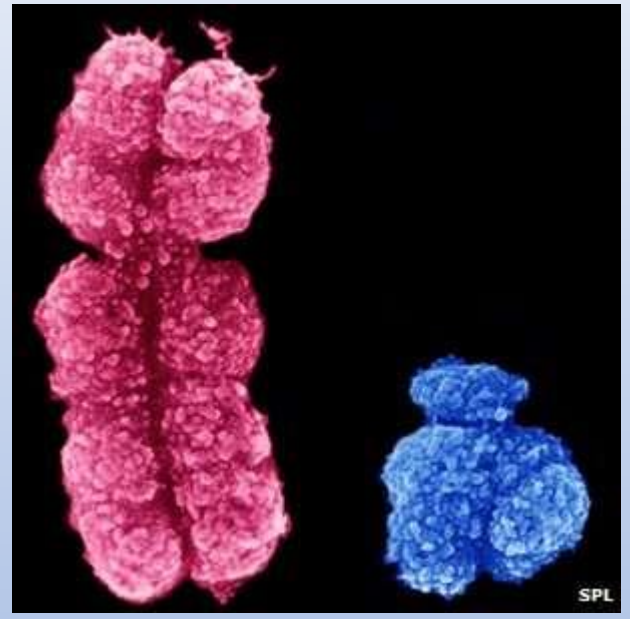
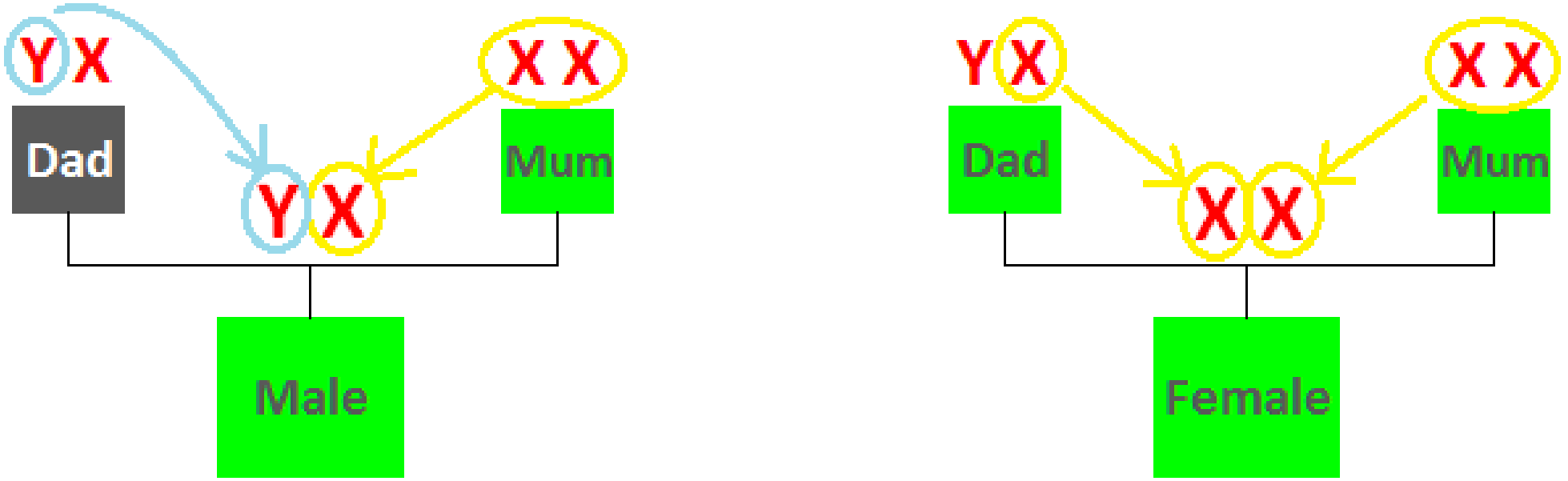
The Complicated and Unruly X Chromosome

Actual Images of the Two Sex Chromosomes
The Bix X and the Little y



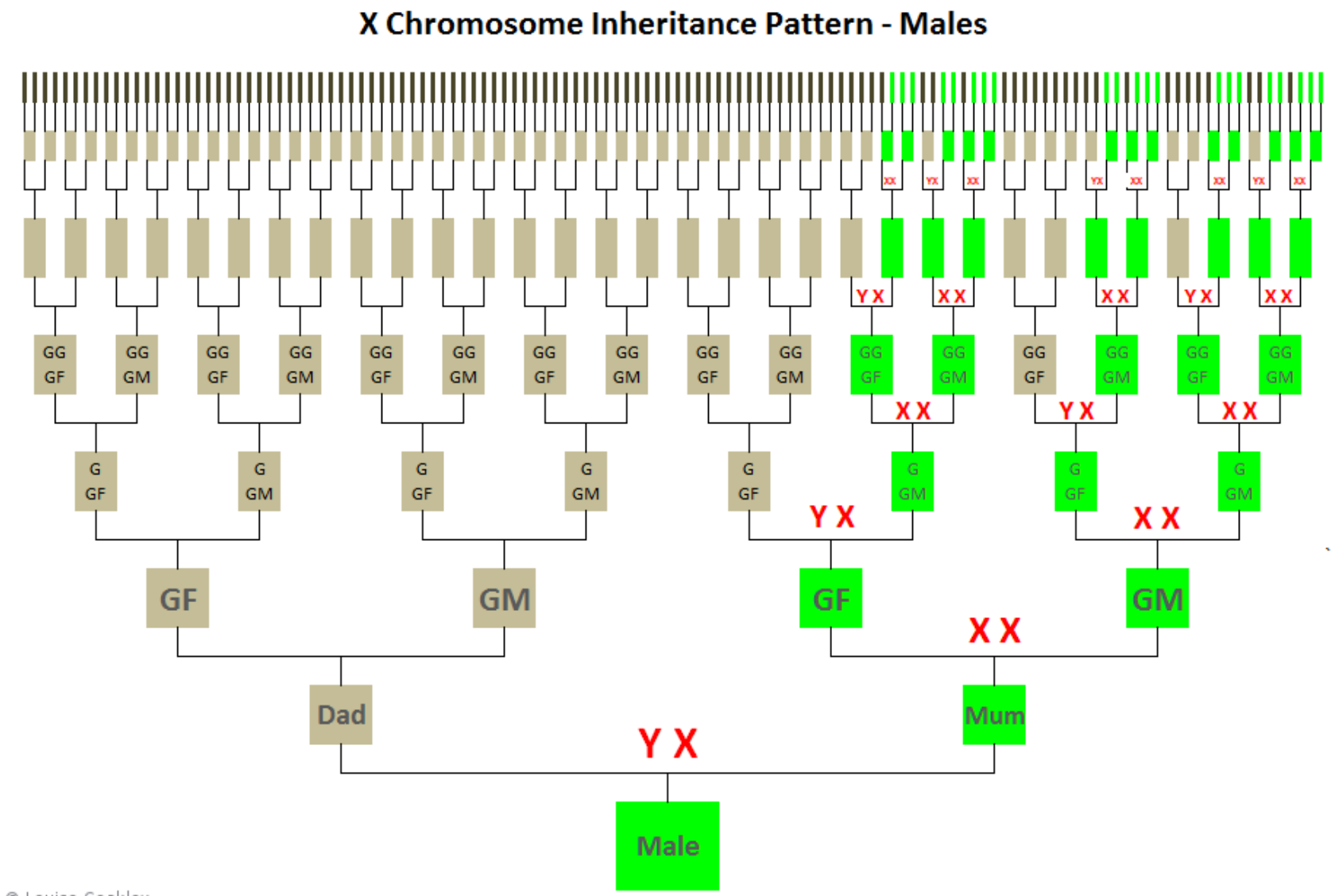
Scientist believe the Y Chromosome is getting smaller over time. What do you think that means?

X Inheritance Pattern



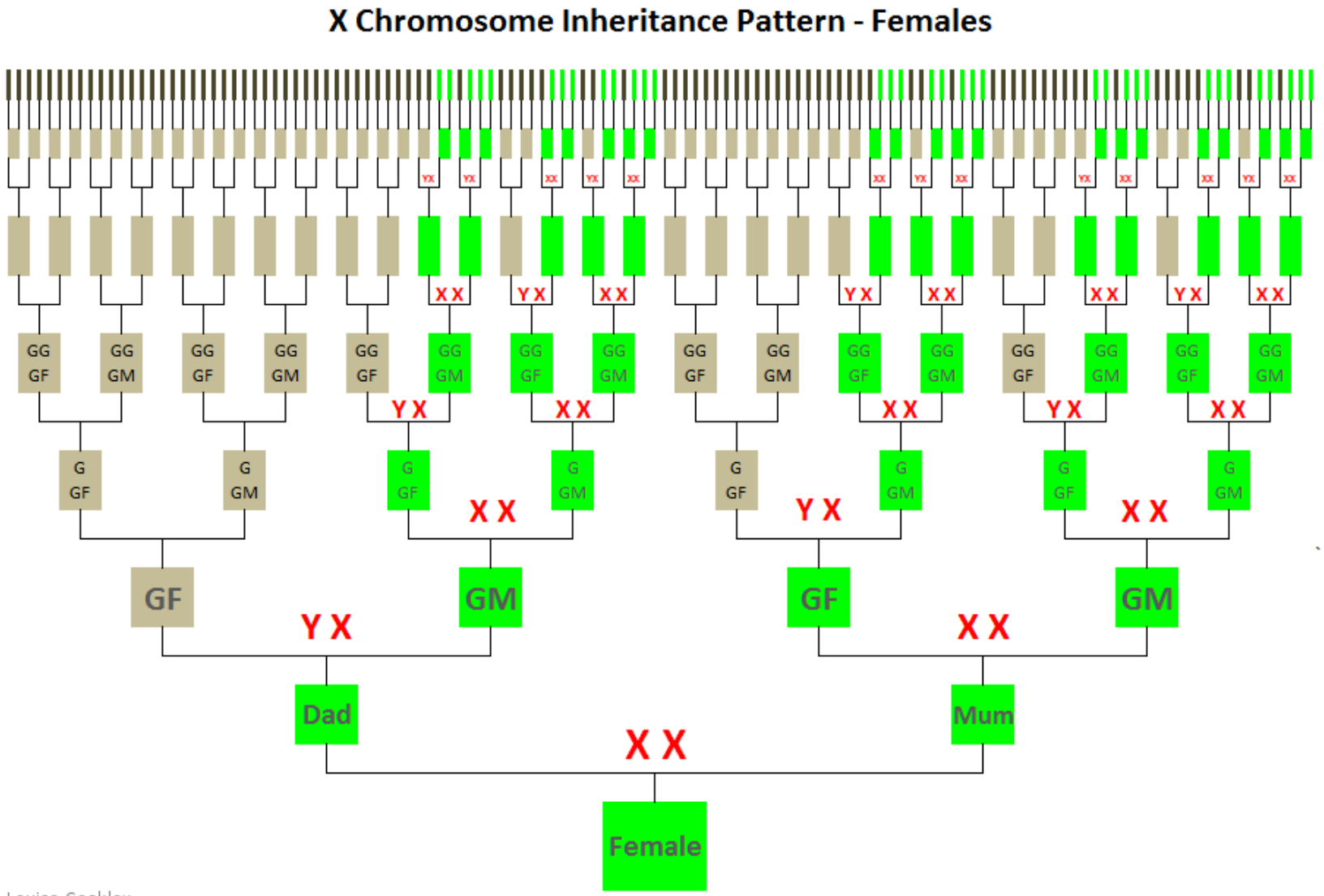
Note there is no X history transferred from the left side. That is because the Dad only passed on his Y Chr. to his son.

If Dad's GF and GM did not have a daughter, all the info for the the X chromosome is lost forever. At least for this genetic strain



On the other hand, if Mom and Dad have a daughter, Dad's X DNA is passed on and the history the X is continued, but only for GM's branch.

So if a female gets 2 X's, do they get mixed during the transfer like autosomal (1-22) chromosomes?



We could spend the entire class trying to understand this table.

Key points to remember:

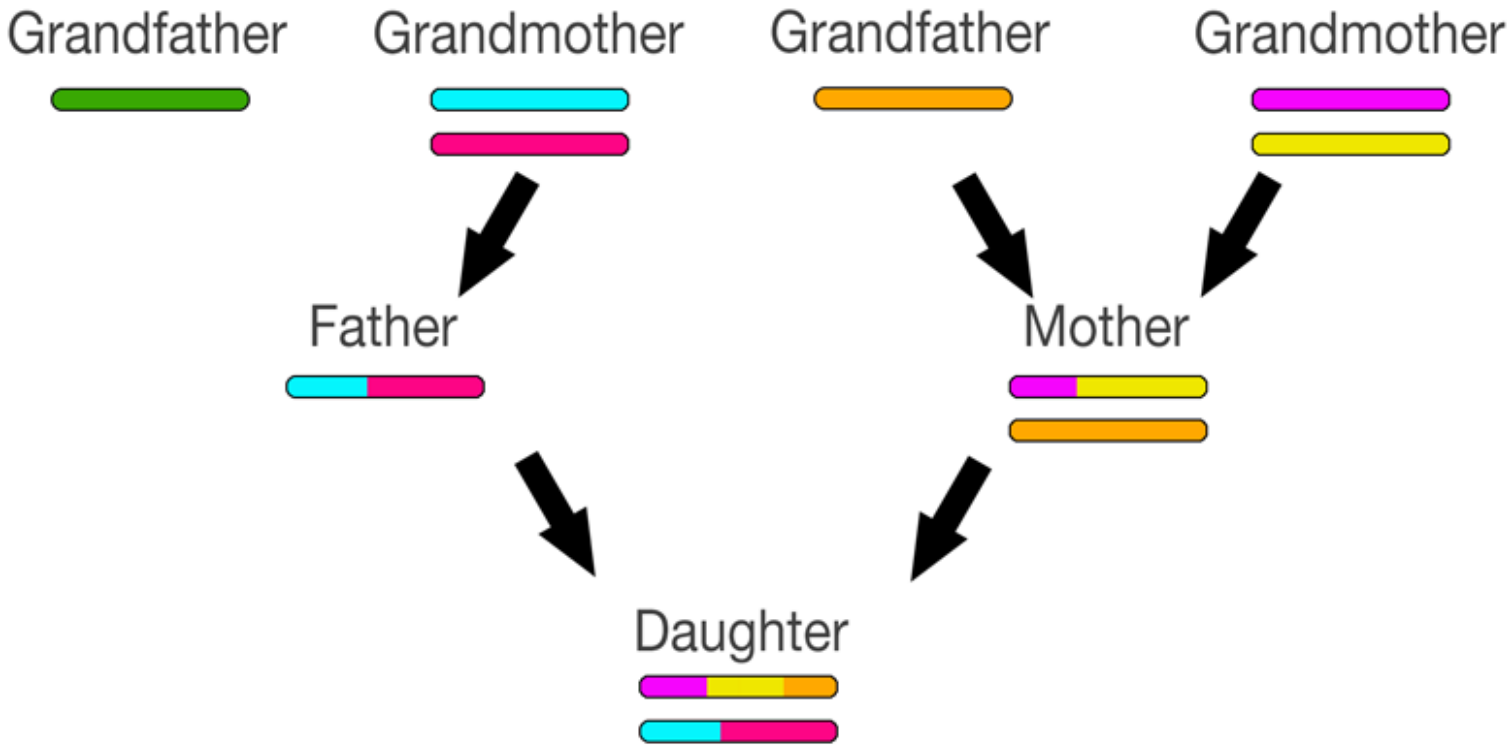
1)Because an X-chromosome is passed exactly from father to daughter, it will remain unchanged for that generation. This means that X chromosomes change less often along father-daughter pedigree lines.

2) The Mom transfers one of her two X chromosome which may or may not have been mixed with her other X chromosome

This is the way 'recombination' is supposed to work

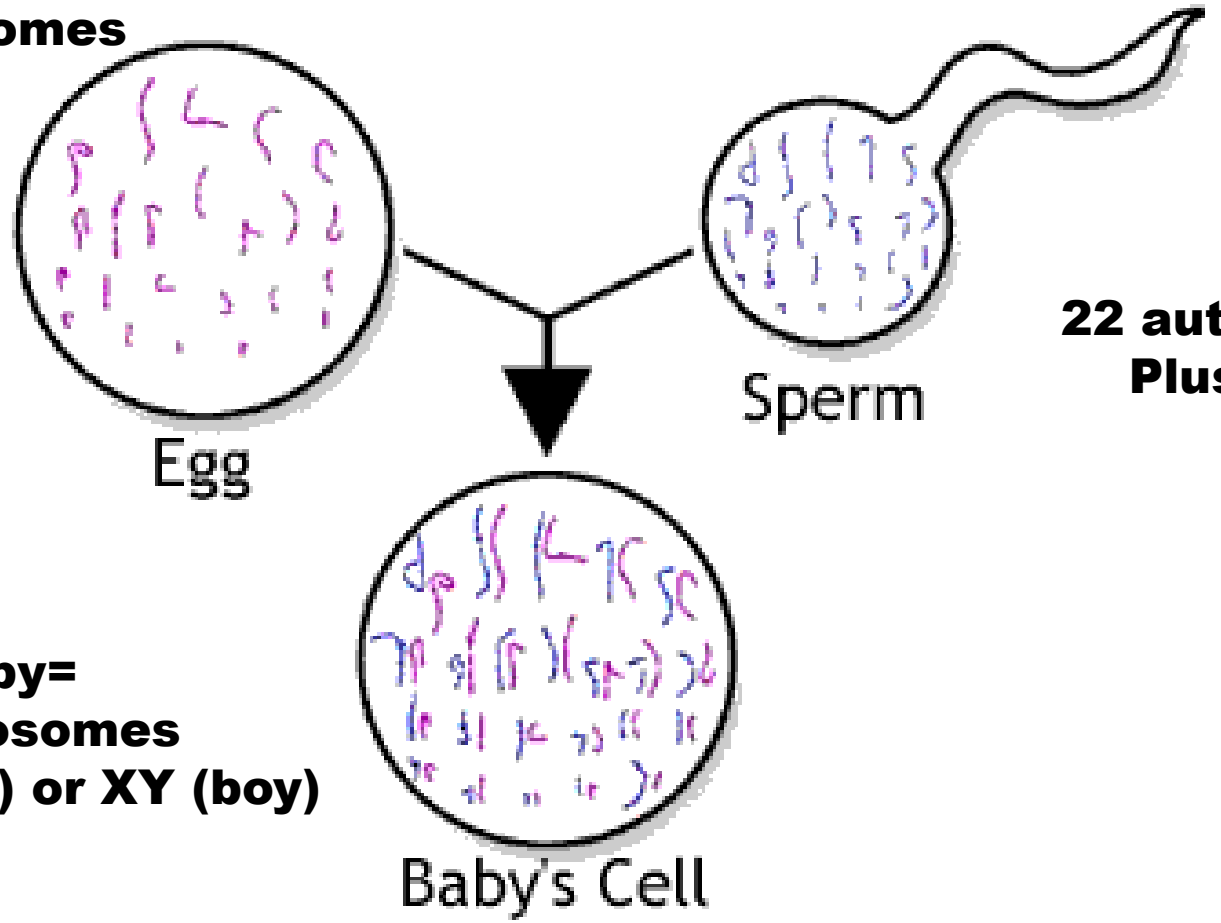
Generation	Average Autosomal %	Avg X Maternal Only (men)	Average X Total (Women)
GGGG-Grandparents	1.56%	12%, 6.25%, 3.125%, 1.56%	6.25%, 3.125%, 1.56%, 0.78%
GGG-grandparents	3.12%	25%, 12.5%, 6.25%	12%, 6.25%, 3.125%, 1.56%
Great-Great-grandparents	6.25%	25% - mother's father's grandparents 25% - mother's mother's grandmother through grandfather 12.5% - mother's mother's grandparents through grandmother	25%, 12.5%, 6.25%
Great-grandparents	12%	50% - mother's father's parents 25% - mother's mother's parents	25% - father's grandparents 25% - mother's father's grandmother 12.5% - mother's mother's parents
Grandparents	25%	50%	50% father's mother 25% - mother's parents
Parents	50%	100%	50%
You	100%	100%	100%

The two X Chromosomes of a female <u>may</u> re-combine when a new egg is made, but <u>*not*</u> the X the father passes on since he has only a single X paired with a Y. There is nothing to re-combine with



This is where it all starts.

Egg=
22 autosome chromosomes
Plus an X



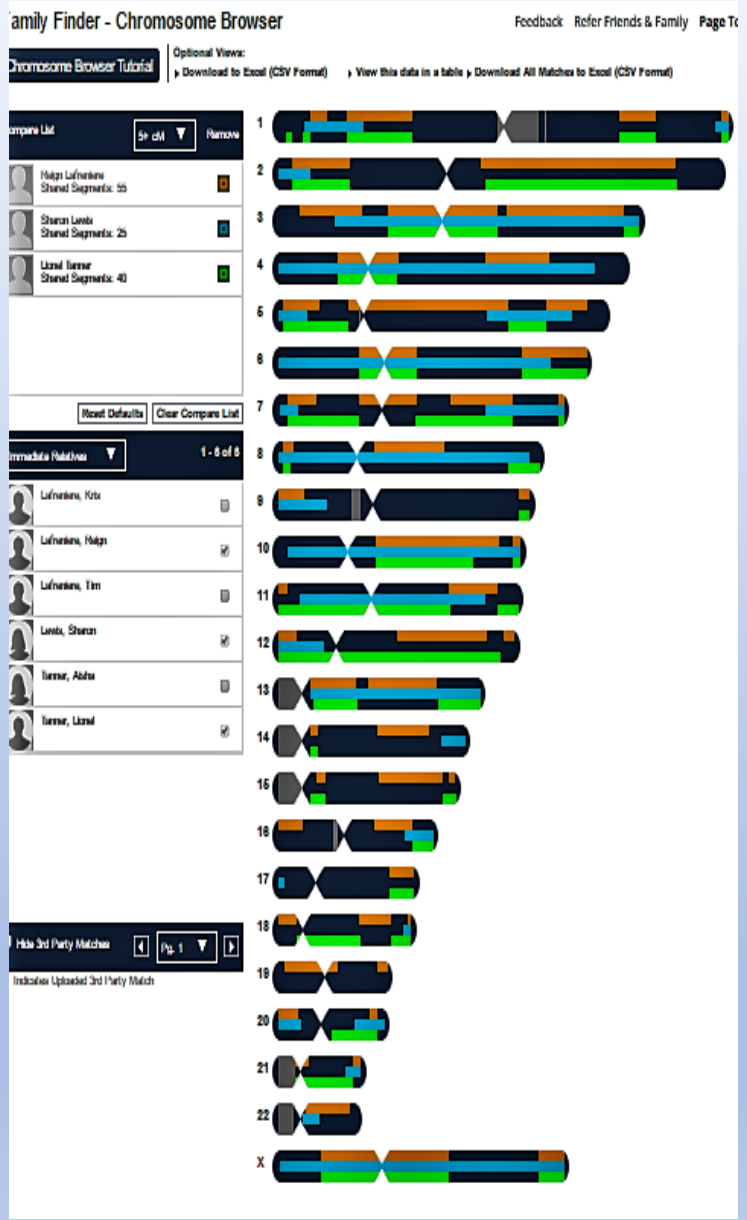
Sperm=
22 autosome chromosomes
Plus either an X or a Y

Baby=
44 autosomes
Plus XX (girl) or XY (boy)

DNA Analyzed for Youngest Son's Family

Maternal Side		Paternal Side	
Grandfather – L.T			Grandmother- S.L
Mom – A.T.			Dad– T.L
Son – R.T. Prev. Marriage	Daughter P.F		

To reduce clutter and increase clarity not all DNA results included



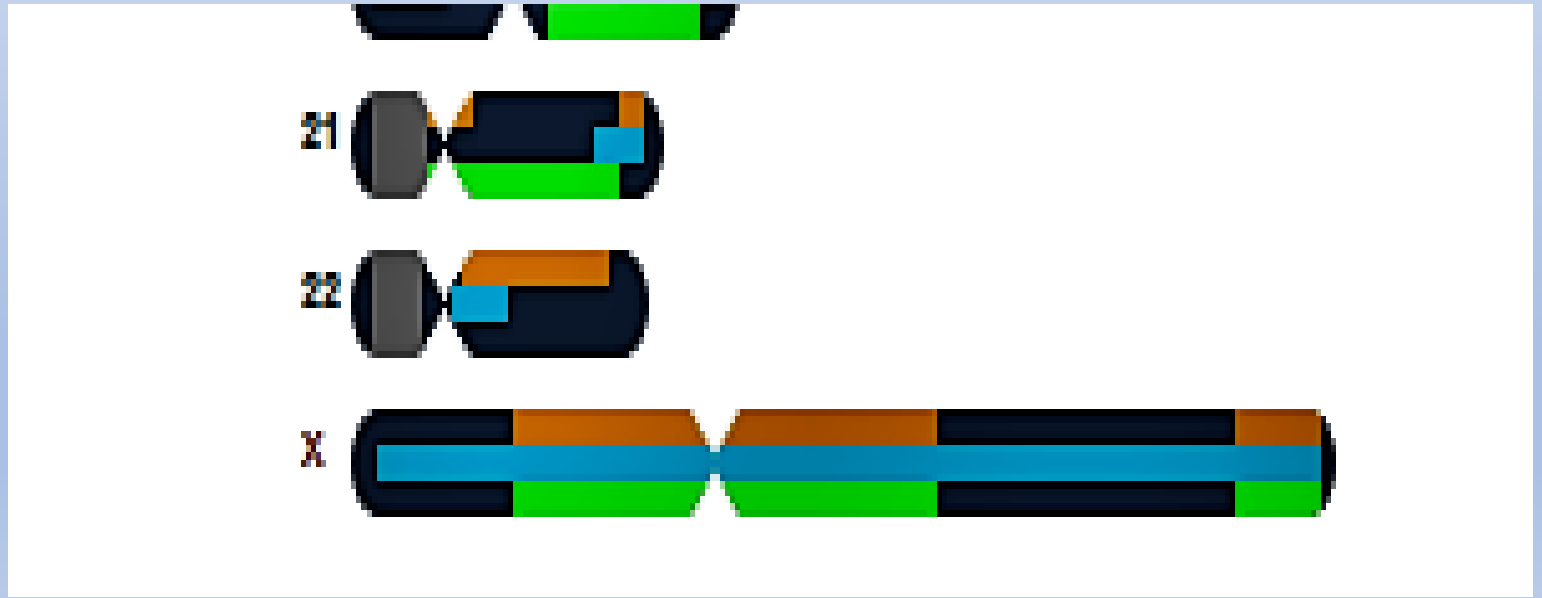
So how did my Grand Daughter's (P.L.) DNA get mixed?

FTDNA Chromosome Browser
(colored bar indicates a DNA match in each Chromosome)

Gold - Half Brother – R.L. (total match = 24%)

Green – Maternal Grandfather – L.T. (total match =21%)

Blue – Paternal Grandmother – S.L. (total match = 24%)
(Note the Blue 100% match on the X Chromosome)



X Chromosome Inheritance

Grandmother (GM) to Granddaughter (GD)

The Blue at the bottom of the image indicates where a “match” occurs between GD and GM. In this case it is a 100% match. But note the top is primarily Yellow. That indicates a “half match”. That is because GD gets two X’s. The other would always be a 100% with her mother.

So this is common

GEDmatch.Com X-DNA Comparison - V2.1.0(a)

Base Pairs with Full Match =
Base Pairs with Half Match =
Base Pairs with No-call =
Base Pairs with No Match =
Base Pairs not included in comparison =

Matching segments greater than 7 centiMorgans =

Comparing Kit M219936 (Sharon Lewis)(F) and T527989 (Phoenix Lafreniere)(F)

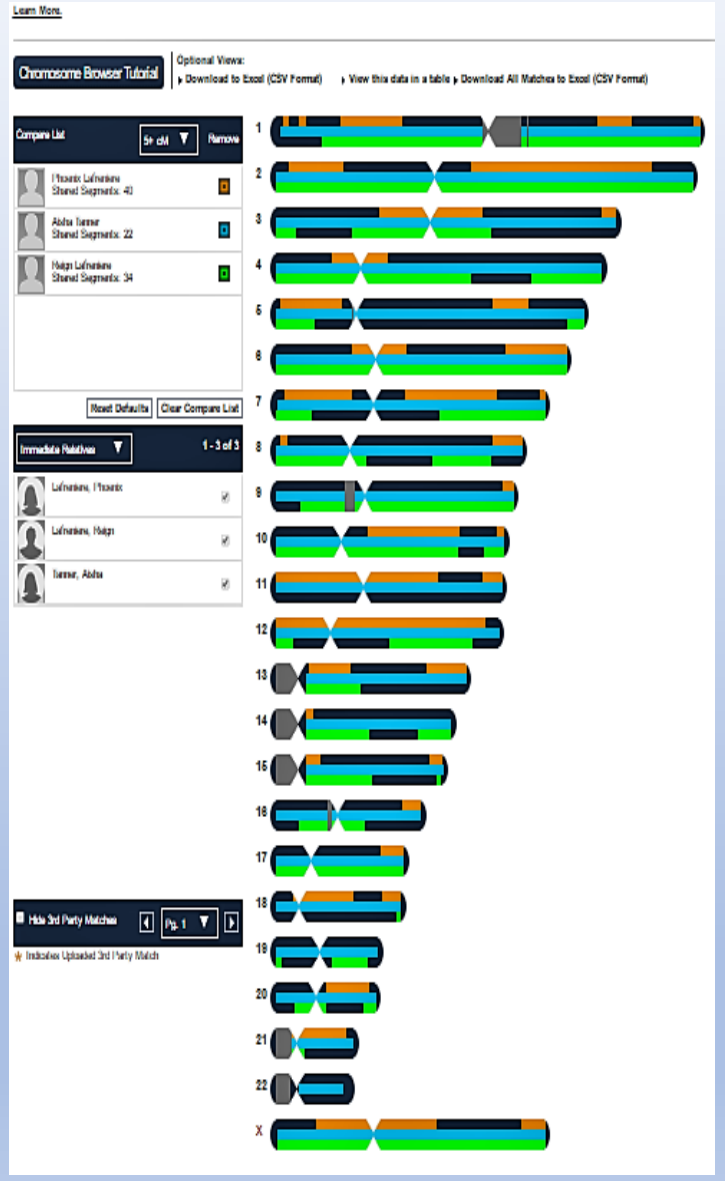
Minimum threshold size to be included in total = 700 SNPs
Mismatch-bunching Limit = 350 SNPs
Minimum segment cM to be included in total = 7.0 cM

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
X	2,710,157	154,551,755	196.0	16,538

Chr 23

Image size reduction: 1/17

Largest segment = 196.0 cM
Total of segments > 7 cM = 196.0 cM Actual.

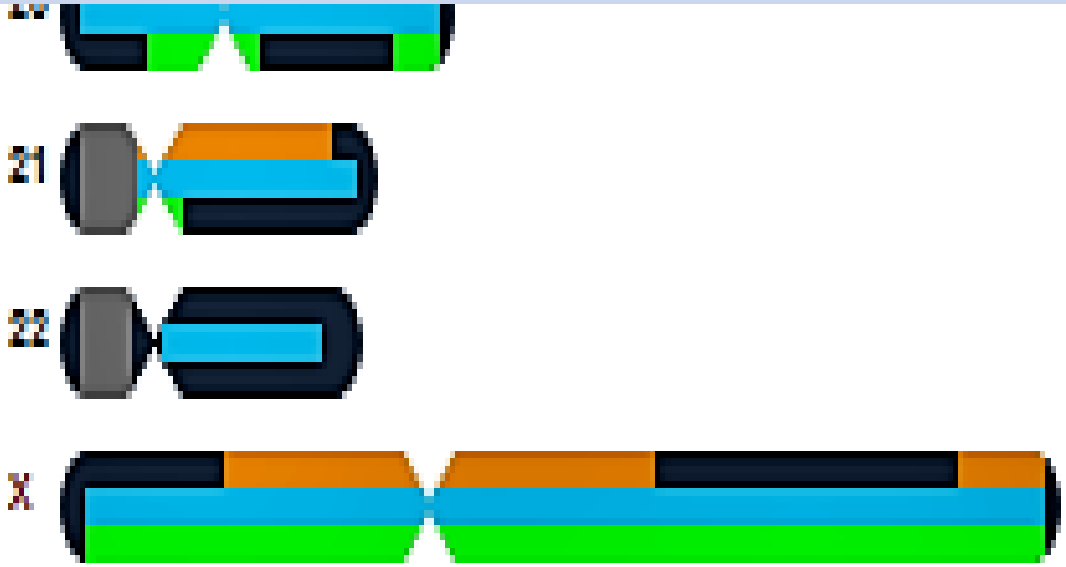


Chromosome Browser for adopted Grandson

Gold- Half Sister (total match = 24%)

Blue – Mother (total match = 100%)

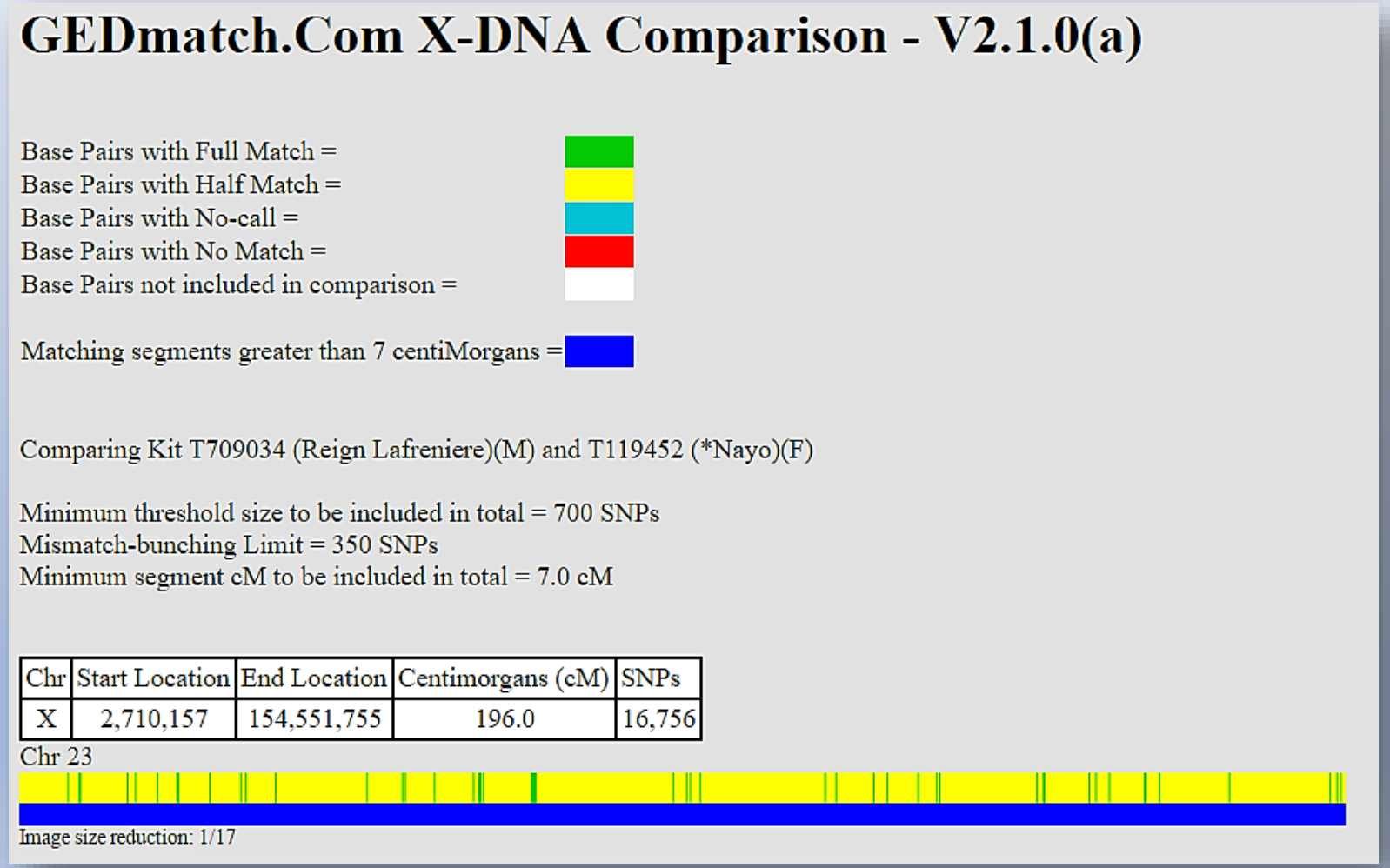
Green – Maternal Gradmother (tot. match = 32%)
(Note the Green Bar in the X Chromosome)



X Chromosome Inheritance

Mother to Son

As should be expected, there is a 100% match on the X chromosome from Mother to Son since he received the X only from her and a Y from dad. Note, however, that it is a half match (yellow) since he only received one of her two X chromosomes



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Matching segments greater than 7 centiMorgans =

Comparing Kit T709034 (Reign Lafreniere)(M) and T119452 (*Nayo)(F)

Minimum threshold size to be included in total = 700 SNPs
Mismatch-bunching Limit = 350 SNPs
Minimum segment cM to be included in total = 7.0 cM

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
X	2,710,157	154,551,755	196.0	16,756

Chr 23

Image size reduction: 1/17

X Chromosome Inheritance
Grandfather to Grandson

This “match” is much more unusual. As noted in Slide 7 , the X Chromosome tends to recombine from generation to generation. But not always. Note the graph is both all Blue, but also all Green. Which indicates a 100% Full match between the Grandfather and Grandson. That means the Grandson has the exact X DNA that his Great Grandmother was born with almost two centuries earlier.

GEDmatch.Com X-DNA Comparison - V2.1.0(a)

Base Pairs with Full Match =
Base Pairs with Half Match =
Base Pairs with No-call =
Base Pairs with No Match =
Base Pairs not included in comparison =

Matching segments greater than 7 centiMorgans =

Comparing Kit T278909 (Lionel Tanner)(M) and T709034 (Reign Lafreniere)(M)

Minimum threshold size to be included in total = 700 SNPs
Mismatch-bunching Limit = 350 SNPs
Minimum segment cM to be included in total = 7.0 cM

Chr	Start Location	End Location	Centimorgans (cM)	SNPs
X	2,710,157	154,551,755	196.0	16,694

Chr 23

Image size reduction: 1/17

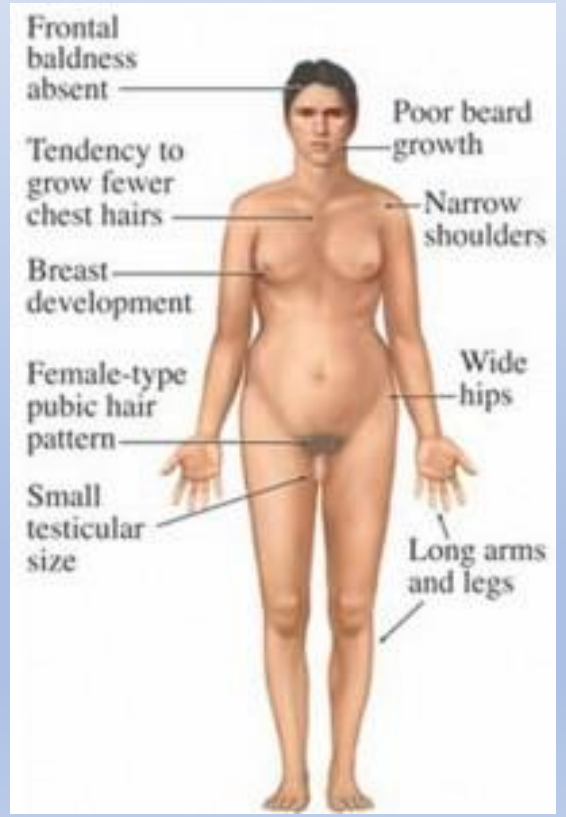
Largest segment = 196.0 cM
Total of segments > 7 cM = 196.0 cM Actual.

So what does all this mean? How is X DNA used in the body. As the presentation title suggests:

!*!*! It's complicated !*!*!

Related Diseases: 1 in every 650 births has an X Chromosome related disease

Klinefelter Syndrome – Male inherits both X's from mother, thus XXY instead of XY. Symptoms: lack of body hair, enlarged breasts, Narrow sloped shoulders.



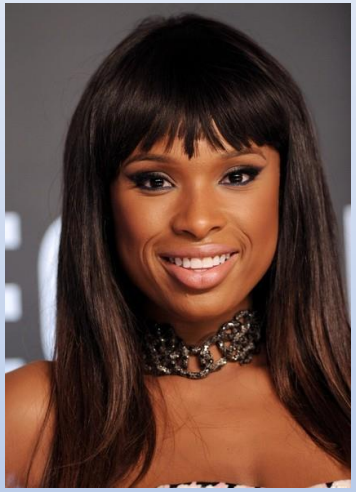
TARP Syndrome – Club foot, Heart Arterial Deformities

DENT Disease – Chronic Kidney Disorder

Microphthalmia – Small Eye Syndrome

Forest Whitaker

Jennifer Hudson

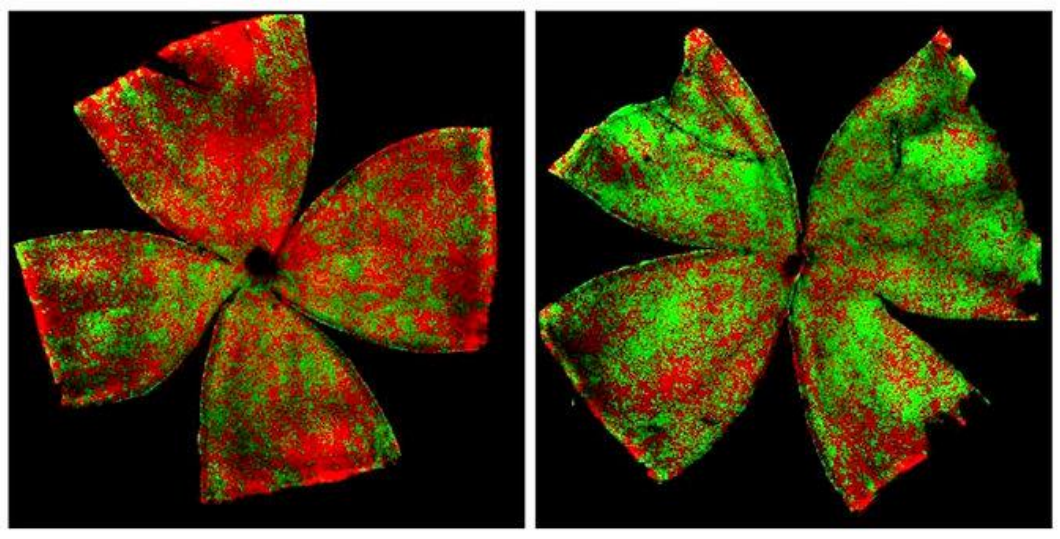


Penelope Cruz

So the X chromosome is not just related to whether a fertilized egg becomes Male (XY) or Female (XX). It has over 1,000 genes. Beside the various X related diseases listed above, and that is a small sample, an area that is getting much more attention is the "in- or de-activation" of one X chromosome in every new cell made from the fertilized egg. That seems to make sense. Why would you need two X chromosomes since the genes are the same in each X. You only need one, right? Not exactly, it's complicated.

Which one is In- De-activated – the male donated X or the female X? Early on geneticist thought it was random. But new studies show that is not always the case. Note the pictures below. Scientists developed a way to stain the X chromosome from a female mouse Green and the male X chromosome Red. These are images of a mouse's retinas. Why is the left retina primarily male X and the right mostly female X?

A calico cat (always female) is another example. The orange and black are the result of different X de-activation. White means no pigment



Cases have been found where there is a 95:5 ratio of forced male or female X chromosome deactivation. In those cases there is invariably a disease process included. Those disorders include:

1) **History of miscarriage,**

2) **Deficit of live male births, or**

3) **An otherwise unknown cause of mental retardation in a male family member.**

Non-deactivation is also present in cancer cells. Several forms of breast cancer have cells where both X Chromosomes are active.

Summary

XY – Male
XX – Female

An X chromosome may go several generations without recombining, especially in the father-to-daughter transfer. Estimates are that occurs in 3-4% of new babies, but that may be low.

X Chromosome contains over 1,000 genes affecting much more than sexual orientation. 1 in 650 births have some form of X related disease.

Very early in new cell development one of the two X chromsomes in a female cell is "in- or de-activated" supposedly random between male and female contributed X's BUT evidence is accumulating that is not always the case and the consequences may be good or bad.

!*!*! It's Complicated !*!*!

Dave Lewis – “Semi-Pro DNA Genealogist”

Stuff619@gmail.com (916) 626-2795

Available for Consultation and/or Tutoring