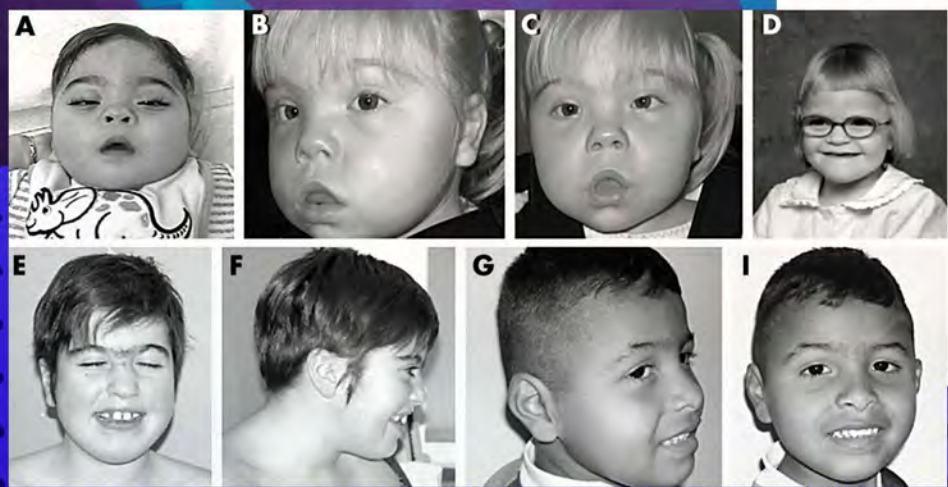
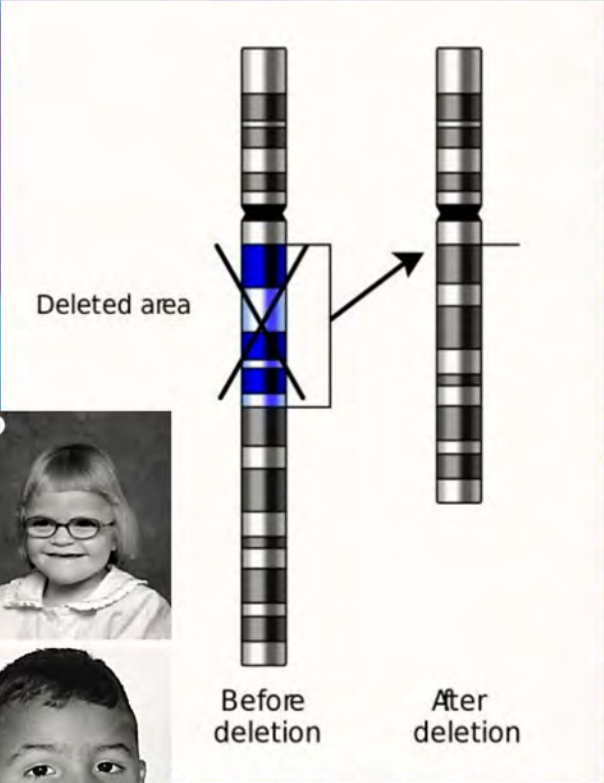




# KLEEFSTRA SYNDROME

A VISUAL ESSAY BY ABBEY WALTZ



YOURGENOME. (2017, JANUARY 6). CHROMOSOMES. [HTTPS://WWW.FLICKR.COM/PHOTOS/YOURGENOME/31768020130](https://www.flickr.com/photos/yourgenome/31768020130)  
MIRMILLON. (2013, SEPTEMBER 10). PELETION ON A CHROMOSOME [HTTPS://EN.WIKIPEDIA.ORG/WIKI/PELETION\\_\(GENETICS\)](https://en.wikipedia.org/wiki/Peletion_(genetics))  
YATSENKO, S. A. (N.P.). FACIAL PHOTOGRAPHS [HTTPS://IMG.BMJ.COM/CONTENT/42/4/328](https://img.bmj.com/content/42/4/328)



WHEN WE THINK OF DNA, WE USUALLY PICTURE A TWISTED LADDER STRUCTURE LIKE THIS...

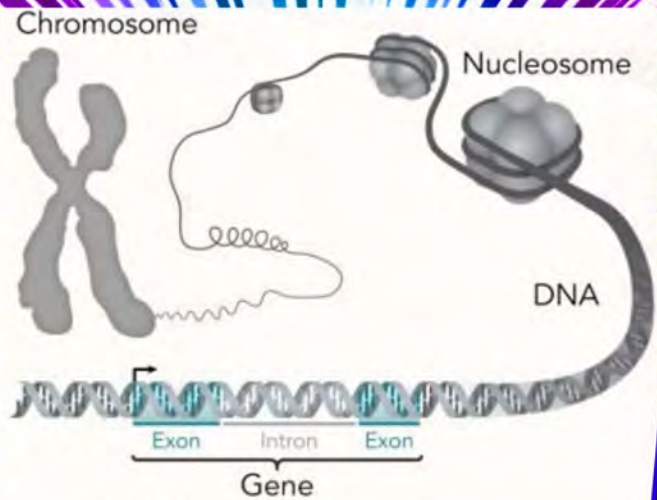


SIMPLE DNA STRAND [HTTPS://WWW.CLIPART.EMAIL/DOWNLOAD/905754.HTML](https://www.clipart.email/download/905754.html)

... BUT INSIDE THE BODY, DNA IS ACTUALLY FOUND IN TIGHTLY PACKED STRUCTURES CALLED:

# CHROMOSOMES

YOURGENOME. (2017, JANUARY 6). CHROMOSOMES. [HTTPS://WWW.FLICKR.COM/PHOTOS/YOURGENOME/31768020130](https://www.flickr.com/photos/yourgenome/31768020130)



THOMAS SPLETTSTOESSER [HTTPS://EN.WIKIPEPIA.ORG/WIKI/DNA\\_CONDENSATION](https://en.wikipedia.org/wiki/DNA_condensation)

THERES 23 PAIRS TOTAL!



JA, N. (N.P.). HUMAN CHROMOSOME. RETRIEVED FROM [HTTPS://EN.WIKIPEPIA.ORG/WIKI/KLINEFELTER\\_SYNDROME](https://en.wikipedia.org/wiki/Klinefelter_syndrome)

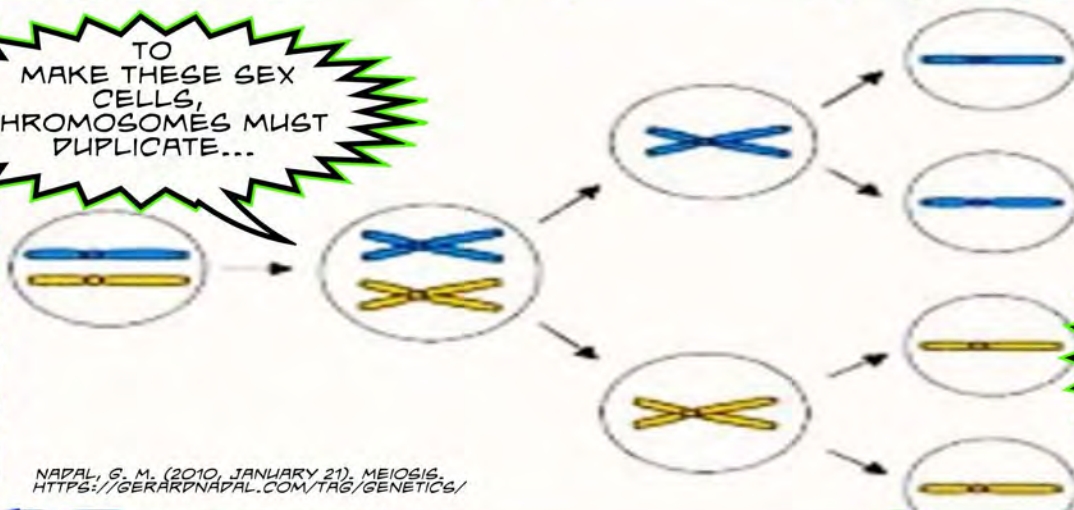


ONE OF THE MOST IMPORTANT FUNCTIONS OF CHROMOSOMES IS CARRYING GENETIC INFORMATION FROM PARENT TO OFFSPRING THROUGH:

# SEX CELLS

ISTOCKPHOTO. (2011, NOVEMBER 22). SPERM FERTILIZING HUMAN EGG. [HTTPS://MAPISON.COM/SPERM-FERTILIZING-HUMAN-EGG-ISTOCK-REMPERING-FILE-PHOTO](https://mapison.com/spERM-fERTILIZING-HUMAN-EGG-ISTOCK-REMPERING-FILE-PHOTO)

TO MAKE THESE SEX CELLS, CHROMOSOMES MUST DUPLICATE...

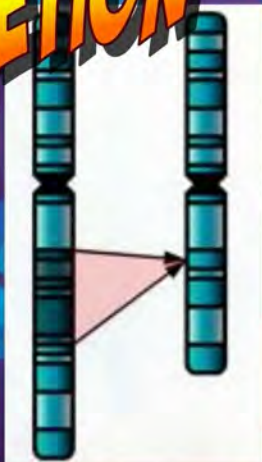


...THIS WAY, EACH SEX CELL CONTAINS ONE OF EACH CHROMOSOME

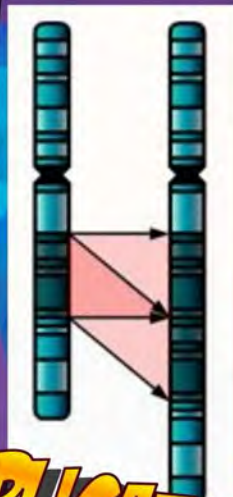
NAPAL, G. M. (2010, JANUARY 21). MEIOSIS. [HTTPS://GERARDNAPAL.COM/TAG/GENETICS/](https://gerardnapal.com/tag/genetics/)

BUT THINGS DON'T ALWAYS GO AS PLANNED DURING REPLICATION...

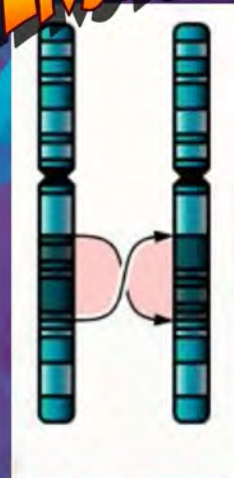
## DELETION



## DUPLICATION



## INVERSION



WHEELER, R. (2014, MAY 24). SINGLE CHROMOSOME MUTATIONS. [HTTPS://EN.WIKIPEDIA.ORG/WIKI/CHROMOSOME\\_ABNORMALITY](https://en.wikipedia.org/wiki/Chromosome_abnormality)



THESE MISTAKES ARE KNOWN AS:

# MUTATIONS

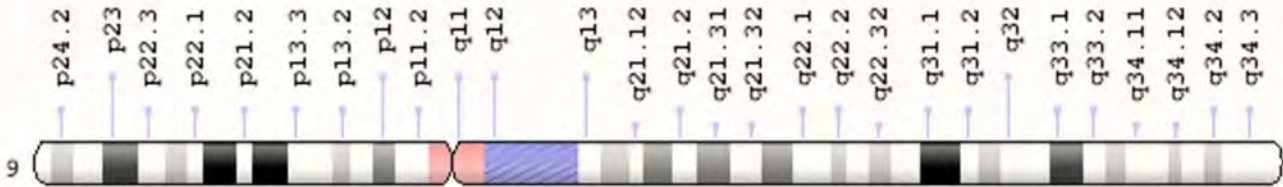
PASCHER, C. (N.P.). CHROMOSOME MUTATIONS  
[HTTPS://WWW.THUGHTCO.COM/CHROMOSOME-MUTATION-373448](https://www.thoughtco.com/chromosome-mutation-373448)



WHICH MAY BE HARMFUL, HELPFUL, OR NEUTRAL

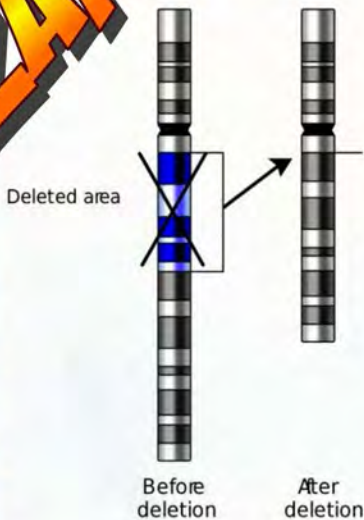
CRIMSON HEXAGON. (N.P.). EMOTION AND SENTIMENT ANALYSIS  
[HTTPS://WWW.CRIMSONHEXAGON.COM/SENTIMENT-ANALYSIS/](https://www.crimsonhexagon.com/sentiment-analysis/)

IF DELETION HAPPENS TO OCCUR AT THE 9P34.3 GENE ON CHROMOSOME 9... [1]



NCIB. (N.P.). CHROMOSOME 9 [HTTPS://GHR.NLM.NIH.GOV/CHROMOSOME/9#IDIOPGRAM](https://ghr.nlm.nih.gov/chromosome/9#idiogram)

# ZZZAP!



...THE OFFSPRING WILL LIKELY BE BORN WITH:

# KLEEFSTRA SYNDROME

MIRMILLON. (2013, SEPTEMBER 10). PELETION ON A CHROMOSOME  
[HTTPS://EN.WIKIPEPIA.ORG/WIKI/PELETION\\_\(GENETICS\)](https://en.wikipedia.org/wiki/Peletion_(genetics))

YOURGENOME. (2017, JANUARY 6). CHROMOSOMES.  
[HTTPS://WWW.FLICKR.COM/PHOTOS/YOURGENOME/31768020130](https://www.flickr.com/photos/yourgenome/31768020130)



CHILDREN BORN WITH KLEEFSTRA SYNDROME MAY HAVE DISTINCT FACIAL FEATURES... [2]



YATSENKO, S. A. (N.P.). FACIAL PHOTOGRAPHS [HTTPS://JMG.BMJ.COM/CONTENT/42/4/328](https://jmg.bmj.com/content/42/4/328)

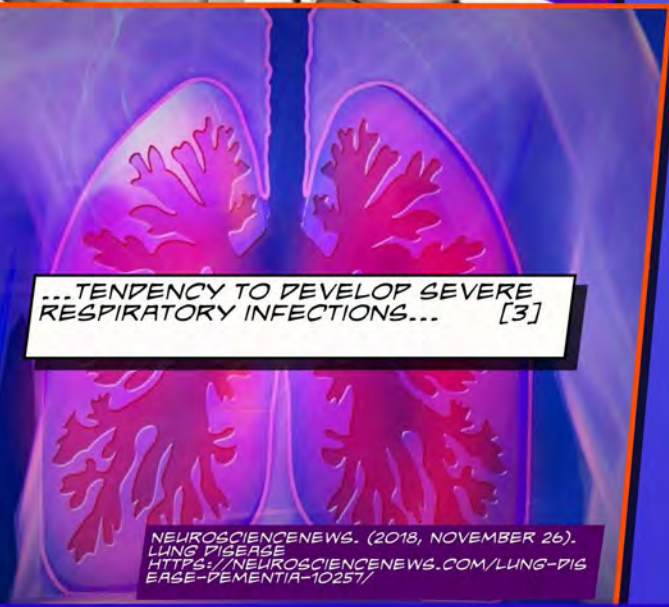


... CONGENITAL HEART MALFORMATIONS... [3]



DIAGRAM OF THE HUMAN HEART (N.P.). [HTTPS://COMMONS.WIKIMEDIA.ORG/WIKI/FILE:DIAGRAM\\_OF\\_THE\\_HUMAN\\_HEART\\_\(CROPPED\).SVG](https://commons.wikimedia.org/wiki/File:Diagram_of_the_human_heart_(cropped).svg)

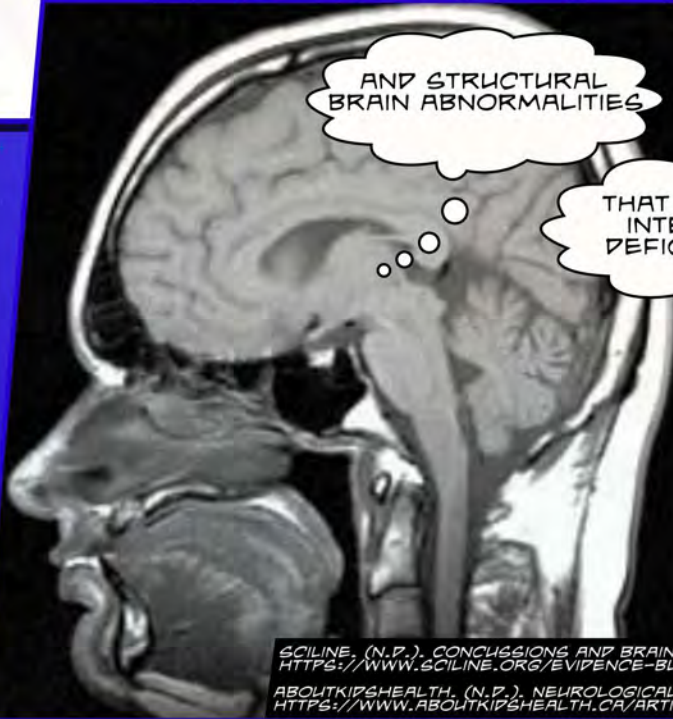
... TENDENCY TO DEVELOP SEVERE RESPIRATORY INFECTIONS... [3]



NEUROSCIENCE NEWS. (2018, NOVEMBER 26). LUNG DISEASE [HTTPS://NEUROSCIENCE NEWS.COM/LUNG-DISEASE-10257/](https://neurosciencenews.com/lung-disease-10257/)

AND STRUCTURAL BRAIN ABNORMALITIES

THAT MAY LEAD TO INTELLECTUAL DEFICIENCIES [3]



SCILINE. (N.P.). CONCUSSIONS AND BRAIN HEALTH [HTTPS://WWW.SCILINE.ORG/EVIDENCE-BLOG/CONCUSSIONS](https://www.sciline.org/evidence-blog/concussions)

ABOUTKID'SHEALTH. (N.P.). NEUROLOGICAL MRI: SAGITTAL VIEW [HTTPS://WWW.ABOUTKID'SHEALTH.CA/ARTICLE?CONTENTID=1334&LANGUAGE=ENGLISH](https://www.aboutkidshealth.ca/article?contentid=1334&language=english)



ALTHOUGH THERE IS NO CURE FOR KLEEFSTRA SYNDROME, THERE ARE SOME EFFECTIVE:

# TREATMENT METHODS

LIKE PHYSICAL THERAPY! [4]



THINKSTOCK. (2017, DECEMBER 1)  
[HTTP://WWW.CALHEALTHREPORT.ORG/2017/12/01/CHILD-PREN-AUTISM-ZIP-COPE-MATTERS](http://www.calhealthreport.org/2017/12/01/child-pren-autism-zip-cope-matters)

SPEECH THERAPY! [4]



PIXABAY. (N.P.). TALK COMMUNICATION TALKING  
[HTTPS://PIXABAY.COM/ILLUSTRATIONS/TALK-COMMUNICATION-TALKING-HAPPY-2782815/](https://pixabay.com/illustrations/talk-communication-talking-happy-2782815/)

AND REGULAR CARDIAC, RENAL, INTESTINAL, AND PULMONARY MONITORING! [4]



VIEWSTOCK. (N.P.). DOCTOR AND PATIENT  
[HTTPS://WWW.123RF.COM/PHOTO\\_30132392\\_DOCTOR-AND-PATIENT.HTM](https://www.123RF.com/photo_30132392_doctor-and-patient.htm)

GENETIC TESTING IS ALSO RECOMMENDED, AS IT CAN BE USED TO DETECT KLEEFSTRA SYNDROME AS EARLY AS POSSIBLE. [5]



NIH. (2018, APRIL 17). NONINVASIVE PRENATAL GENETIC TESTING  
[HTTPS://WWW.GENOME.GOV/27570937/APRIL-17-NONINVASIVE-PRENATAL-GENETIC-TESTING/](https://www.genome.gov/27570937/april-17-noninvasive-prenatal-genetic-testing/)



# REFERENCES

- [1] YAMADA. (N.D.). KLEEFSTRA SYNDROME. RETRIEVED FROM [HTTPS://WWW.MALACARDS.ORG/CARD/KLEEFSTRA\\_SYNDROME](https://www.malacards.org/card/kleefstra_syndrome)
- [2] WHAT IS KLEEFSTRA SYNDROME? (N.D.). RETRIEVED FROM [HTTPS://WWW.KLEEFSTRASYNDROME.ORG/WHAT-IS-KLEEFSTRA-SYNDROME/](https://www.kleefstrasynndrome.org/what-is-kleefstra-syndrome/)
- [3] [HTTPS://GHR.NLM.NIH.GOV/CONDITION/KLEEFSTRA-SYNDROME](https://ghr.nlm.nih.gov/condition/kleefstra-syndrome)
- [4] INTELLECTUAL DISABILITY. (N.D.). RETRIEVED FROM [HTTPS://WWW.ASHA.ORG/PRPSPECIFICTOPIC.ASPX?FOLDERID=858994254ION=TREATMENT](https://www.asha.org/prpspecifictopic.aspx?folderid=858994254ion=treatment)
- [5] [HTTP://WWW.FINDRESOURCES.CO.UK/THE-SYNDROMES/KLEEFSTRA/DIAGNOSIS](http://www.findresources.co.uk/the-syndromes/kleefstra/diagnosis)