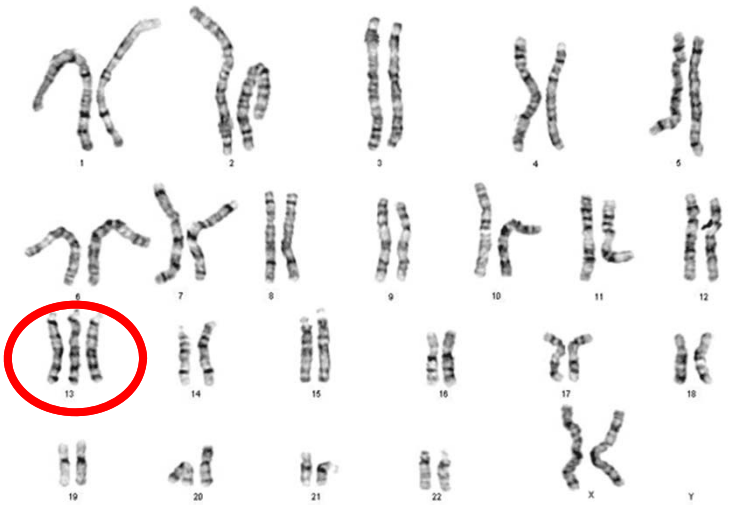
46,XY



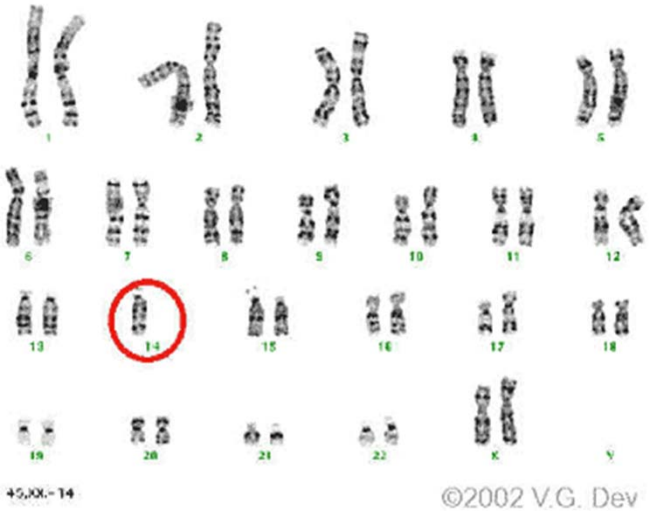
47,XX,+21



47,XX,+13

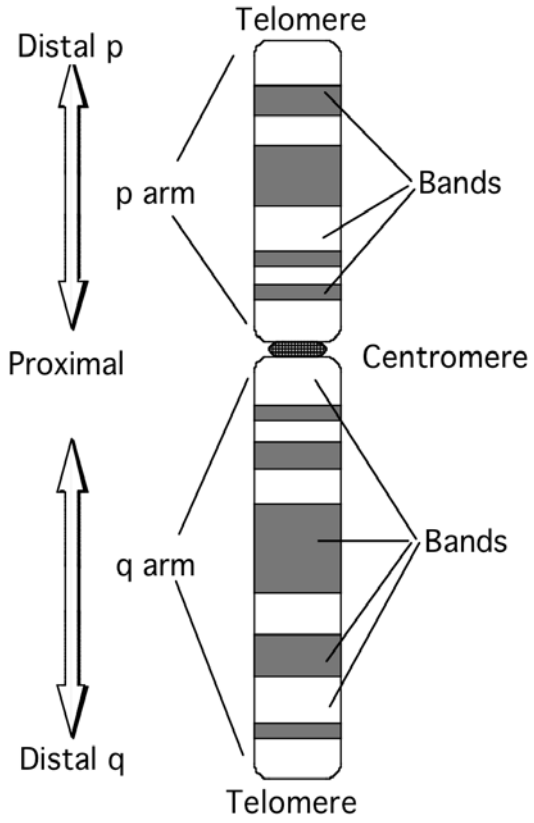


45,XX,-14



45,X

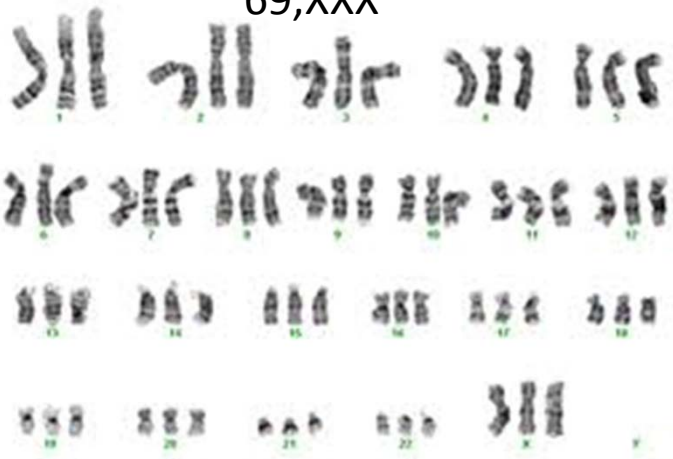
Chromosomes



46,XY,del(5)p

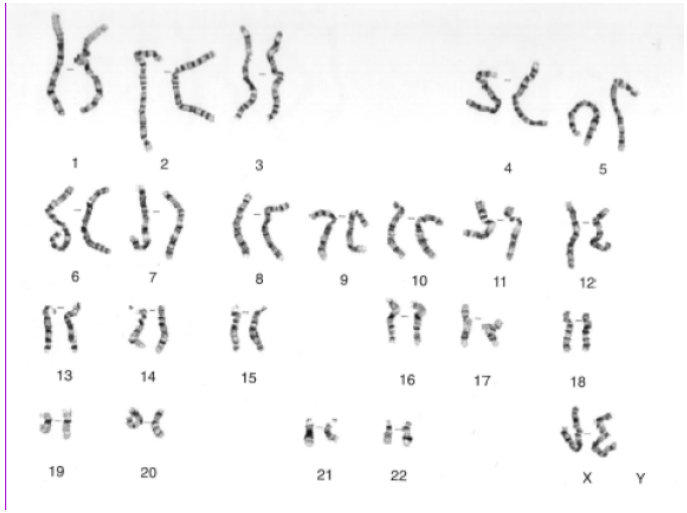
45,XX,t(14;21) or 45,XX,rob(14;21)

69,XXX



Airplane Analogy

Looking at the genetic material like this



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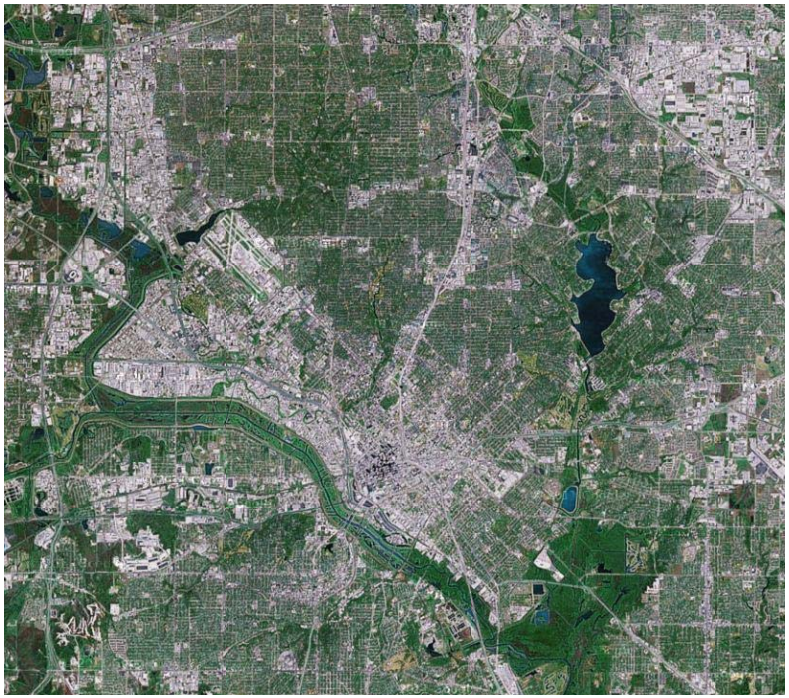
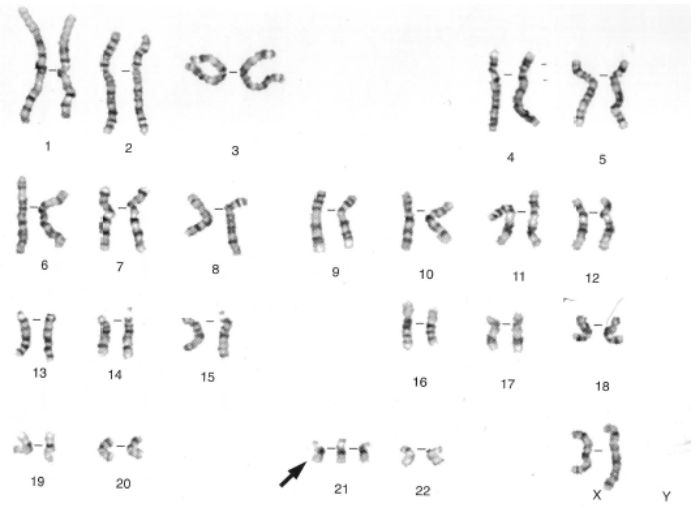
Looking at a city like this



From an Airplane

Having an extra chromosome



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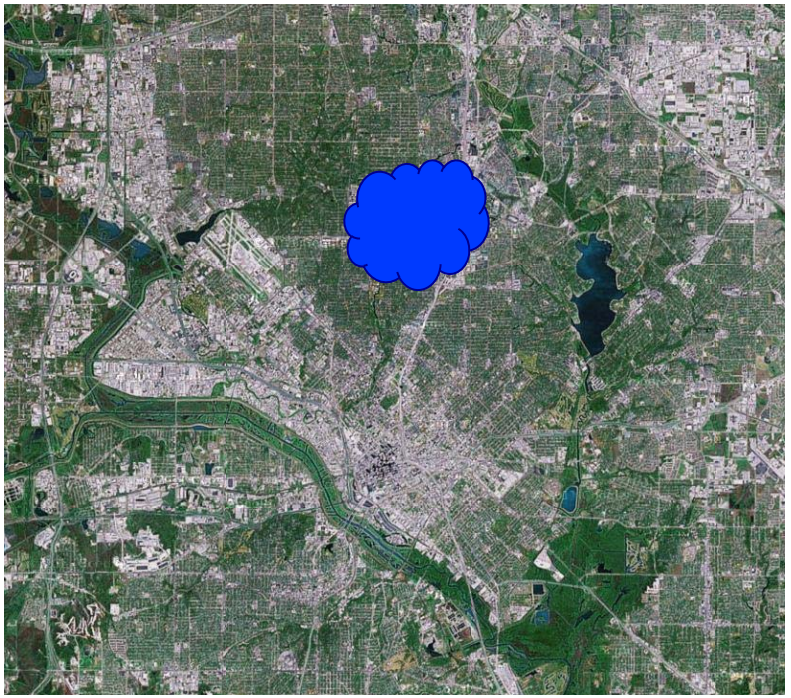
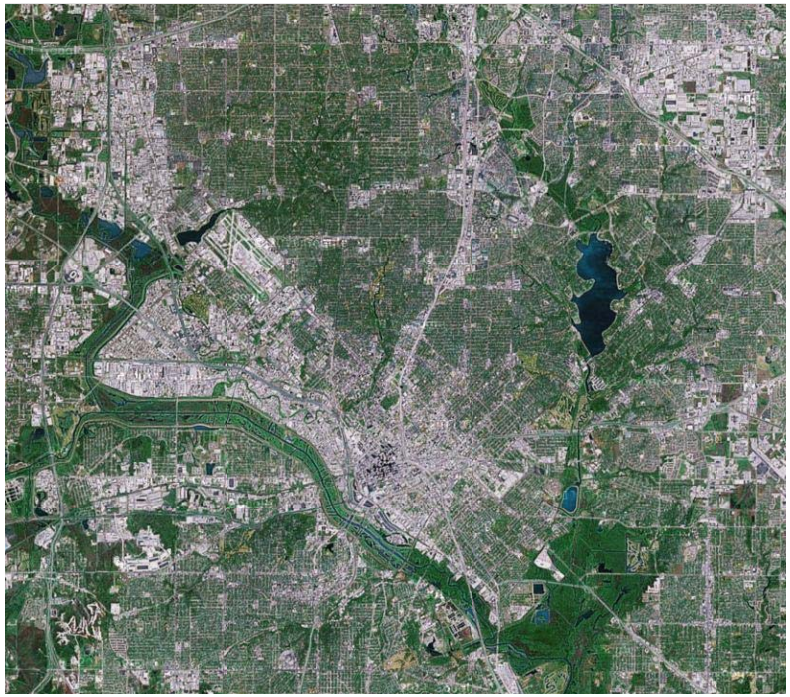
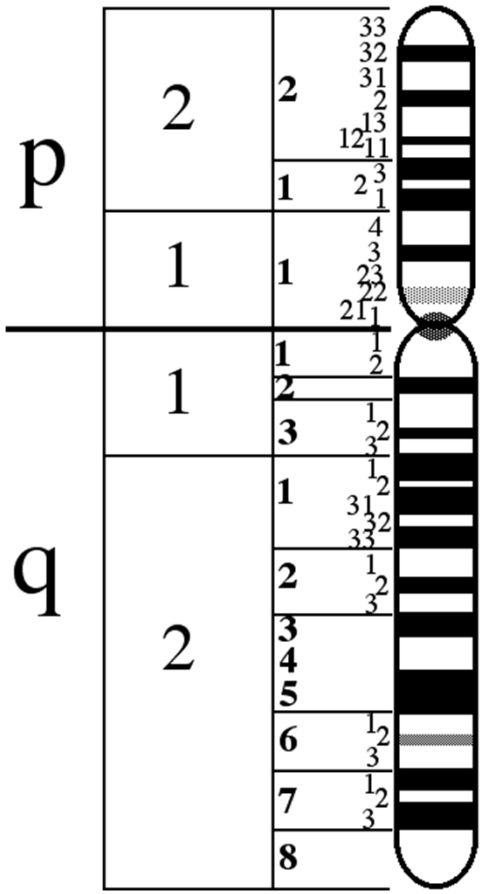
Adding a lake



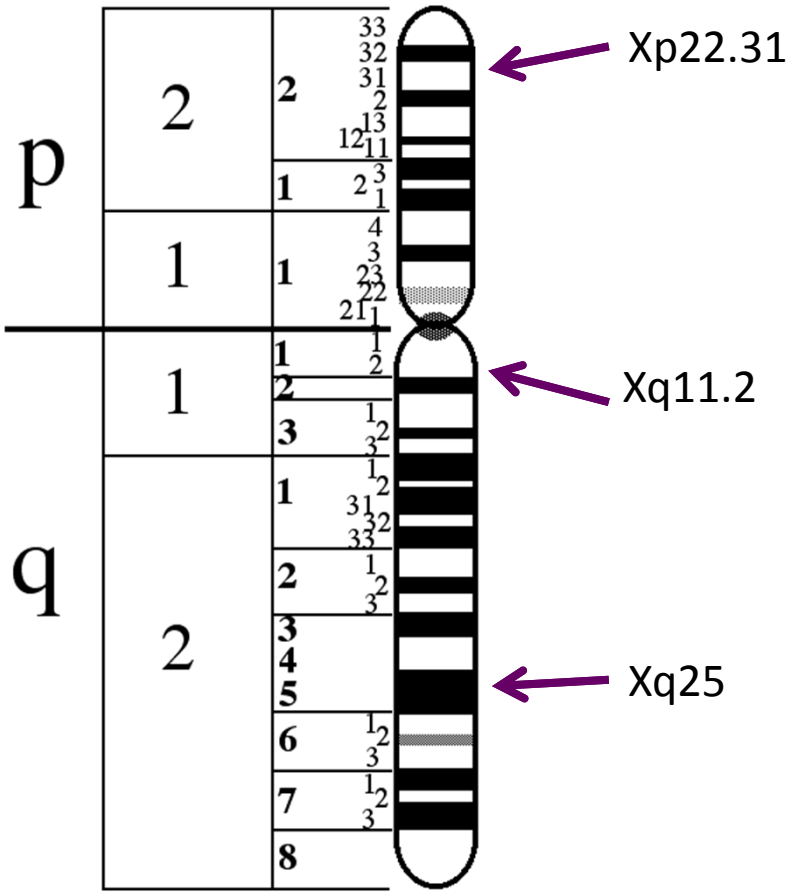
What about a particular address?

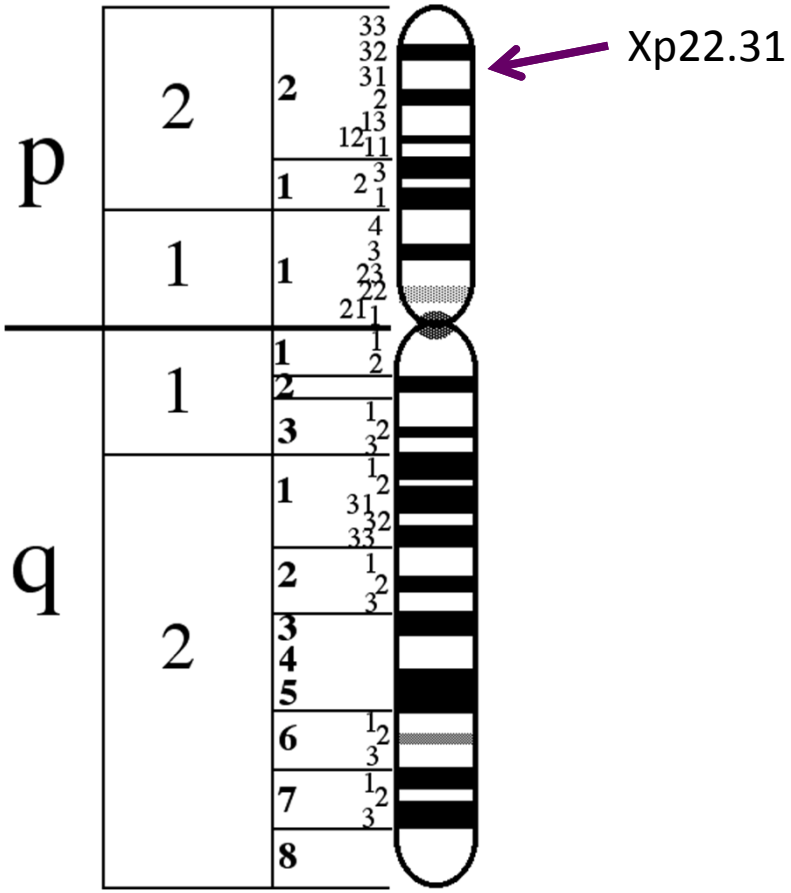


Chromosomes



Chromosomes





9702 Vinewood

What is the correct reading?

22q11.21

A) Twenty-two q eleven point twenty one
B) Two two q one one point two one
C) Twenty-two q one one point two one
D) Two two q eleven point twenty one

What is the correct reading?

22q11.21

A) Twenty-two q eleven point twenty one

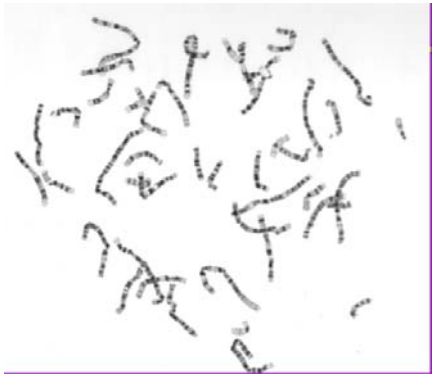
B) Two two q one one point two one

Twenty-two q one one point two one

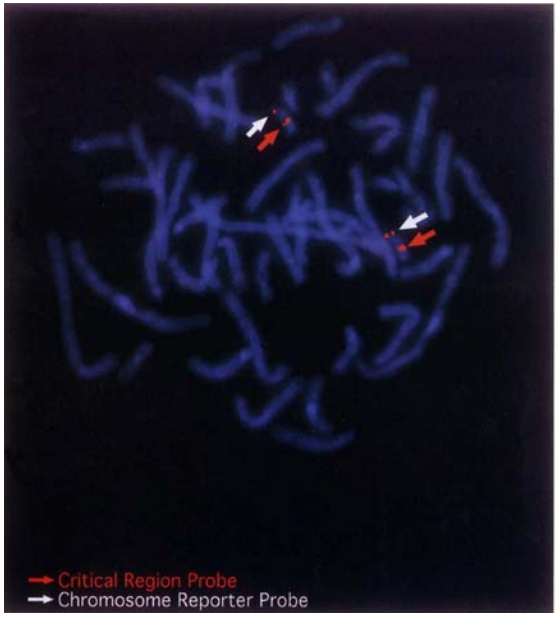
D) Two two q eleven point twenty one

FISH

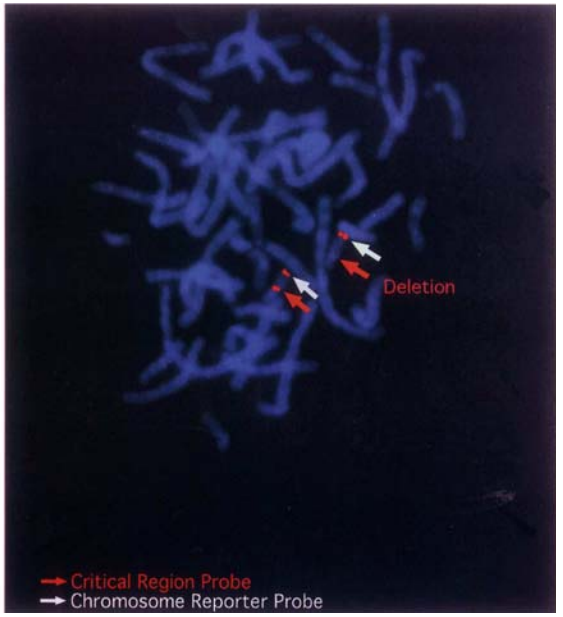
- Will tell you what is going on at a particular address
- Won't tell you what is going on anywhere else.
- Limited usefulness these days as a first-line test
  - You will see it less and less over time
  - Used as confirmation in microarray testing



FISH

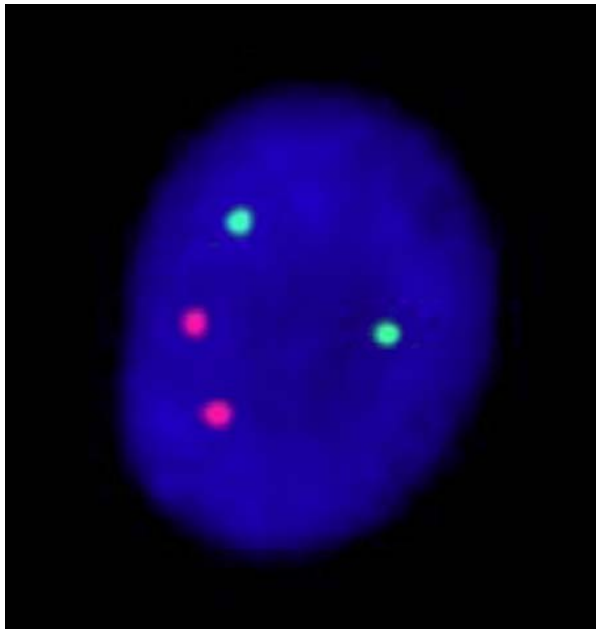


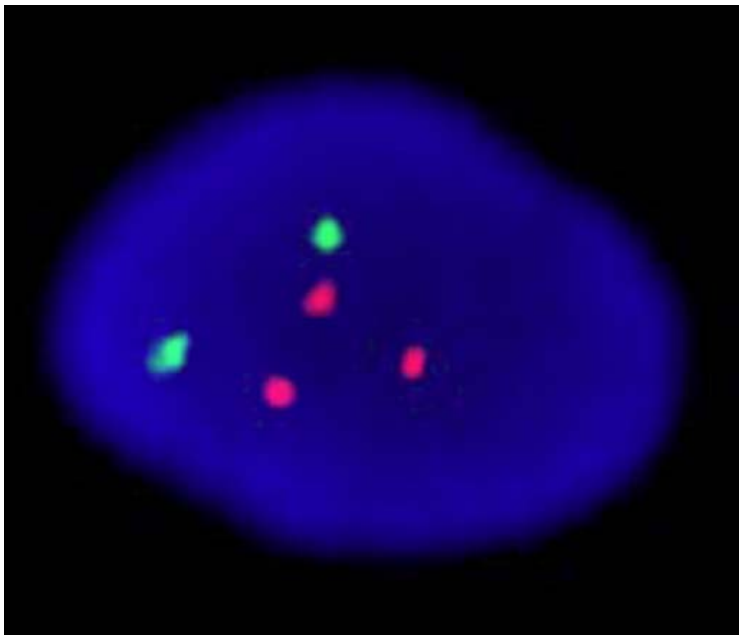
46,XX,ish(22)(q11.2)



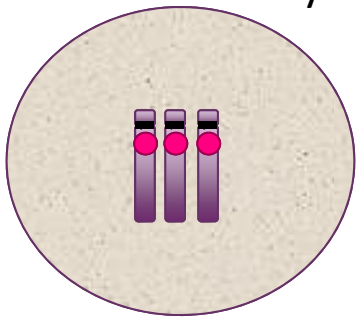
46,XX,ish del(22)(q11.2q11.2)

Interphase FISH

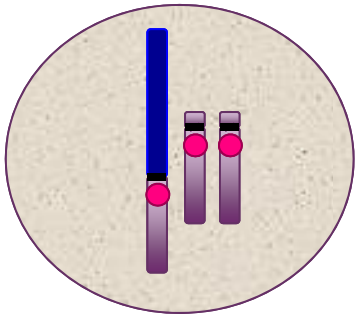




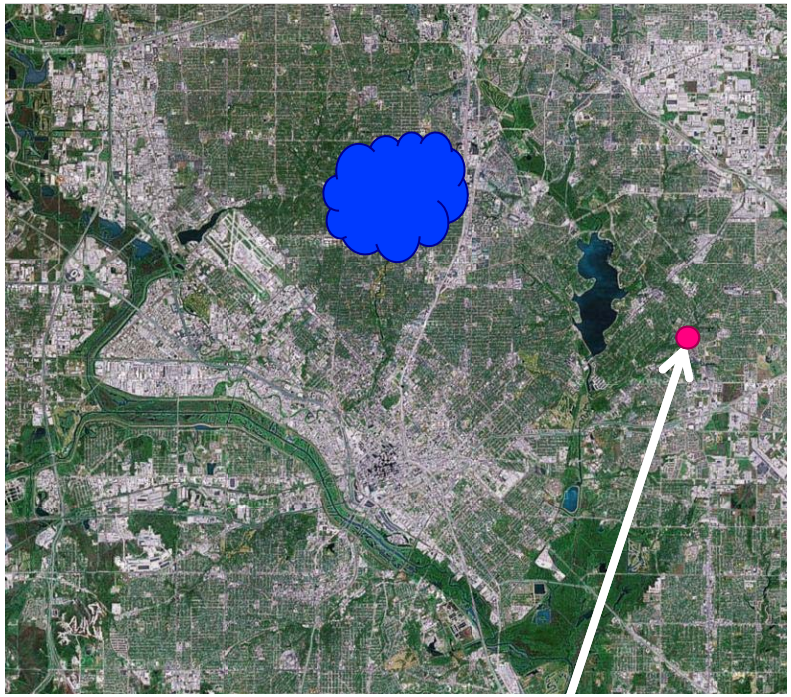
“Free” Trisomy



Translocation Trisomy



Missing the Lake



9702 Vinewood

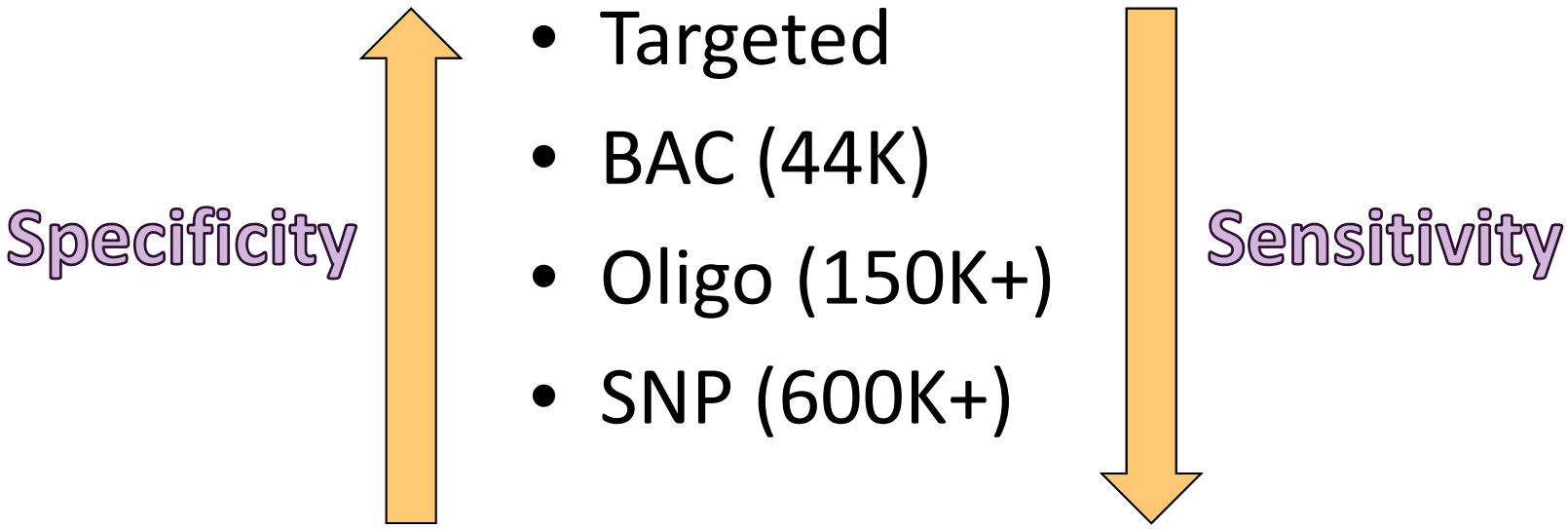
Cytogenomic Microarray

- CMA
  - Cytogenetic Microarray Analysis
- aCGH
  - Array Comparative Genome Hybridization
- Variety of proprietary names
  - Affy, Nimblegen, Roche, etc.
  - Generally identifying the company making the physical hardware

Cytogenomic Microarray

- What it will tell you
  - Submicroscopic additions and deletions
  - Like doing FISH on 200,000 (or more) places
  - SNP arrays will tell about chromosome inheritance
- What it won't tell you
  - Single gene mutations
  - Balanced translocations

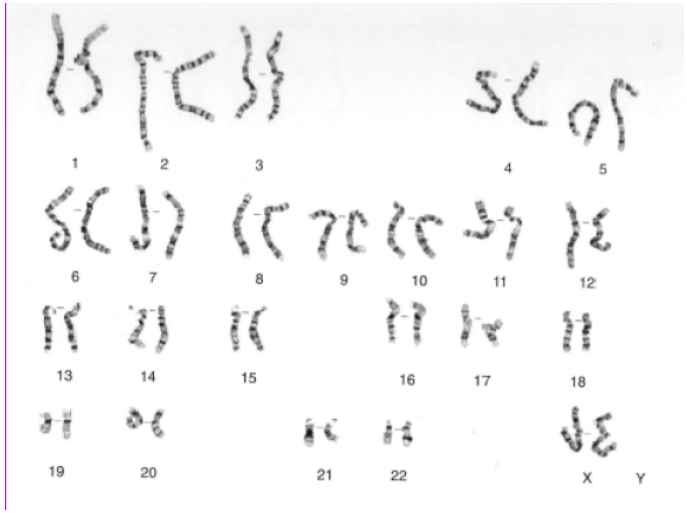
Cytogenomic Microarray



- Targeted
- BAC (44K)
- Oligo (150K+)
- SNP (600K+)

Airplane Analogy

Looking at the genetic material like this



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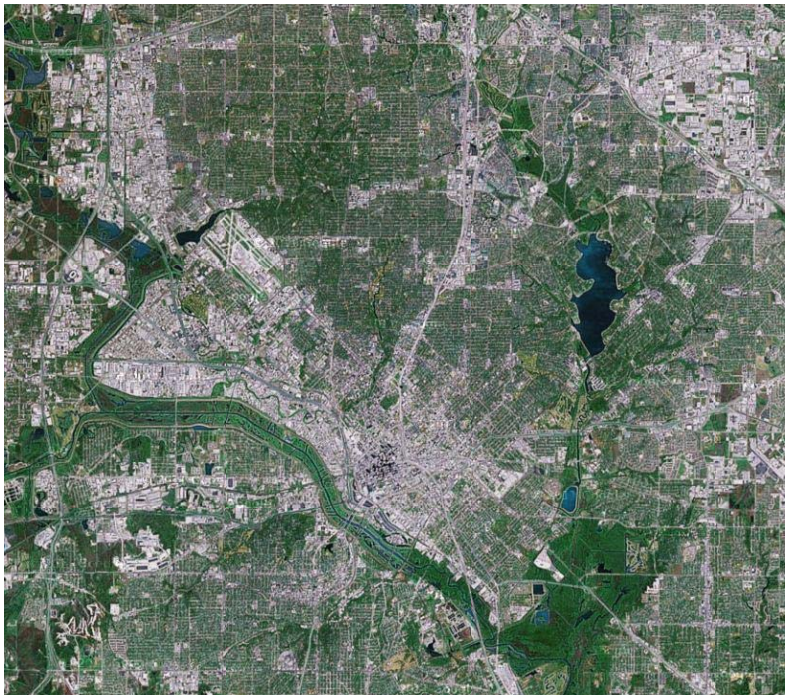
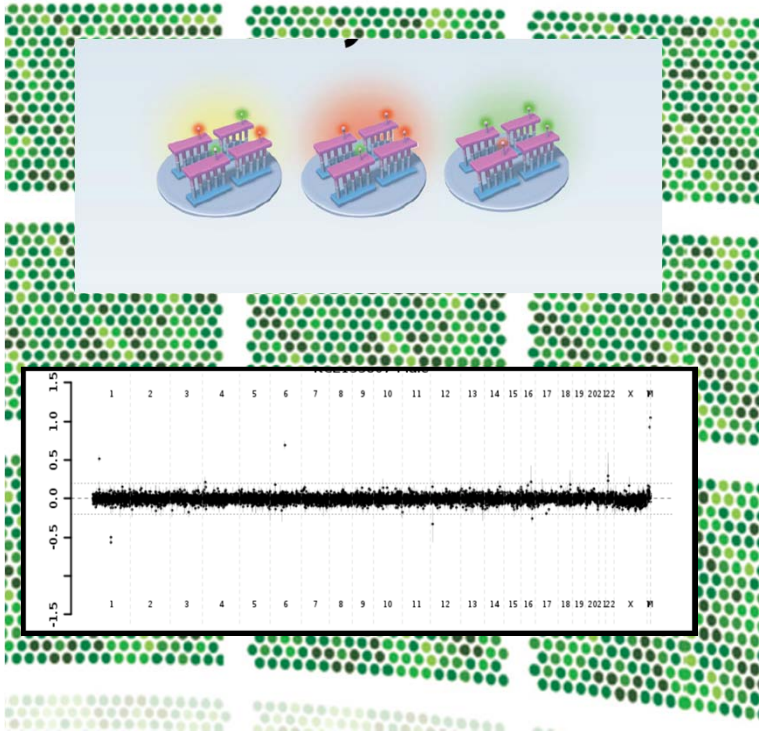
Looking at a city like this

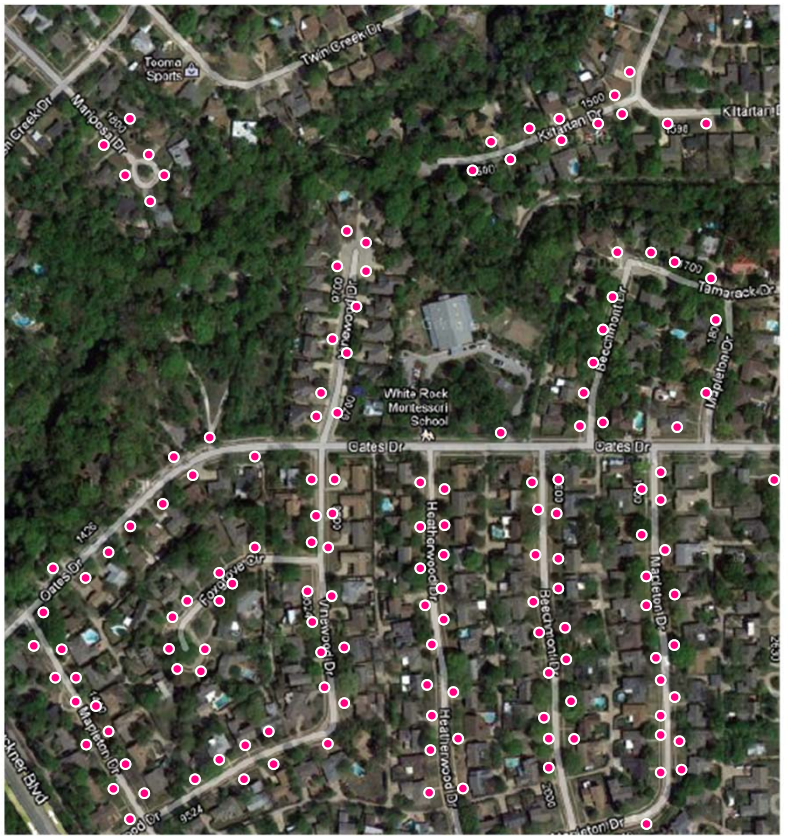


Mailman

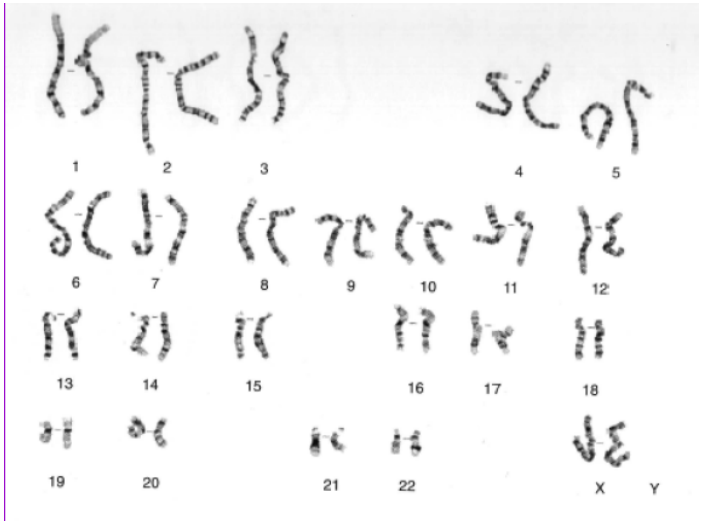
Doing aCGH

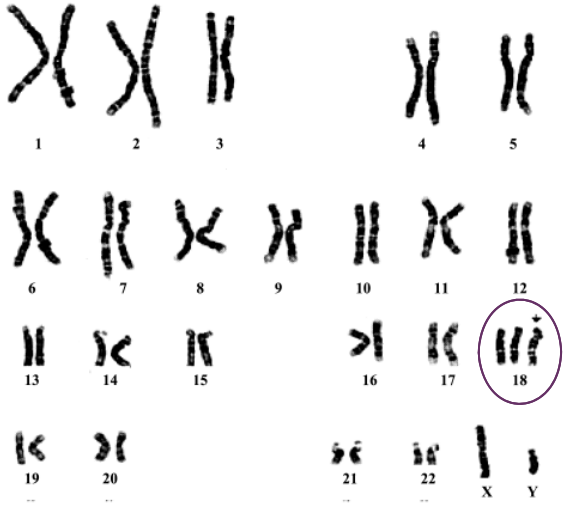


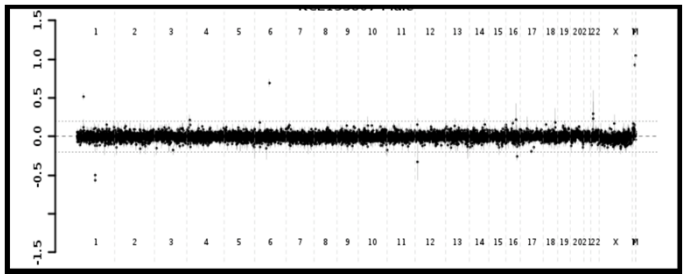
Knowing what mailboxes have mail in them

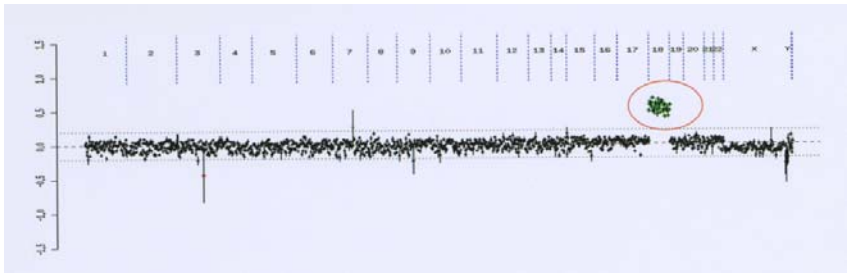


Array

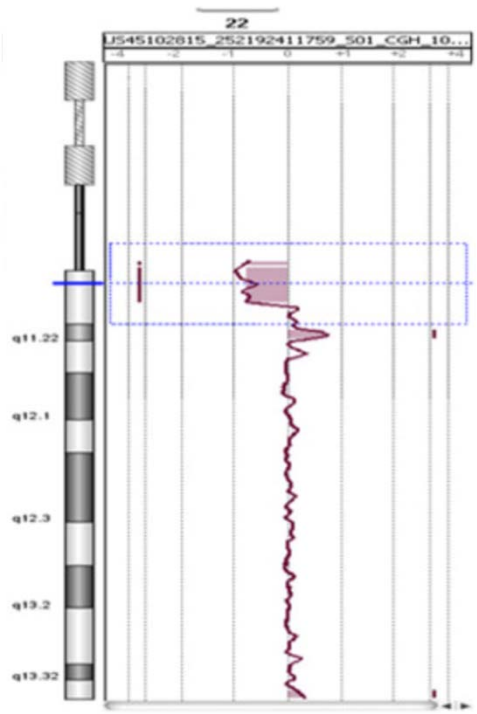




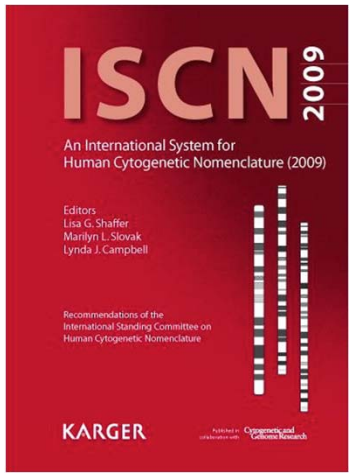




Deletion 22q11.21



Nomenclature



Indication	Abbreviation(s)
FISH was done	ish
Microarray was done	arr
Extra of something	“+”, “x”, add, dup, mar
Something missing	“-“, del, r
Rearrangement	der, ins, inv, rob, t

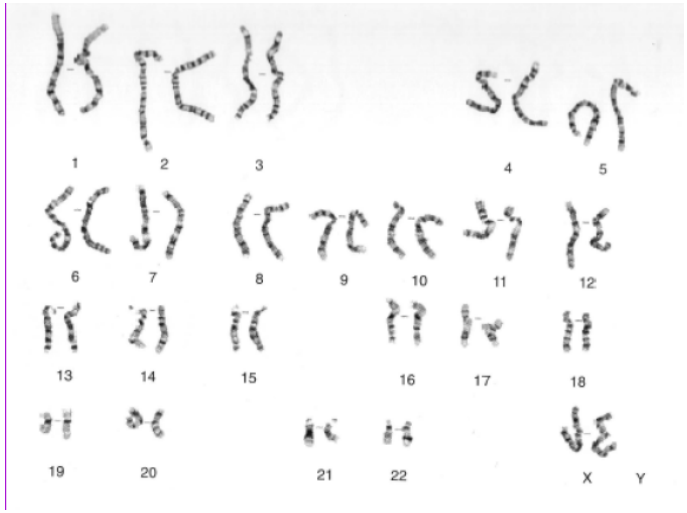
Normal v Abnormal

Technology	Normal Result	Abnormal Result
Routine Kayrotype	46,XX or 46,XY	46,XX,del(22)(q11.2)
FISH	46,XY,ish(22)(q11.2)	46,XY,ish del (22)(q11.2q11.2)
Microarray	Arr (1-22,X)x2	Arr (22)(q11.21q11.21)x1

One Last Analogy

Airplane Analogy

Looking at the genetic material like this



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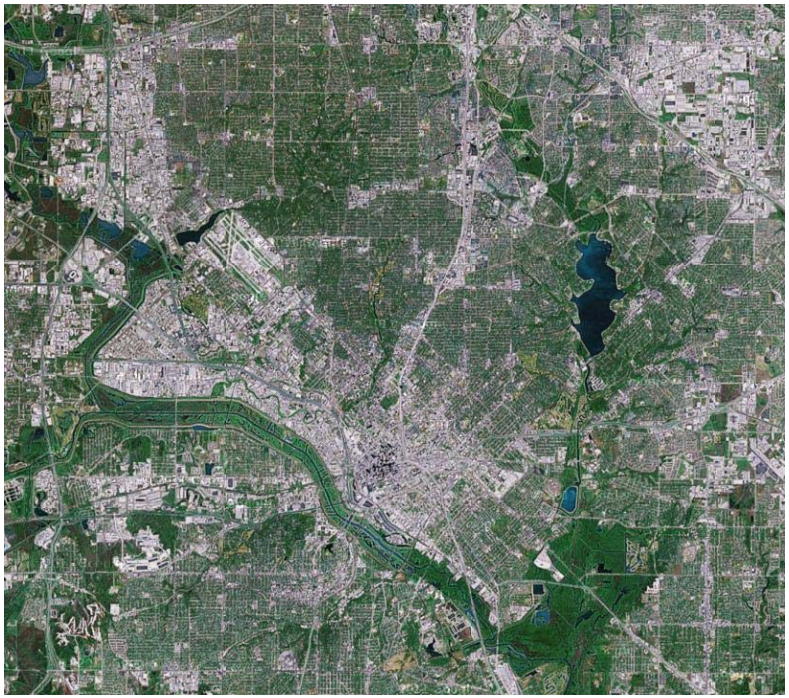
Looking at a city like this



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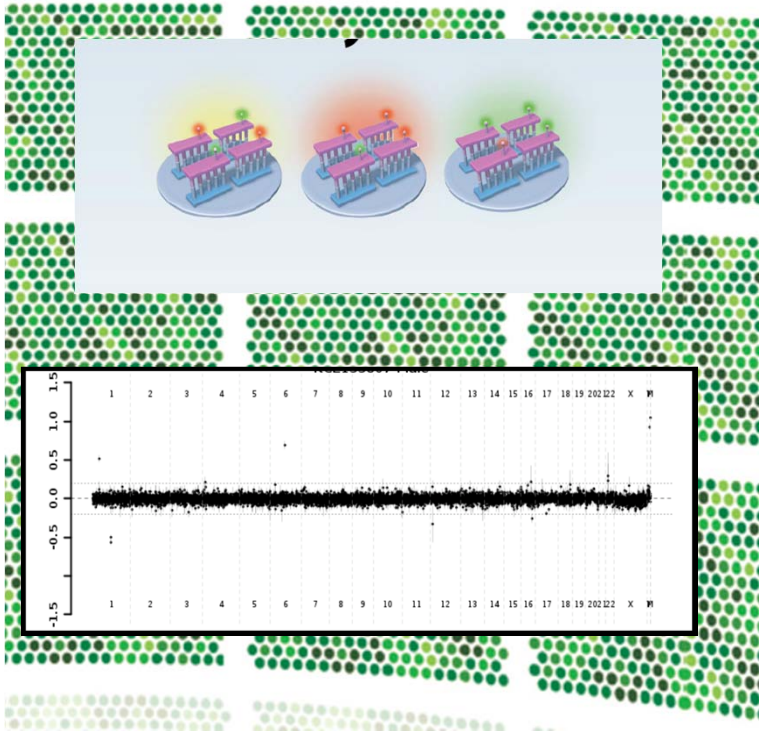

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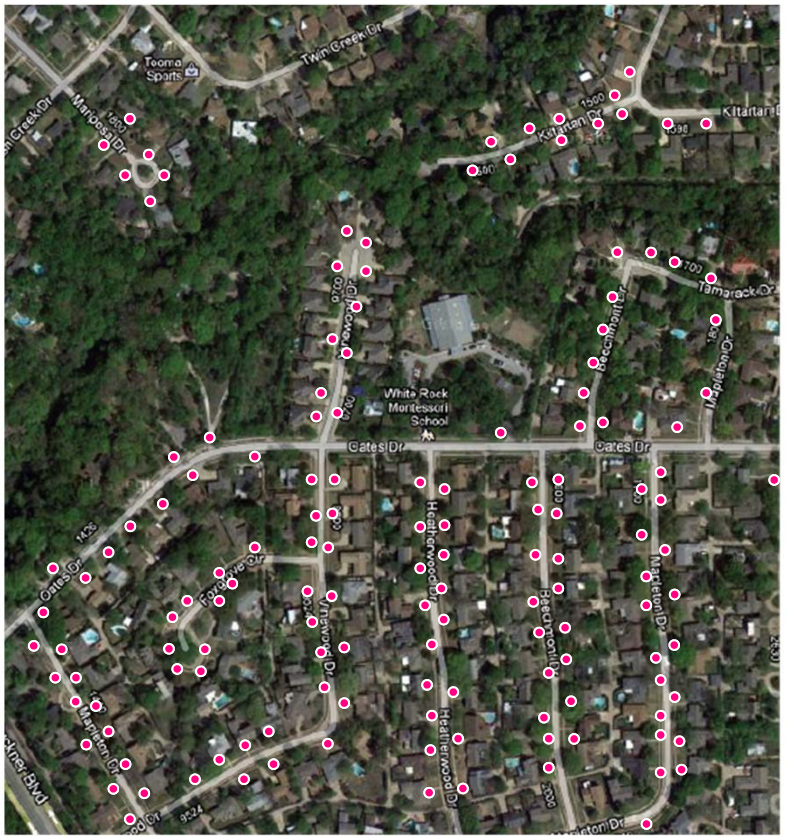
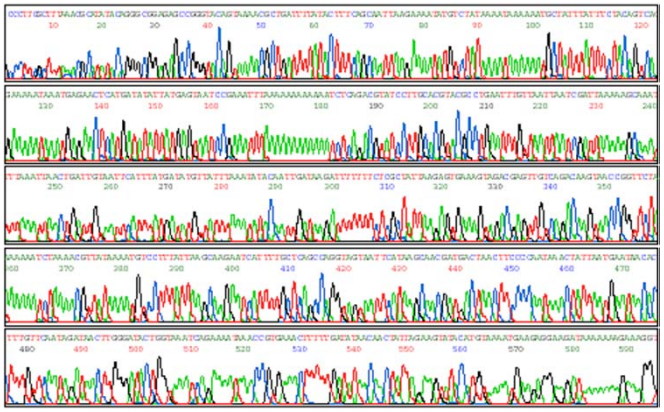
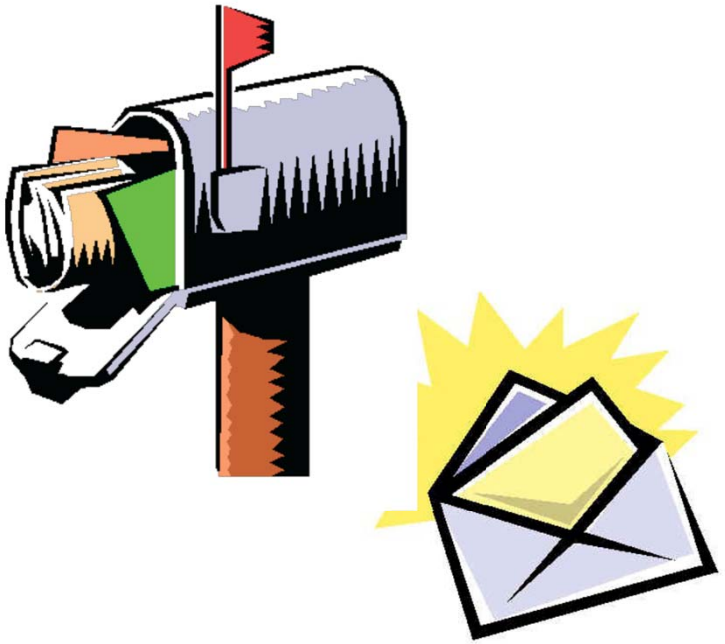


Water Bill

Looking for a mutation
in a single gene



Is like seeing how much I owe
on my water bill



MS. TENA
Reader Advisor
MADAM ROSA
GENETICIST
NICK DOWNES