DR. GREGOR JOHANN MENDEL
(1822-1884)

See: http://www.blc.arizona.edu/courses/181gh/rick/genetics1/graphics/pea_traits.gif
Pea figures from webpage of Richard Halick, U. Arizona

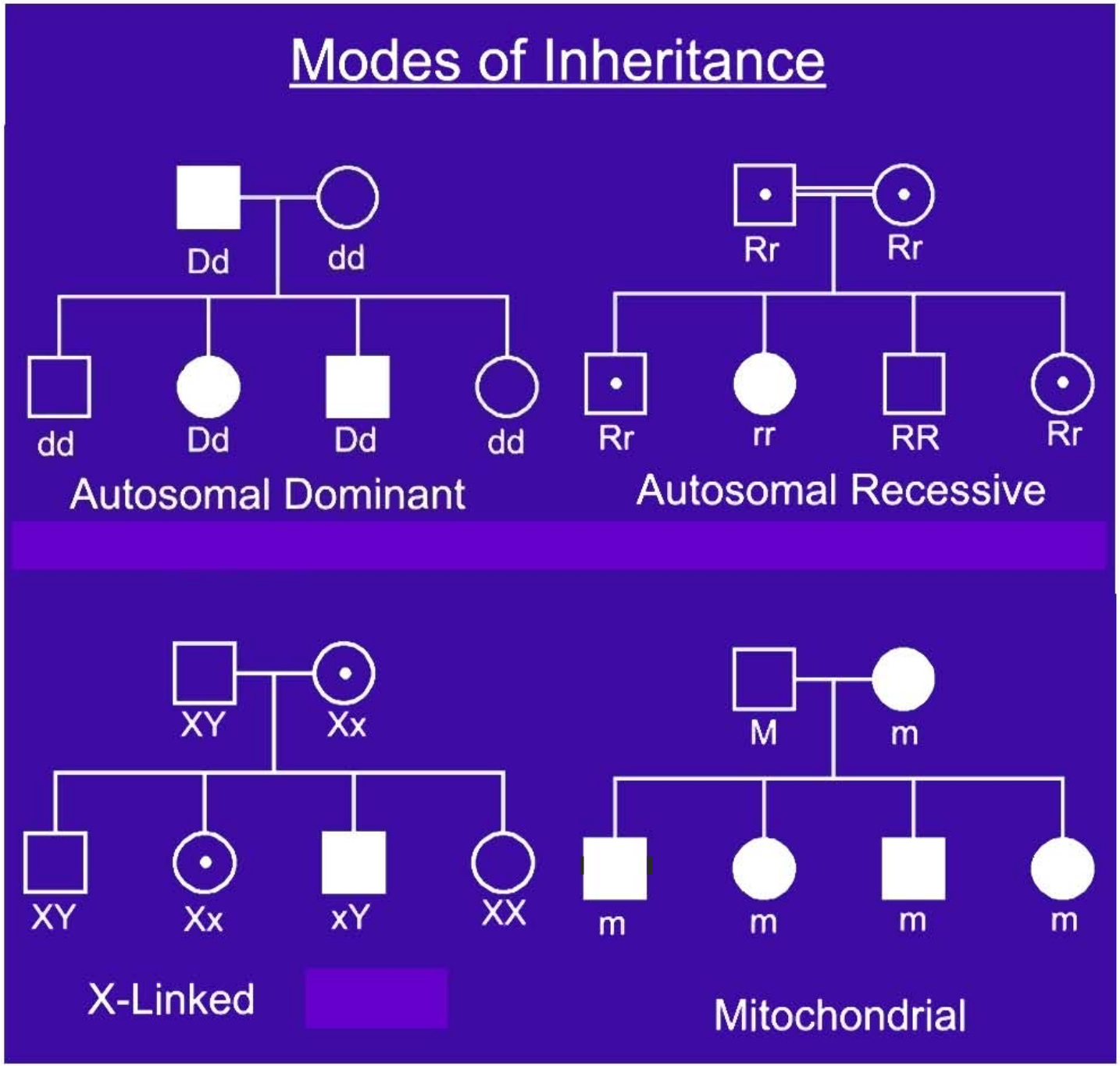
Smooth or dented seeds

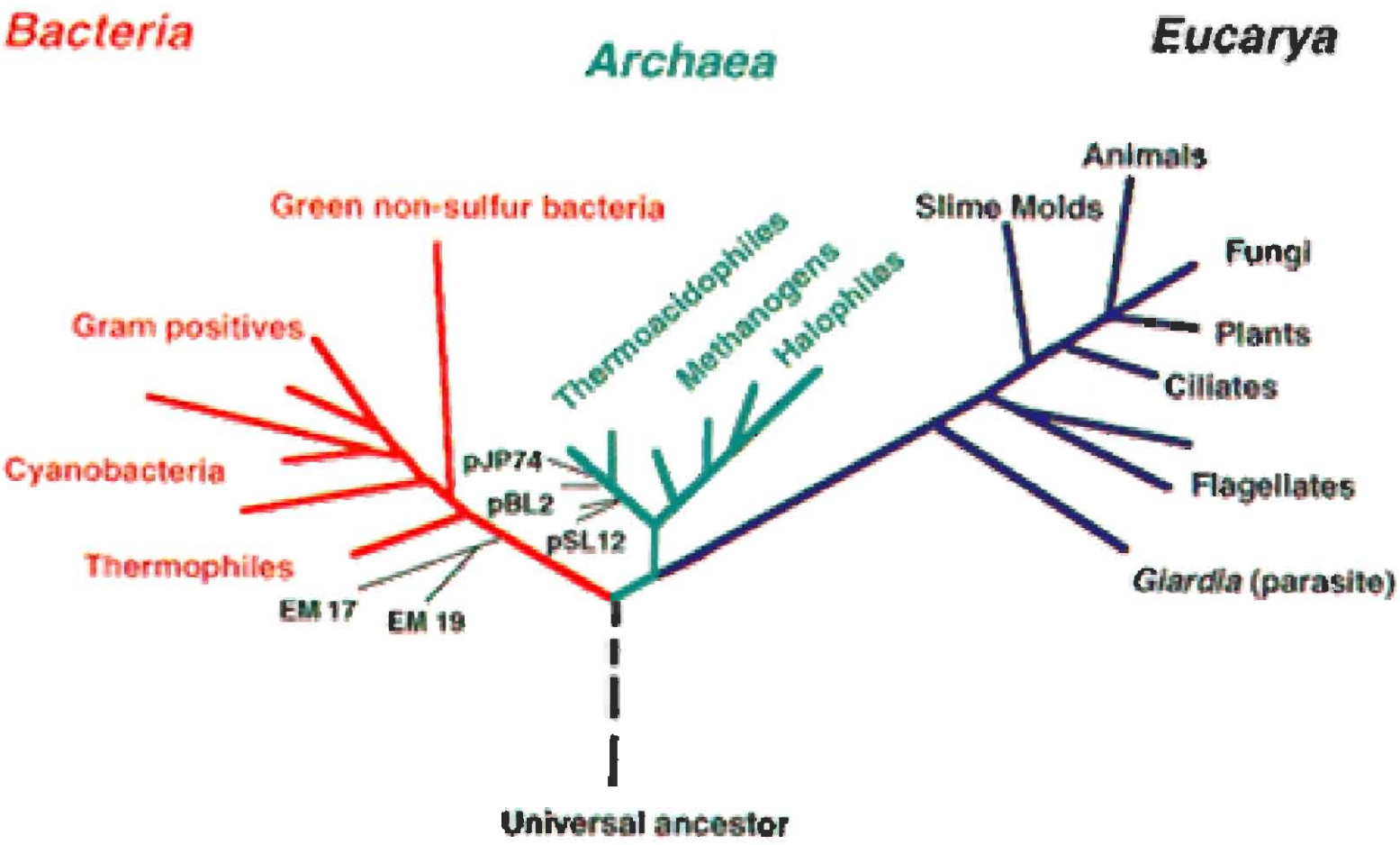
Yellow or green seeds

Axial or terminal flowers

Green or yellow pods

Purple or white flowers


Modes of Inheritance
Dd
dd
dd
Dd
Dd
dd
Autosomal Dominant
Rr
Rr
Rr
rr
RR
Rr
Autosomal Recessive
XY
Xx
XY
Xx
xY
XX
X-Linked
M
m
m
m
m
m
Mitochondrial



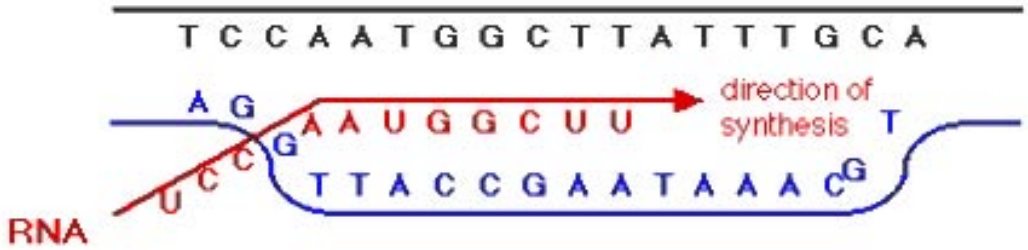
THE UNIVERSAL TREE OF LIFE

The Genetic Code

See Purves et al., Life: The Science of Biology, 4th Edition, by Sinauer Associates (www.sinauer.com) and WH Freeman (www.whfreeman.com).

Transcription of RNA from DNA

- The bottom strand of the DNA molecule above is the template for RNA synthesis.
- RNA polymerase makes a copy of the DNA sequence but substitutes uridine (U) in place of thymine (T)



- The botttom strand of the DNA duplex is used as the template to synthesize RNA. However, the sequence of bases in the RNA is the same as in the top strand of the DNA, with U in place of T

Point mutations

Wild type allele:

M D D Q S R M L Q T L A G V N L

atggacgatcaatccaggatgctgcagactctggccggggtgaacctg

silent (third base pair) mutation:

M D D Q S R M L Q T L A G V N L

atggacgatcaatccaggatgctgcaaactctggccggggtgaacctg

point mutation (missense):

M D D Q S R M L K T L A G V N L

atggacgatcaatccaggatgctgaagactctggccggggtgaacctg

point mutation (nonsense):

M D D Q S R M L stop

atggacgatcaatccaggatgctgtagactctggccggggtgaacctg

frameshift leading to premature termination:

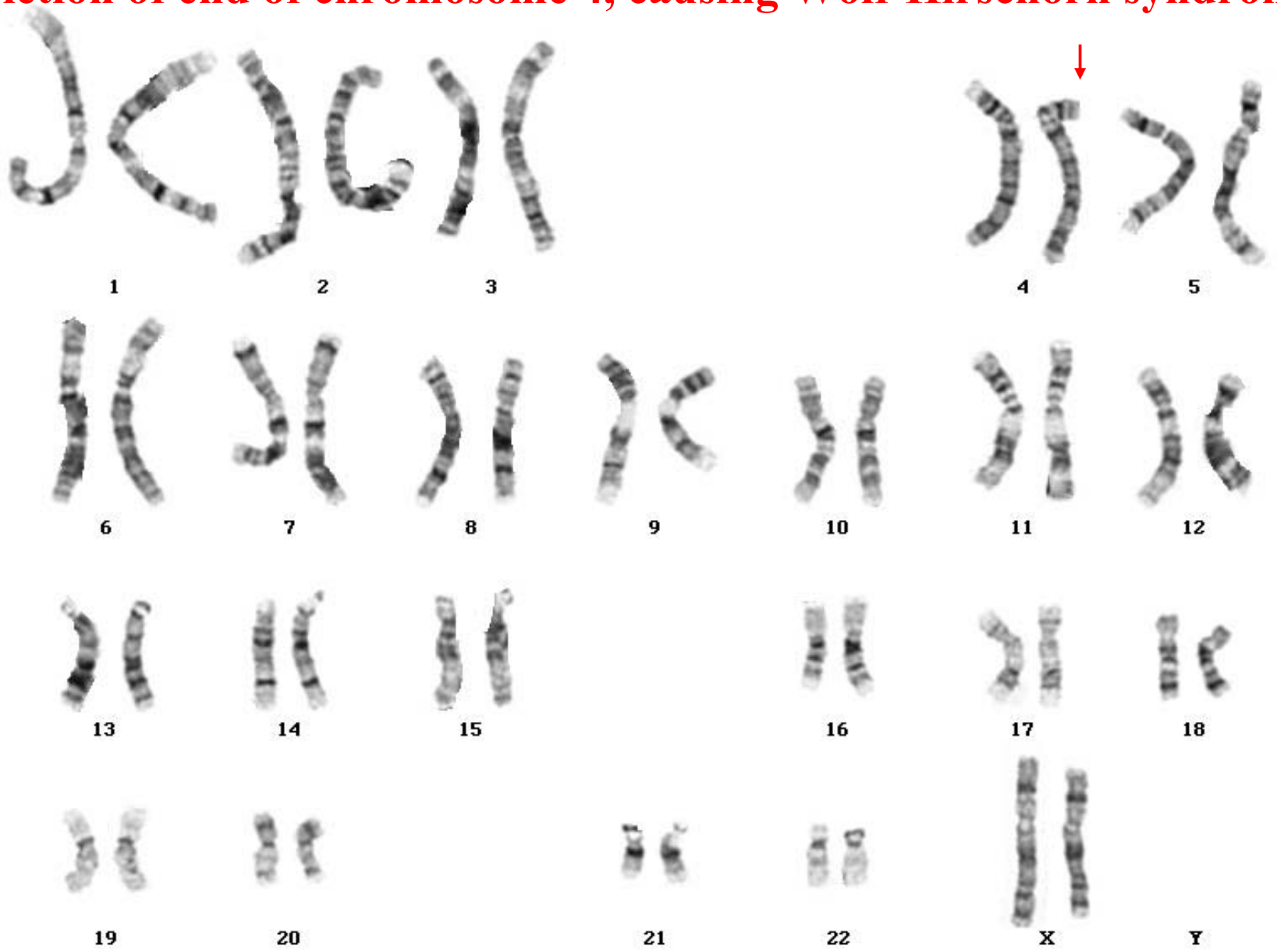
M D D Q S R M L R L W P G stop

atggacgatcaatccaggatgctgagactctggccggggtgaacctg

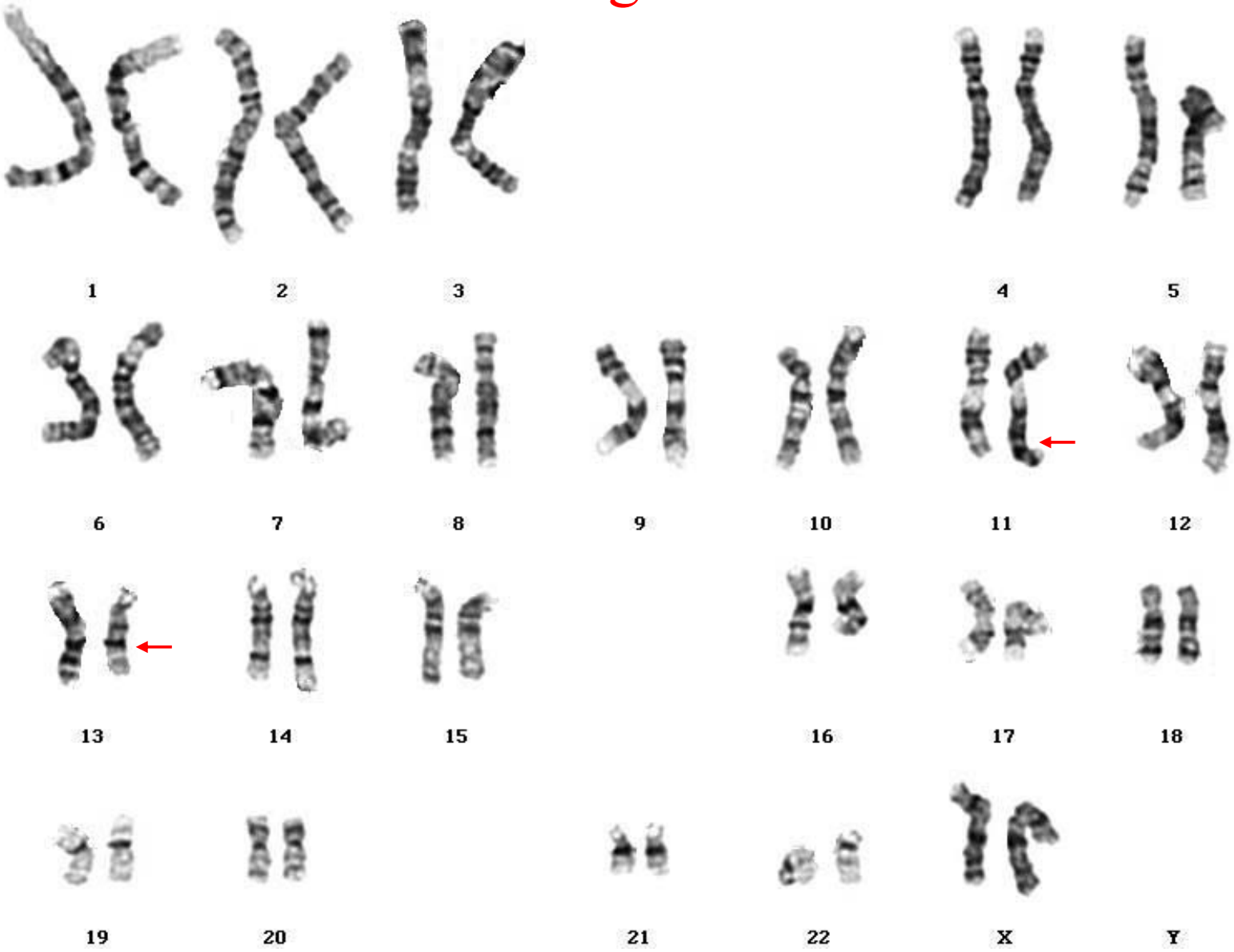
Larger Mutations

Deletions	Can involve deletion of one to many genes. Large deletions may be visible cytogenetically. Phenotype can vary from mild to extreme. Many recognized “syndromes” are caused by chromosome deletions.
Translocations	Interchange of two chromosome pieces. Can be benign if no genes are interrupted. Can lead to gene discovery.
Inversion	Inversion of chromosome section. Can be benign if no genes are interrupted. Can lead to gene discovery.

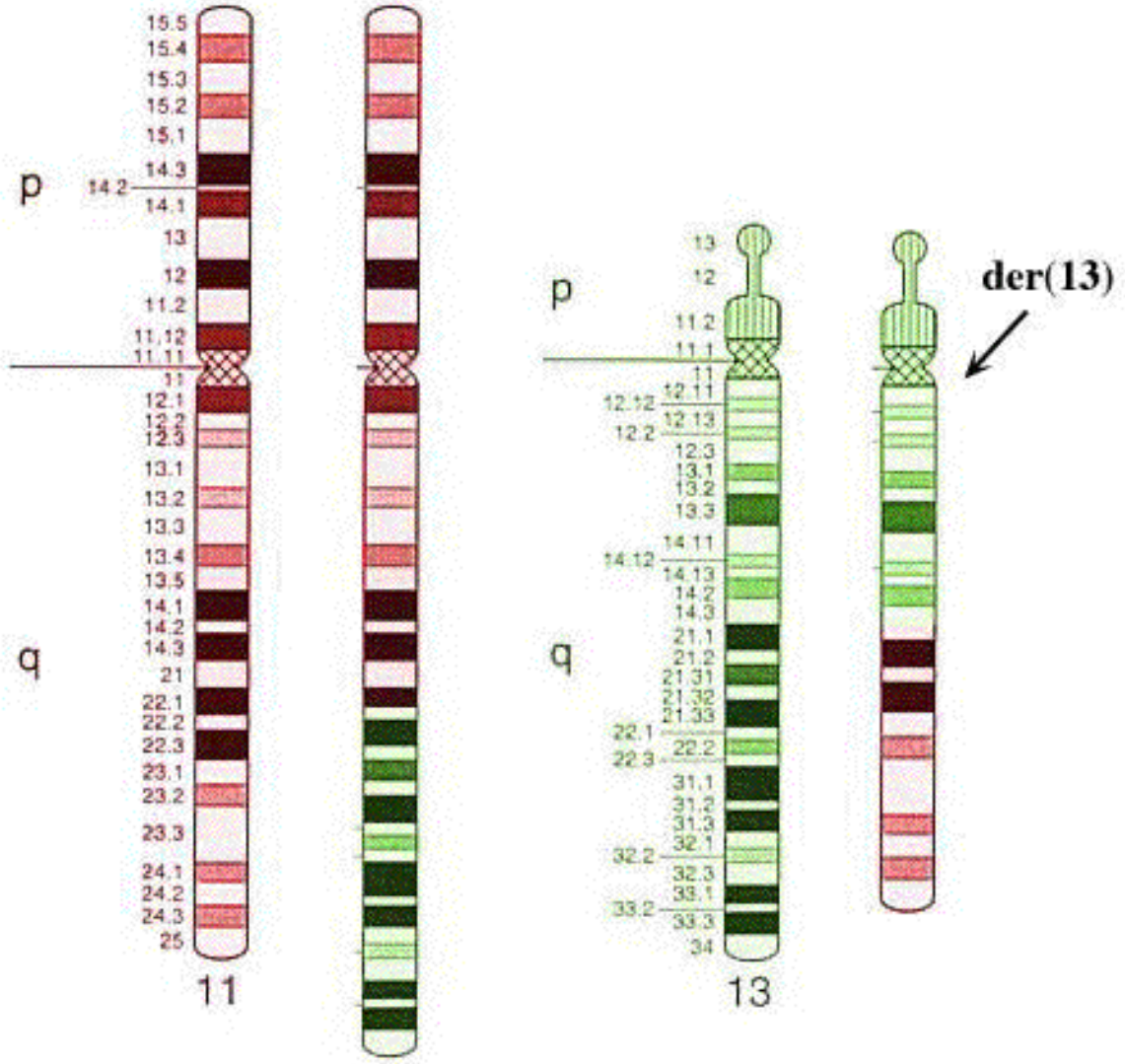
Deletion of end of chromosome 4, causing Wolf-Hirschorn syndrome



Translocation involving chromosomes 11 and 13



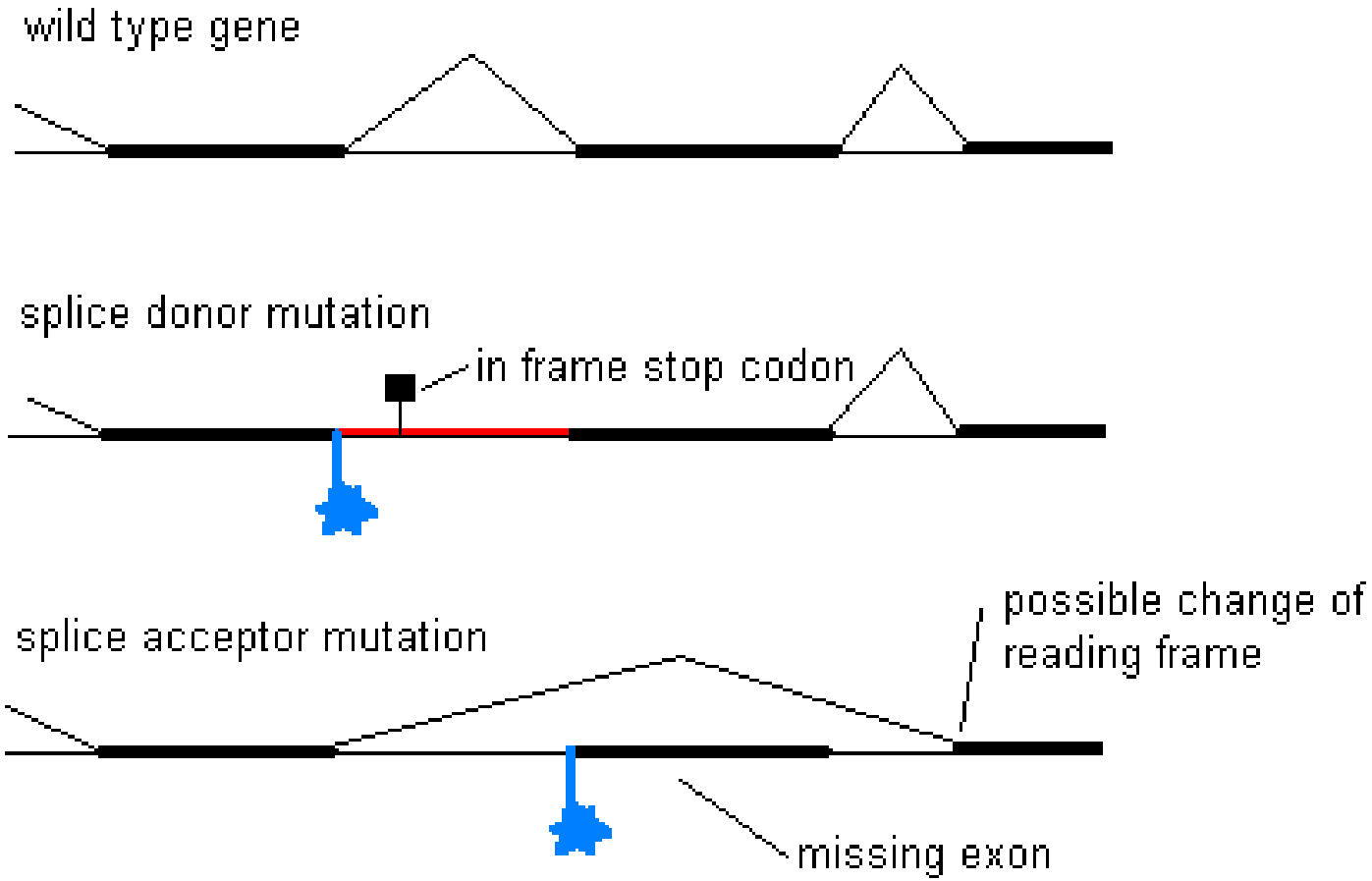
Translocation involving chromosomes 11 and 13



t(11;13)(q21;q14.3)

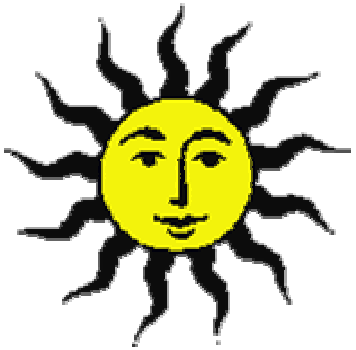
PAX3 at 2q35

See Ishikiriyama *et al*., 1989



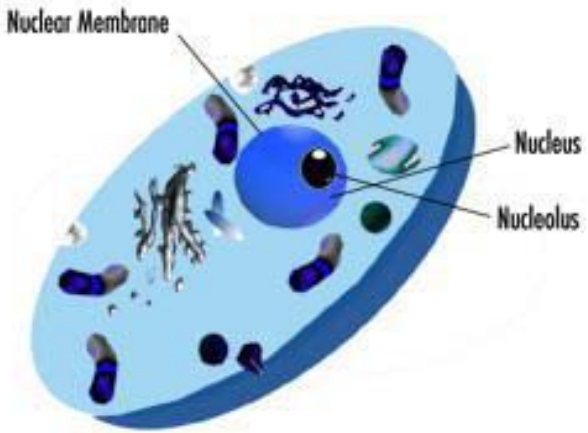
www.ucl.ac.uk/~ucbhjow/ bmsi/bmsi_6.html

How do heritable DNA mutations occur??



UV exposure

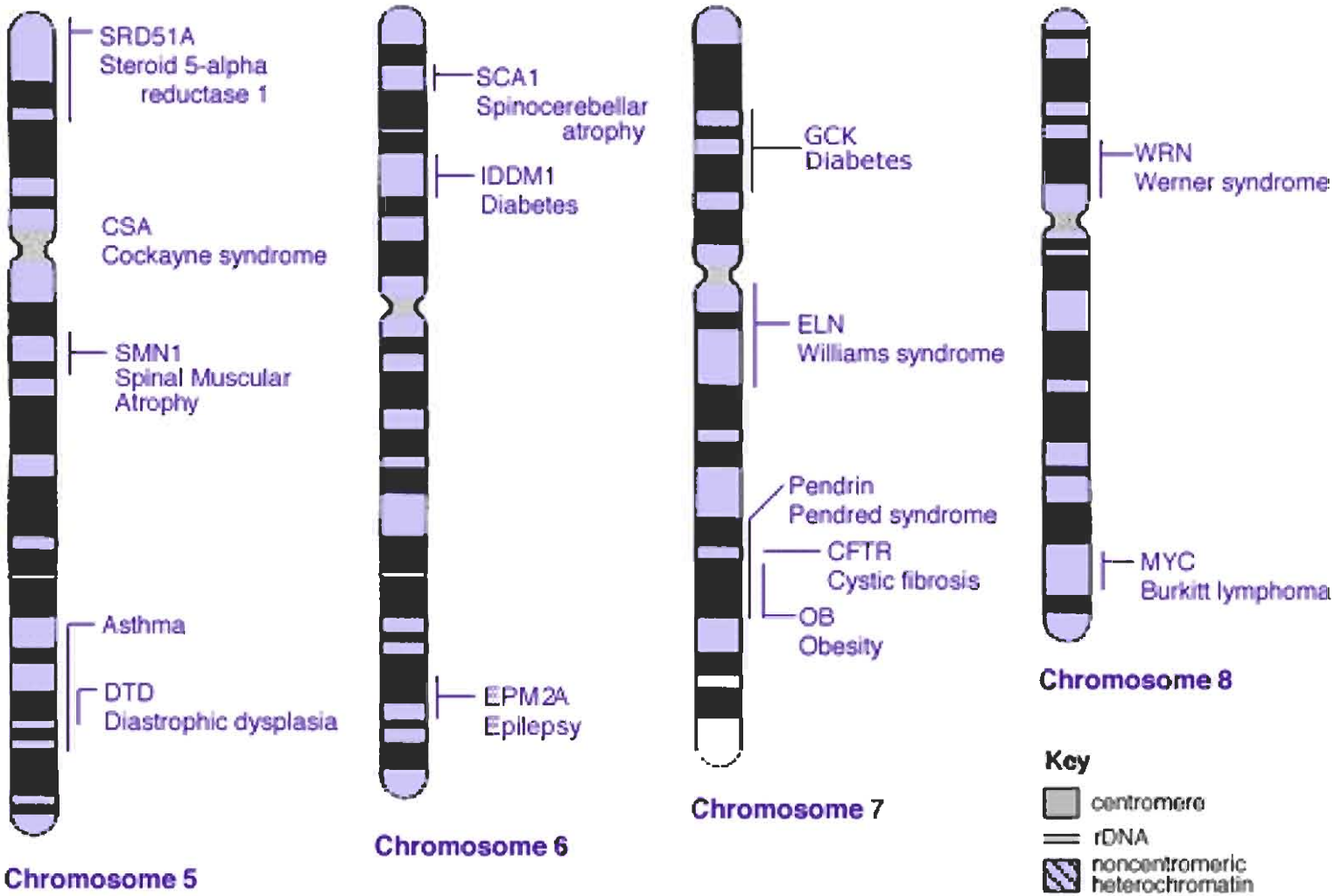
Chemical exposure

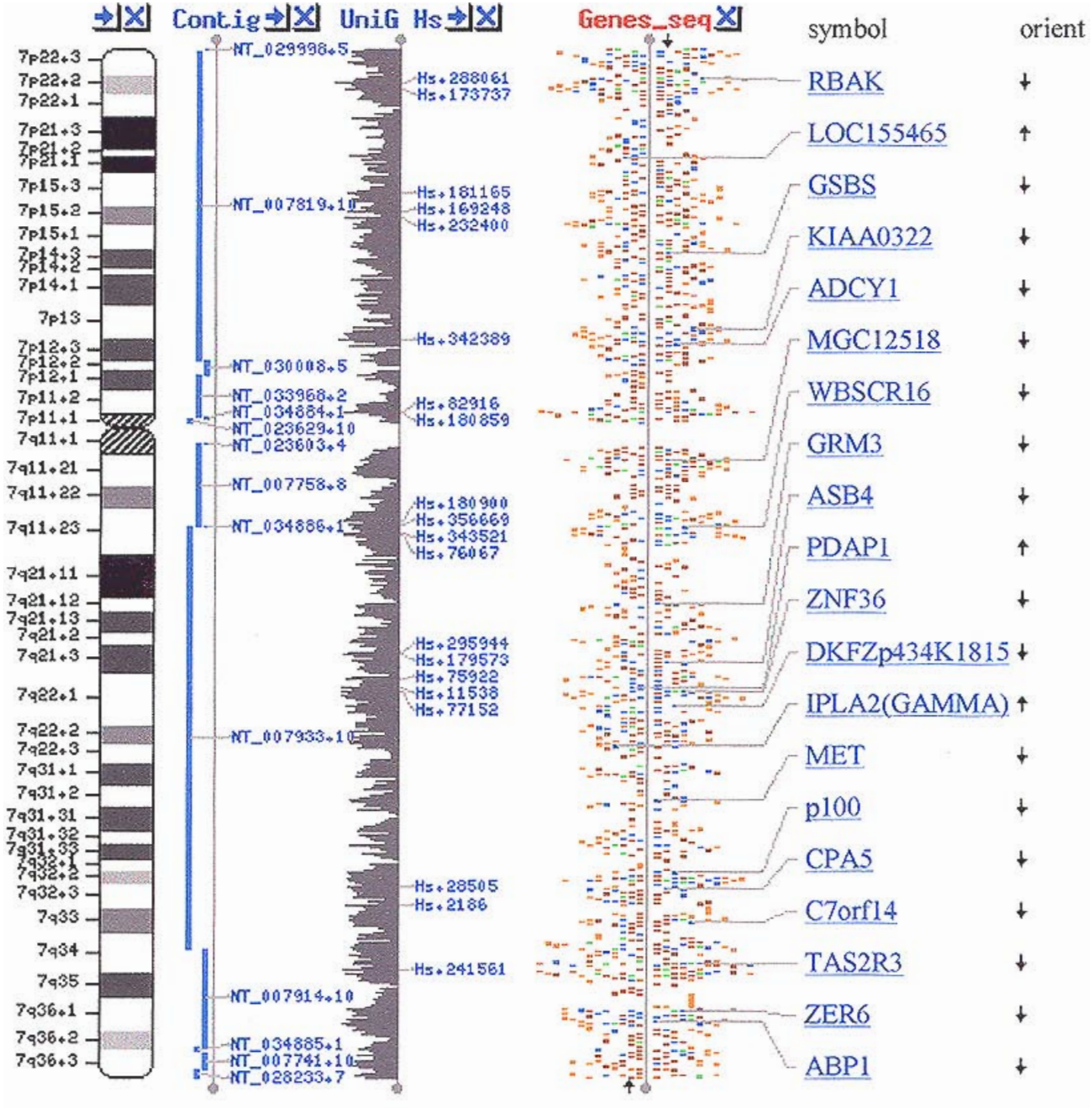


Faulty cellular mechanisms

NCBI Genes and disease Map

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y




Contig
UniG Hs
Genes_seq
symbol
orient
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7p22.2
7p22.1
7p21.3
7p21.2
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7p15.3
7p15.2
7p15.1
7p14.3
7p14.2
7p14.1
7p13
7p12.3
7p12.2
7p12.1
7p11.2
7p11.1
7q11.1
7q11.21
7q11.22
7q11.23
7q21.11
7q21.12
7q21.13
7q21.2
7q21.3
7q22.1
7q22.2
7q22.3
7q31.1
7q31.2
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7q31.32
7q31.33
7q32.1
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7q33
7q34
7q35
7q36.1
7q36.2
7q36.3
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PDAP1
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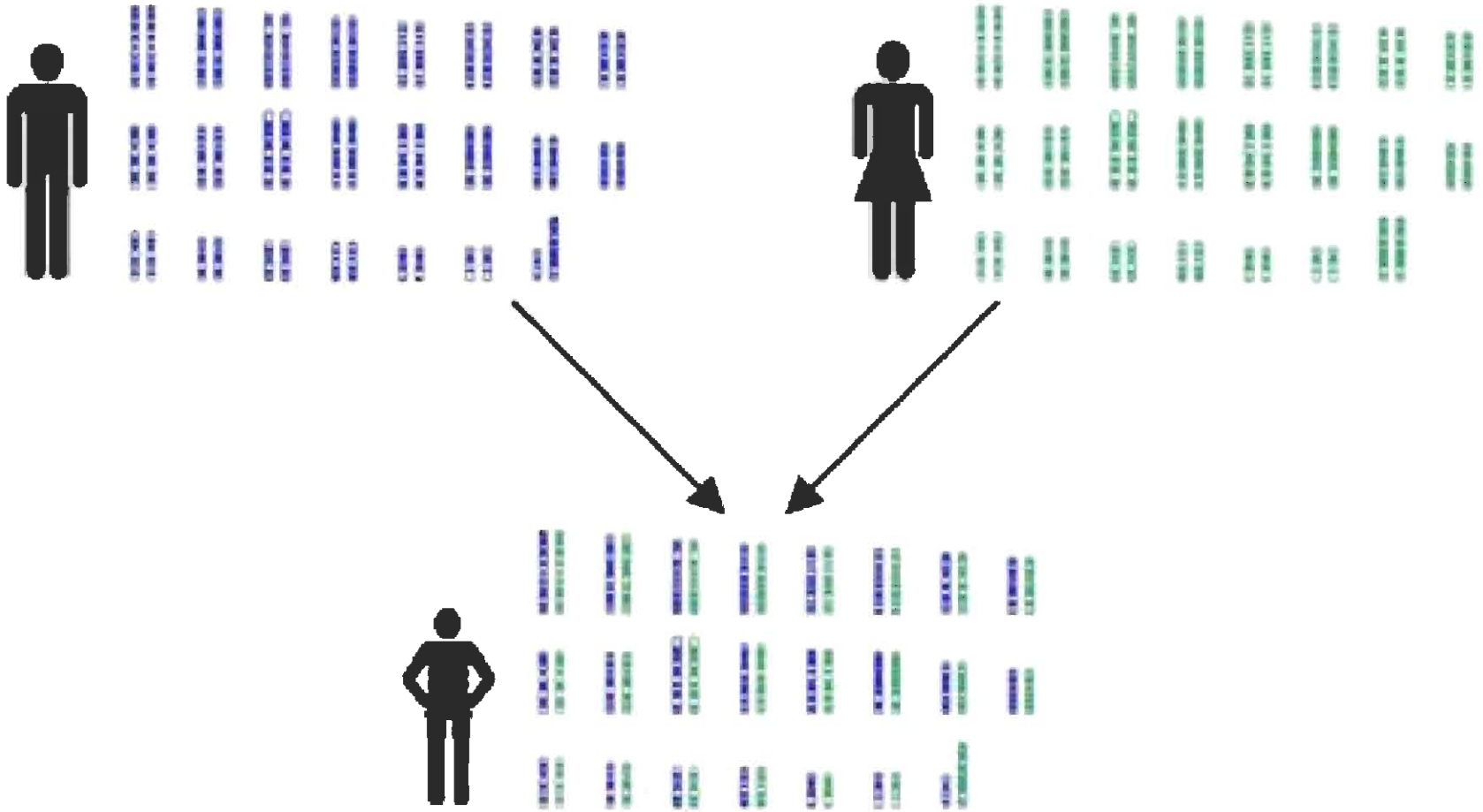
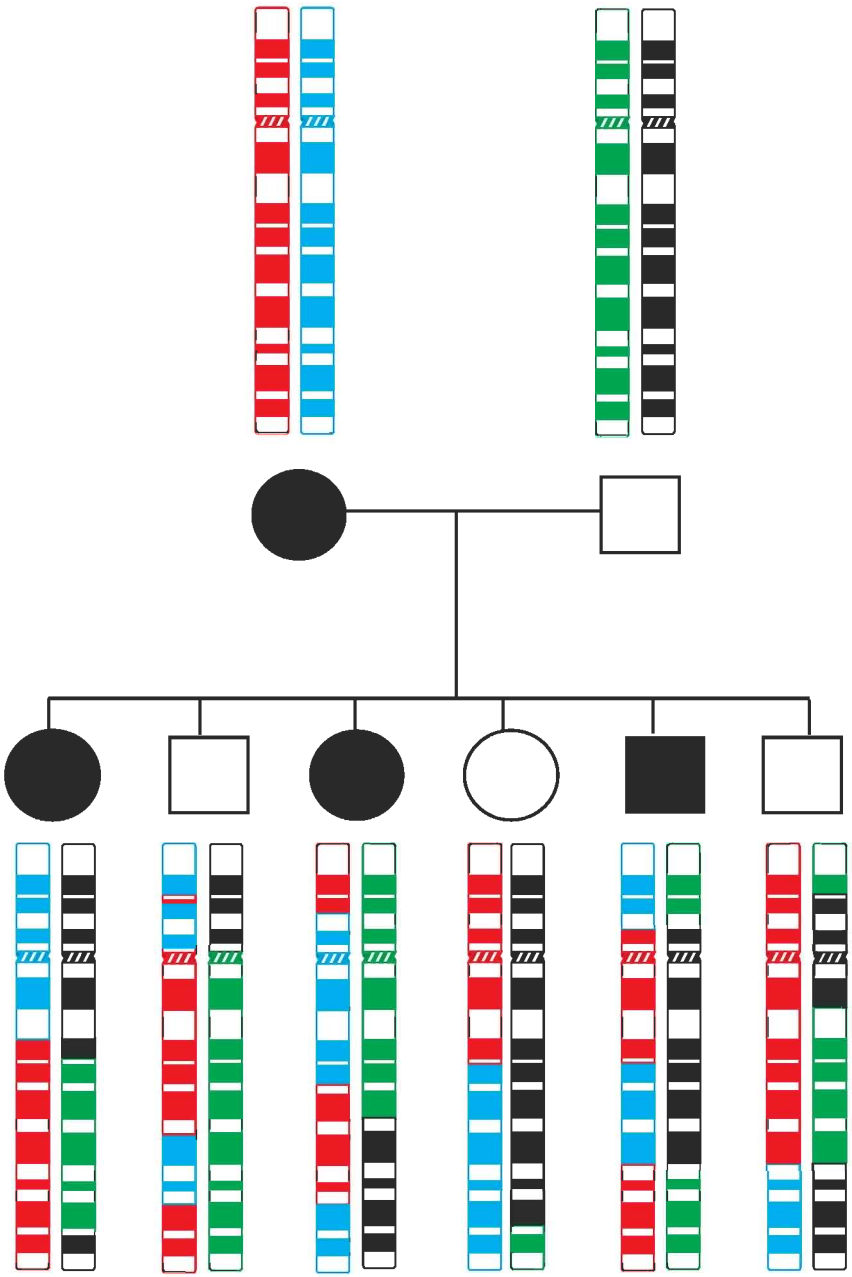
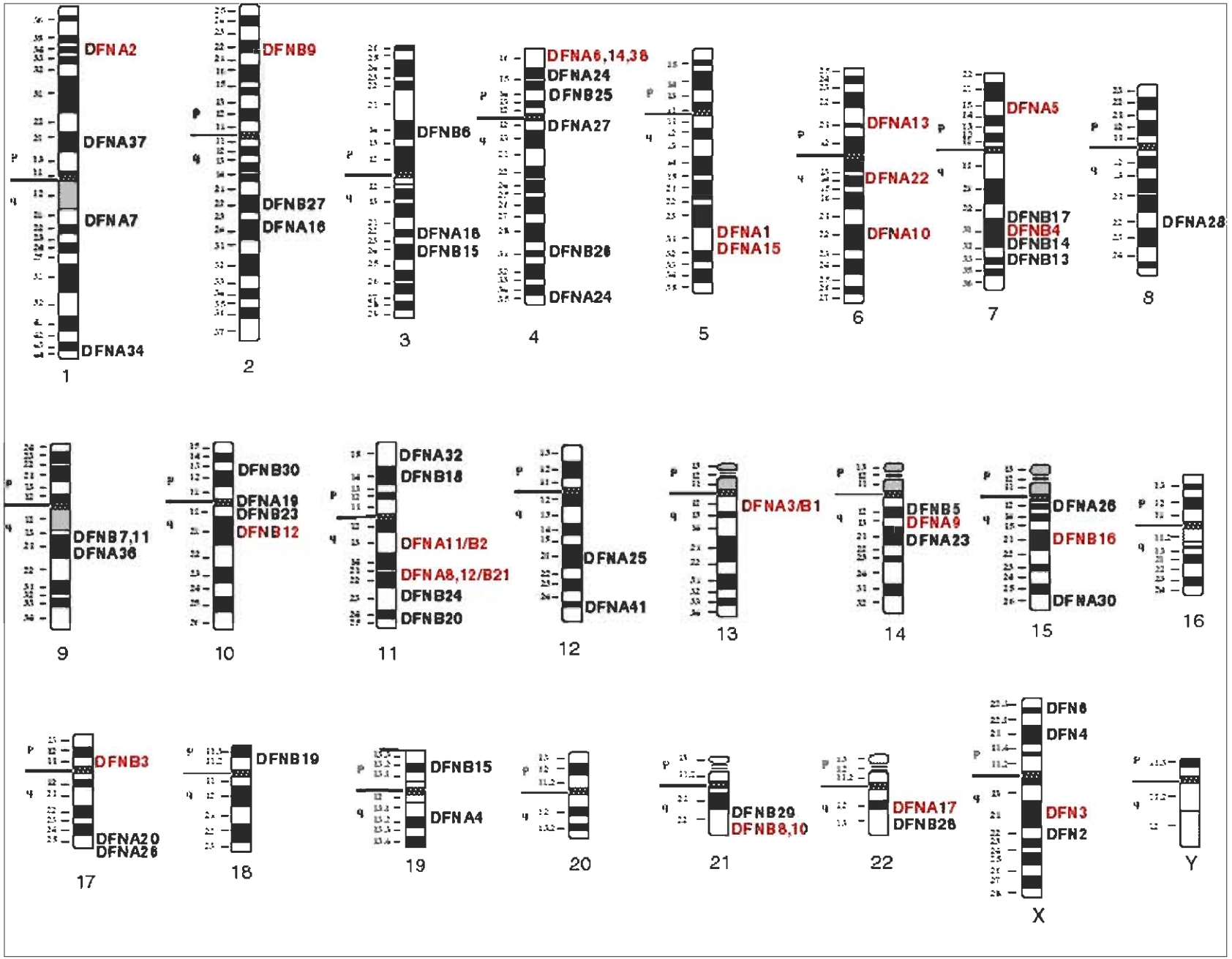
C7orf14
TAS2R3
ZER6
ABP1



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DFNA26
17
DFNB19
18
DFNB15
DFNA4
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20
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21
DFNA17
DFNB28
22
DFN6
DFN4
DFN3
DFN2
X
Y

VNTRs for a genetic locus

See: Thompson & Thompson, Genetics in Medicine, 2001

Microsattelite markers in human DNA

See: Thompson & Thompson, Genetics in Medicine, 2001

DNA fingerprints of MZ vs DZ twins using multiple VNTRs located throughout the genome

See: Thompson & Thompson, Genetics in Medicine, 2001