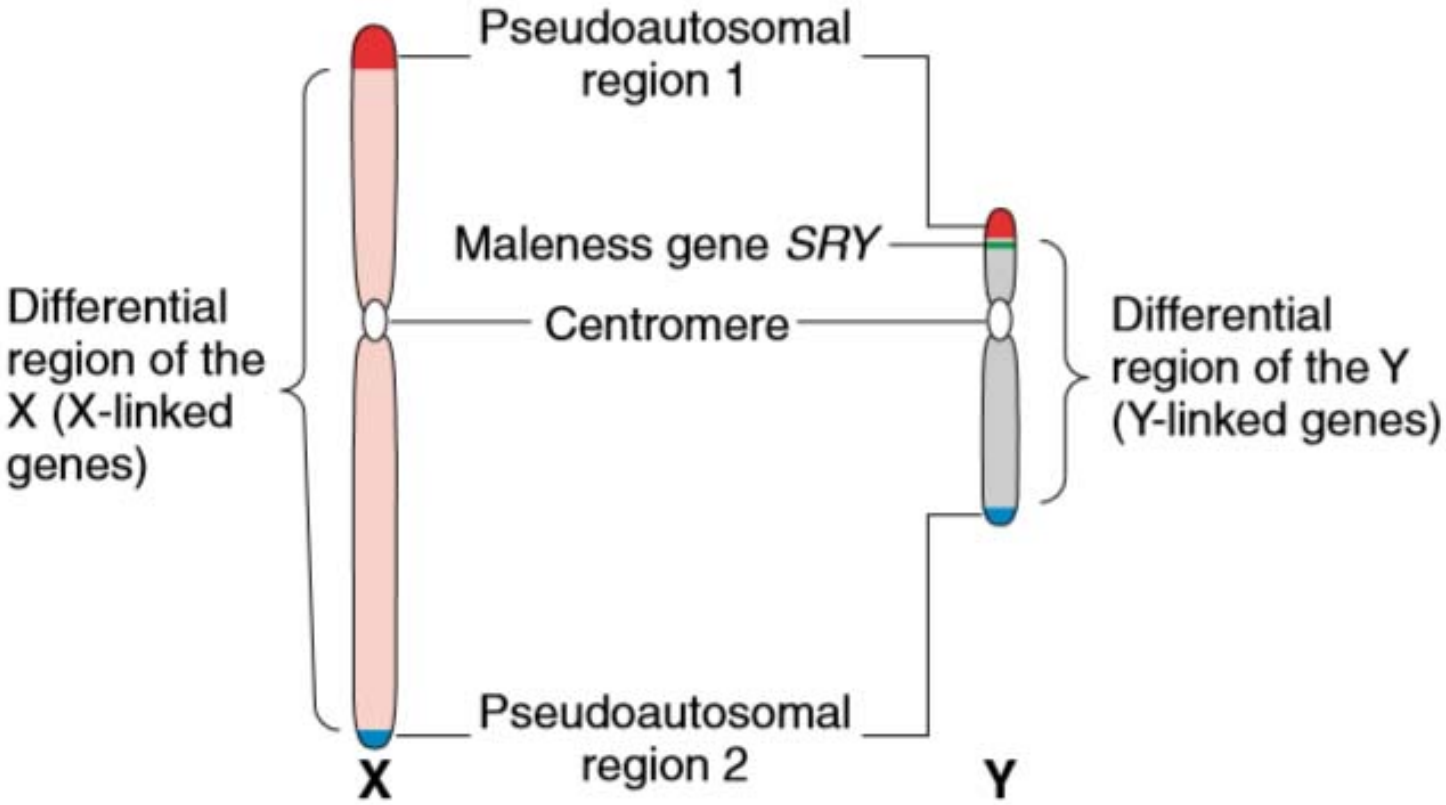
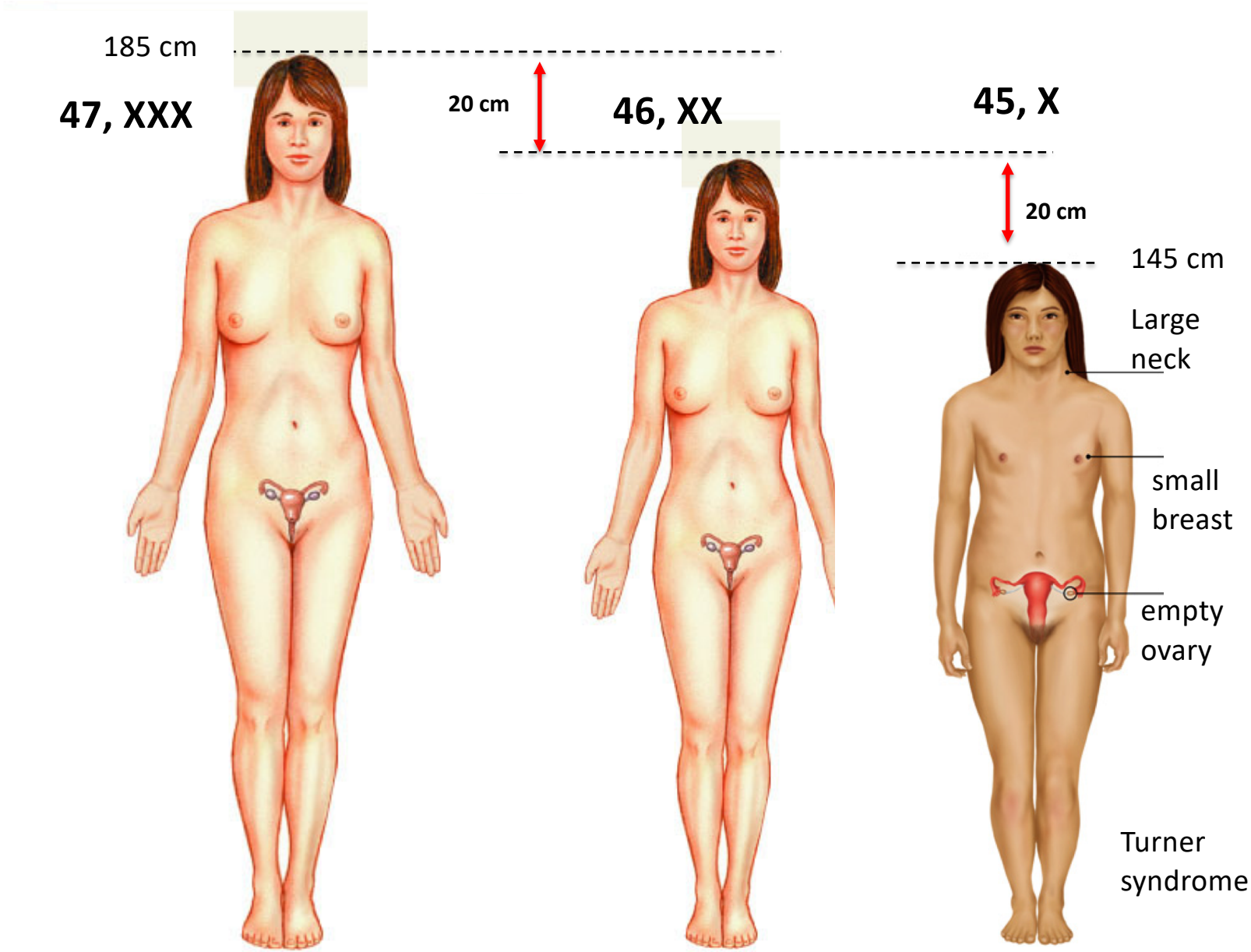
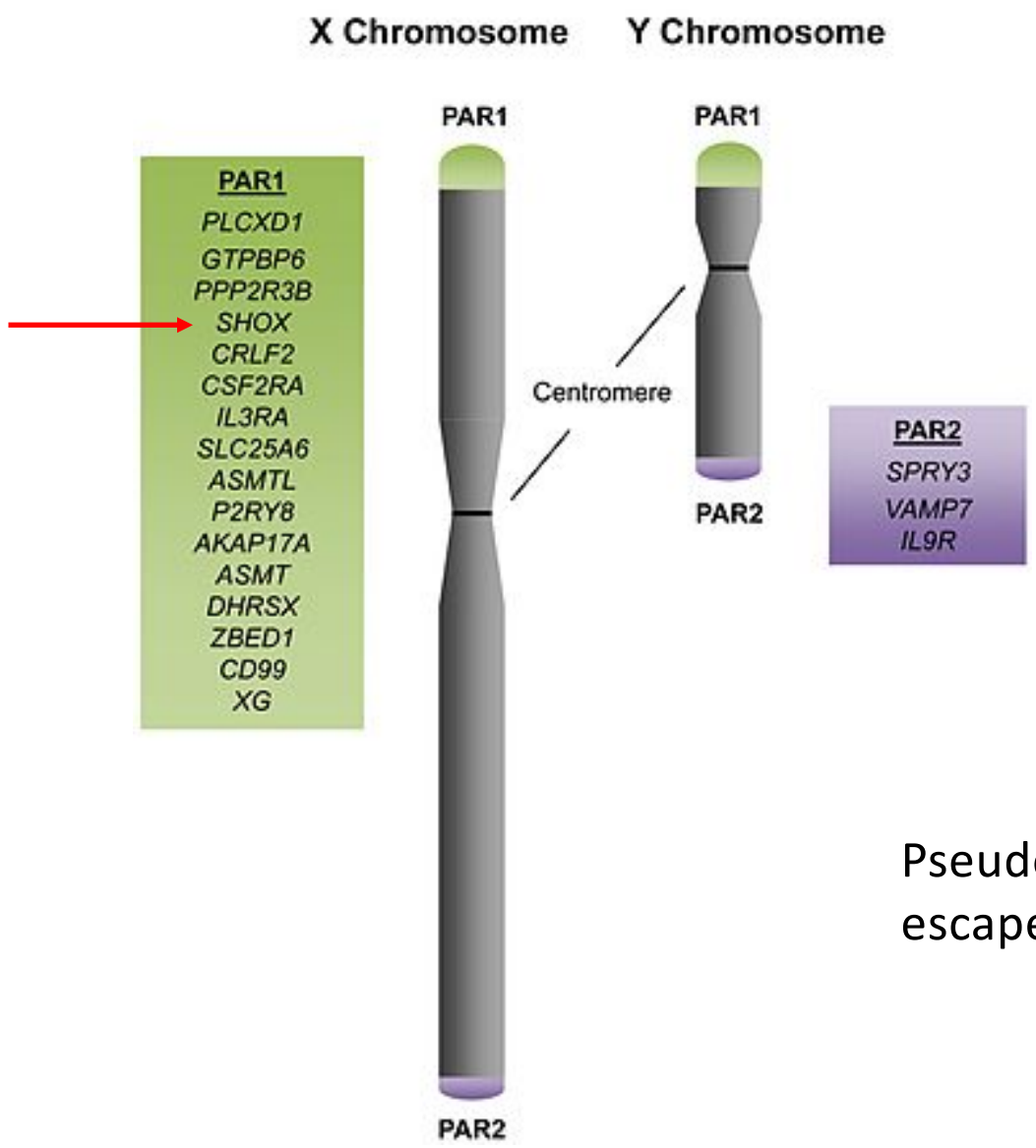
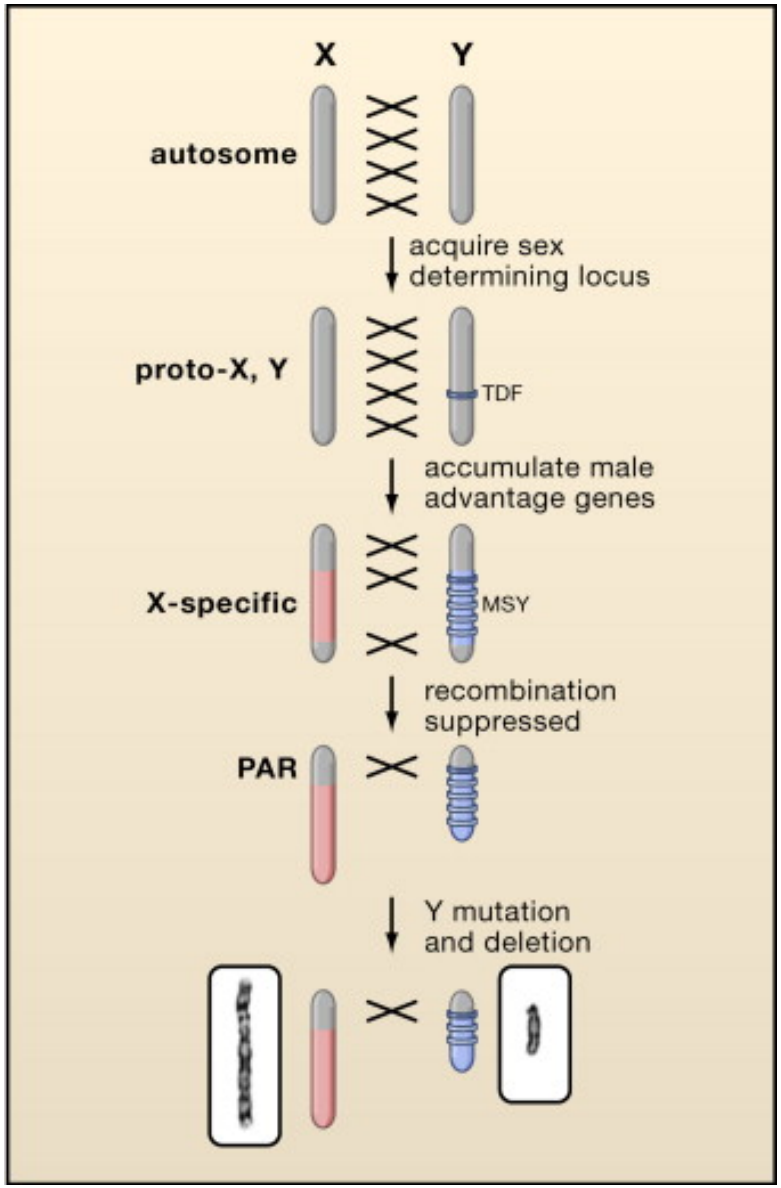

Pseudoautosomal region 1
Maleness gene *SRY*
Centromere
Pseudoautosomal region 2
Differential region of the X (X-linked genes)
Differential region of the Y (Y-linked genes)
X
Y

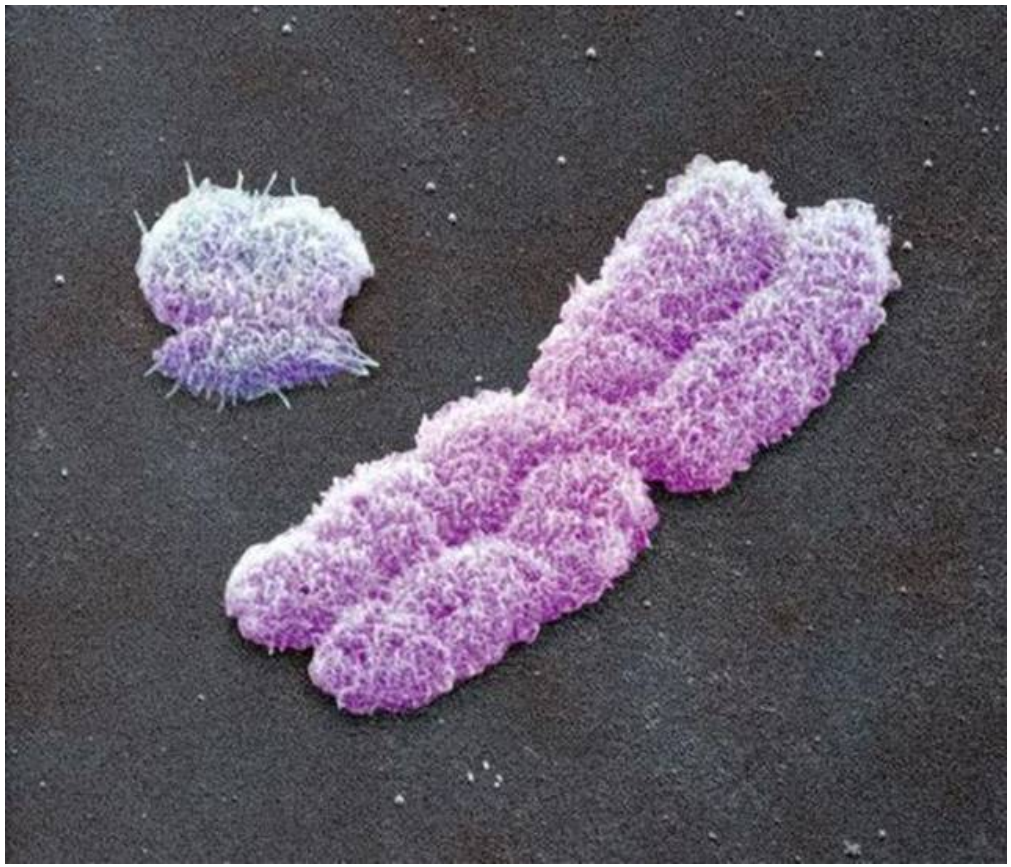

185 cm
47, XXX
20 cm
46, XX
45, X
20 cm
145 cm
Large neck
small breast
empty ovary
Turner syndrome

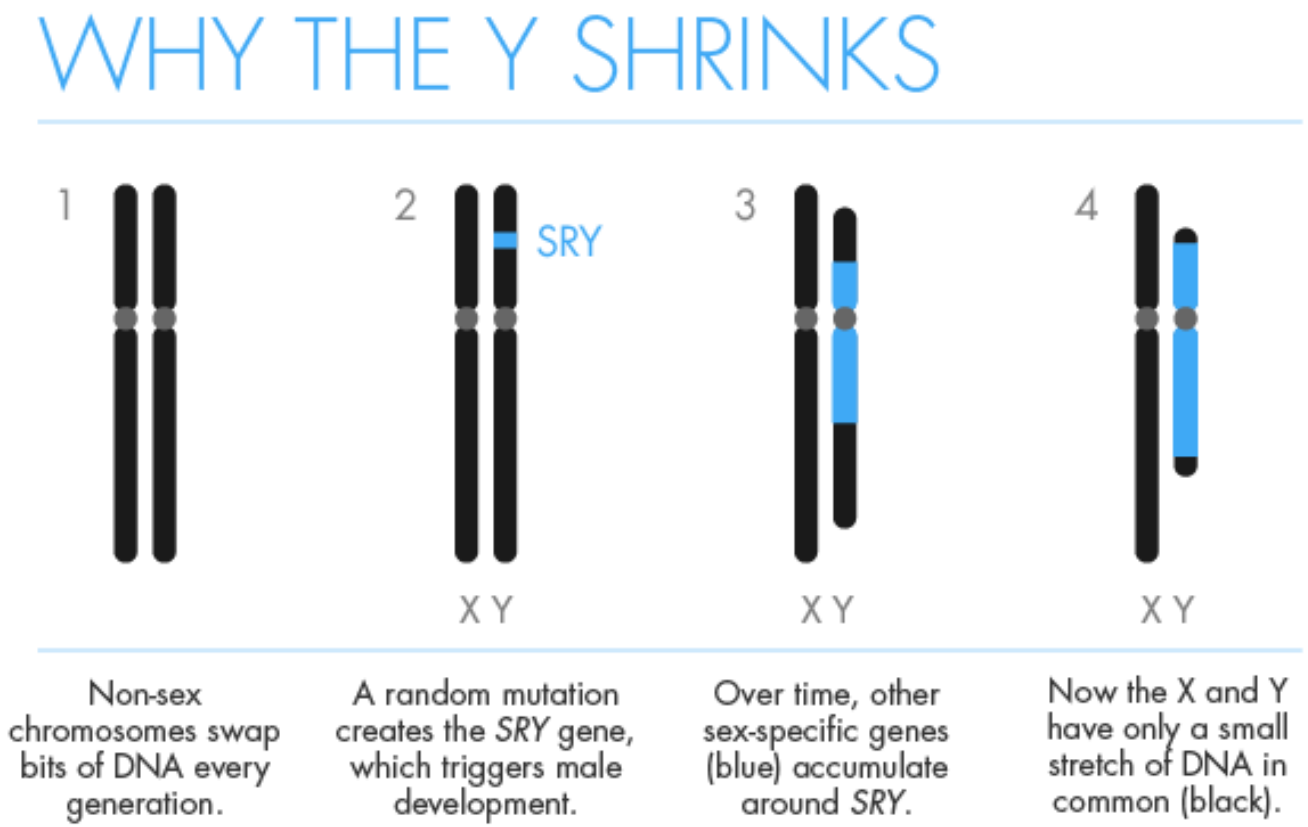


~$2 \cdot 10^6$ bp

Pseudoautosomal regions escape inactivation.

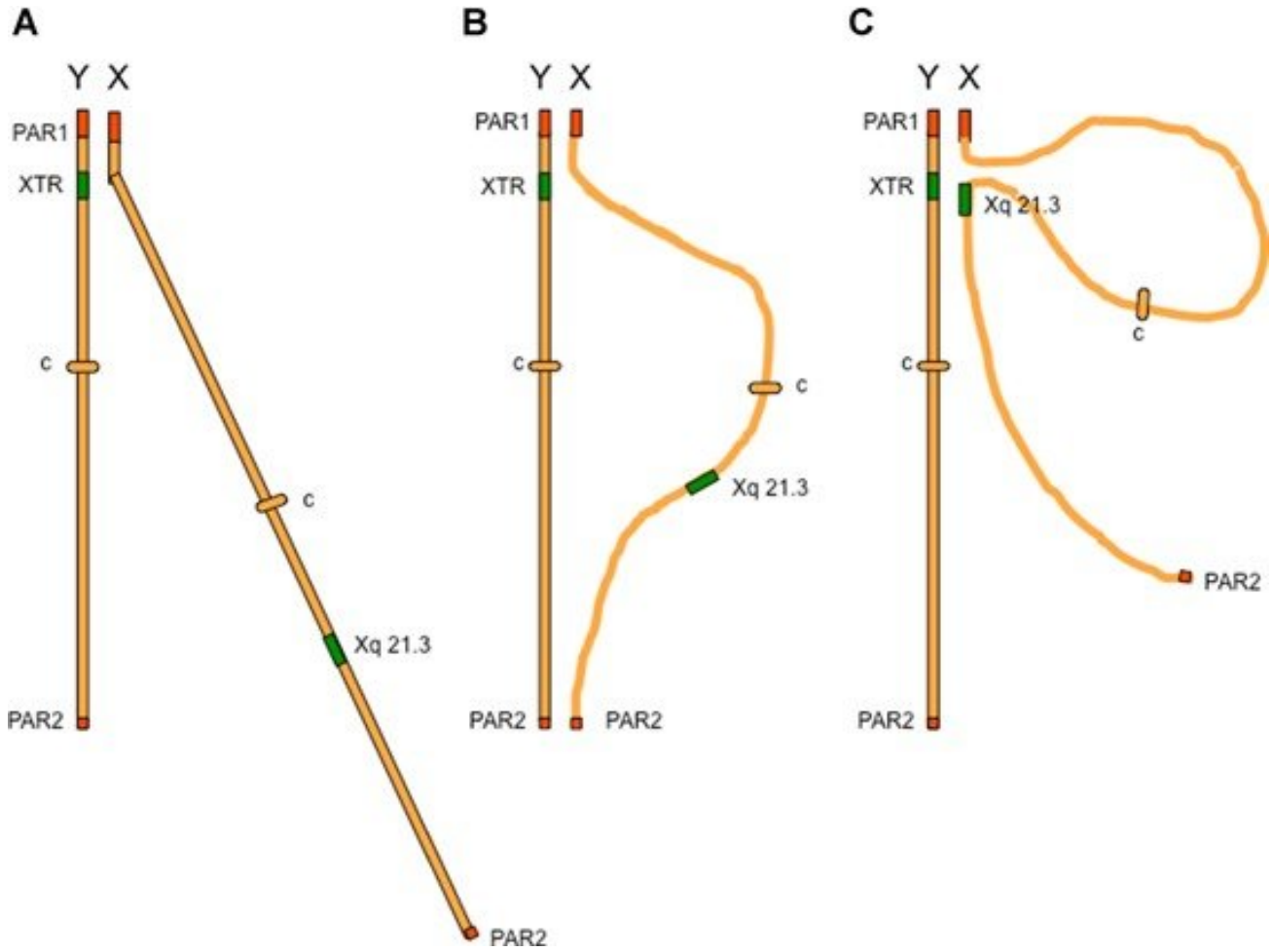

X
Y
autosome
acquire sex determining locus
proto-X, Y
TDF
accumulate male advantage genes
X-specific
MSY
recombination suppressed
PAR
Y mutation and deletion



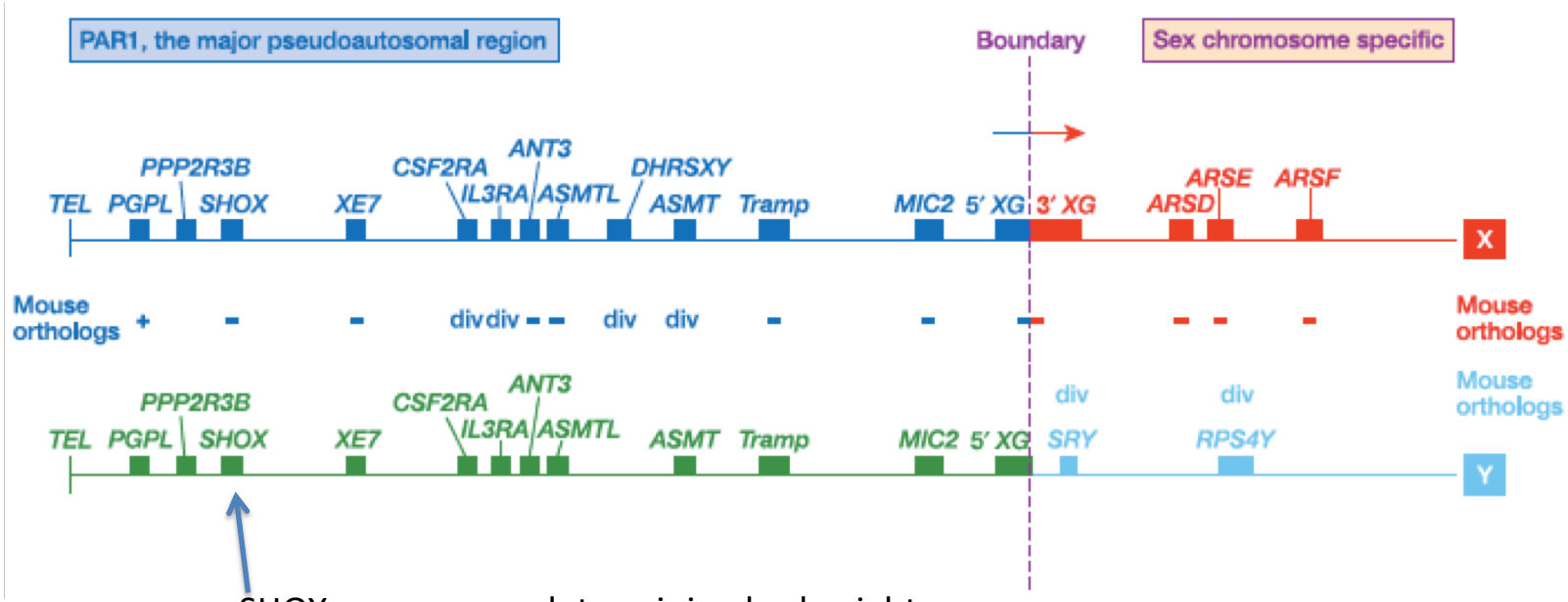


The X and Y chromosomes began life as a matched set of non-sex chromosomes. As sex-specific genes began clustering on the Y chromosome, pieces of the Y flipped around, preventing the X and Y from pairing. No longer able to swap material with the X, the Y couldn't repair itself and began to lose parts.

Male meiosis



Organization and evolutionary instability of the major human pseudoautosomal region (PAR1).

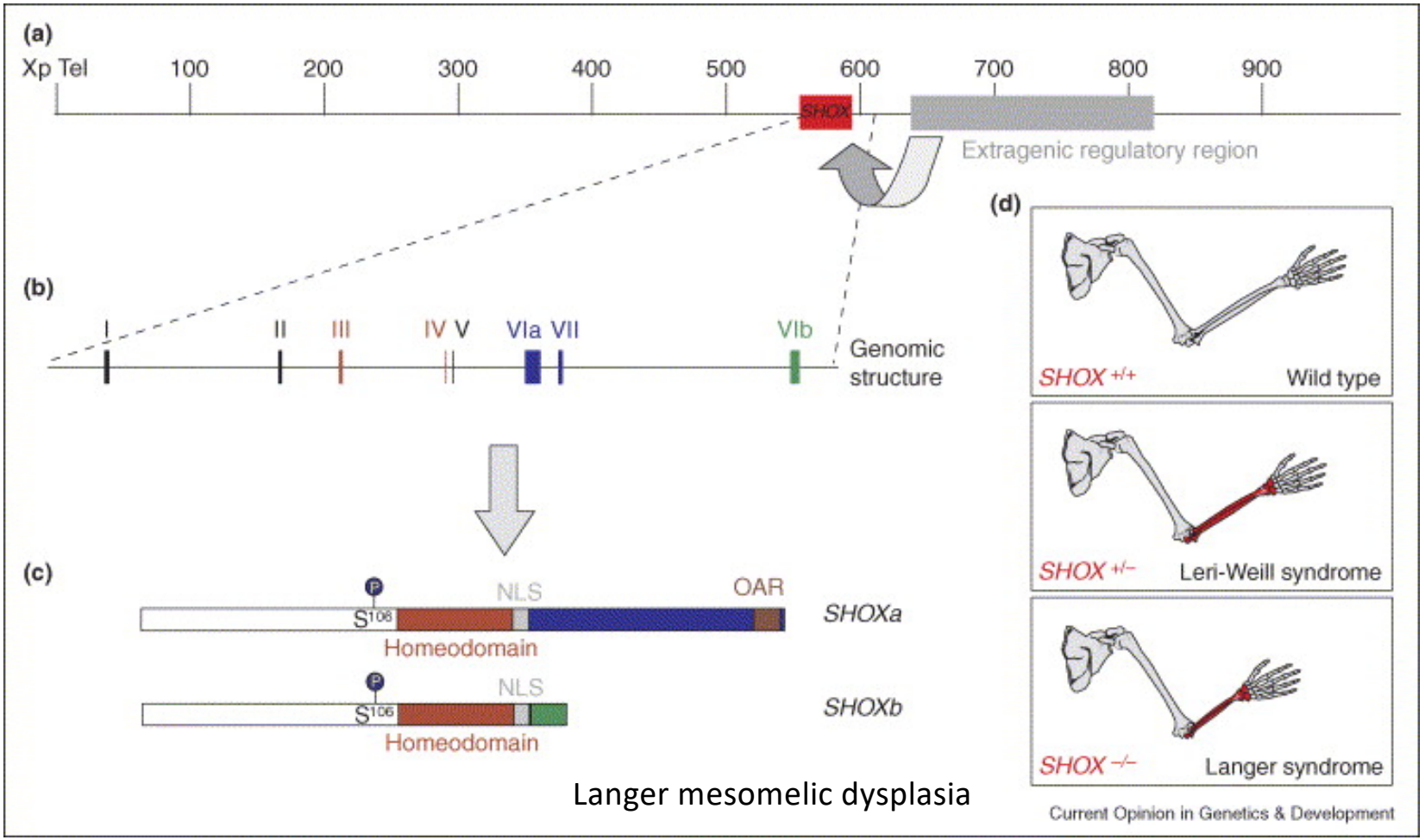


SHOX gene : gene determining body eight

45, X0 → 145 cm

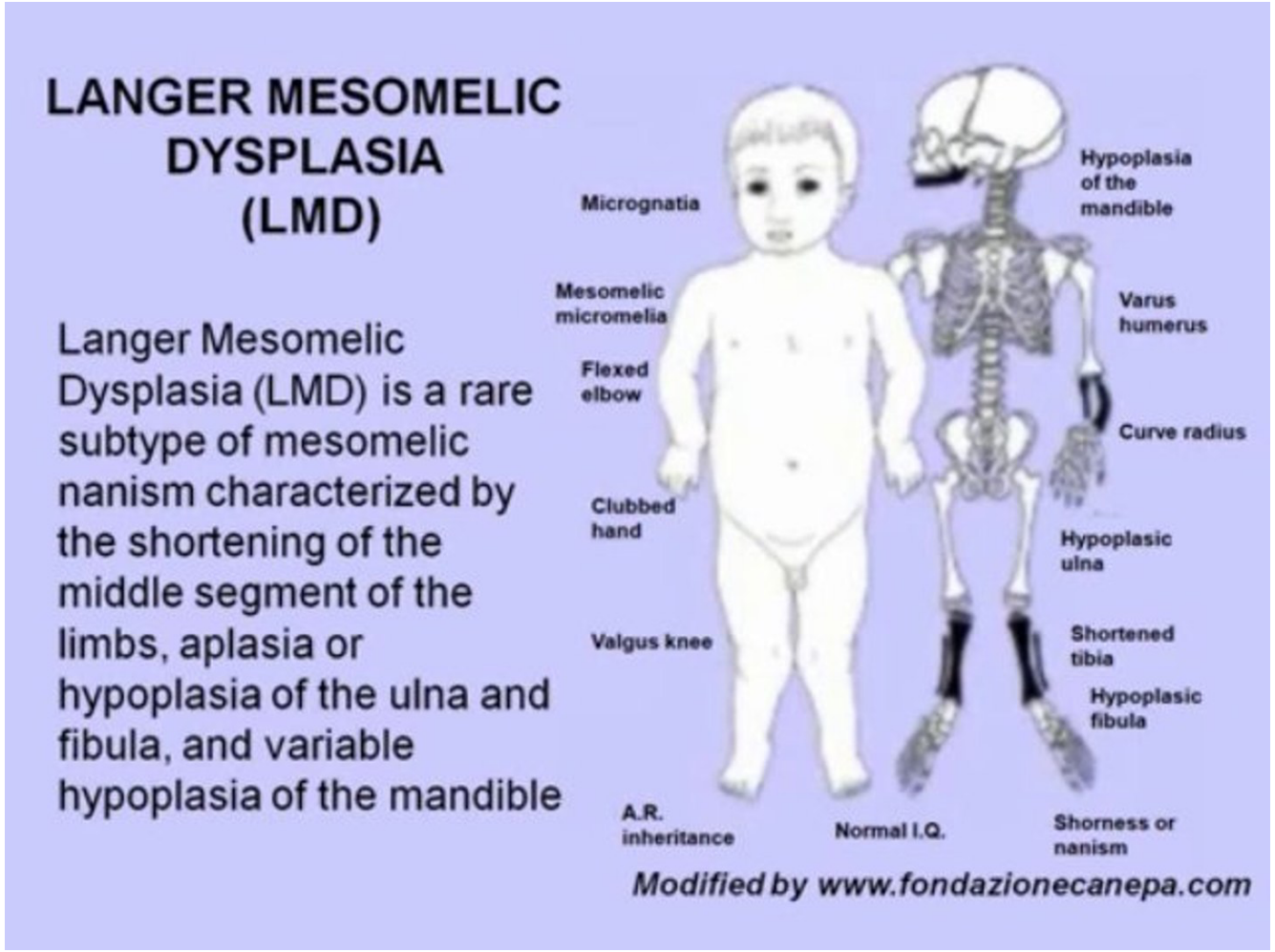
46, XX → 165 cm

47, XXX → 185 cm

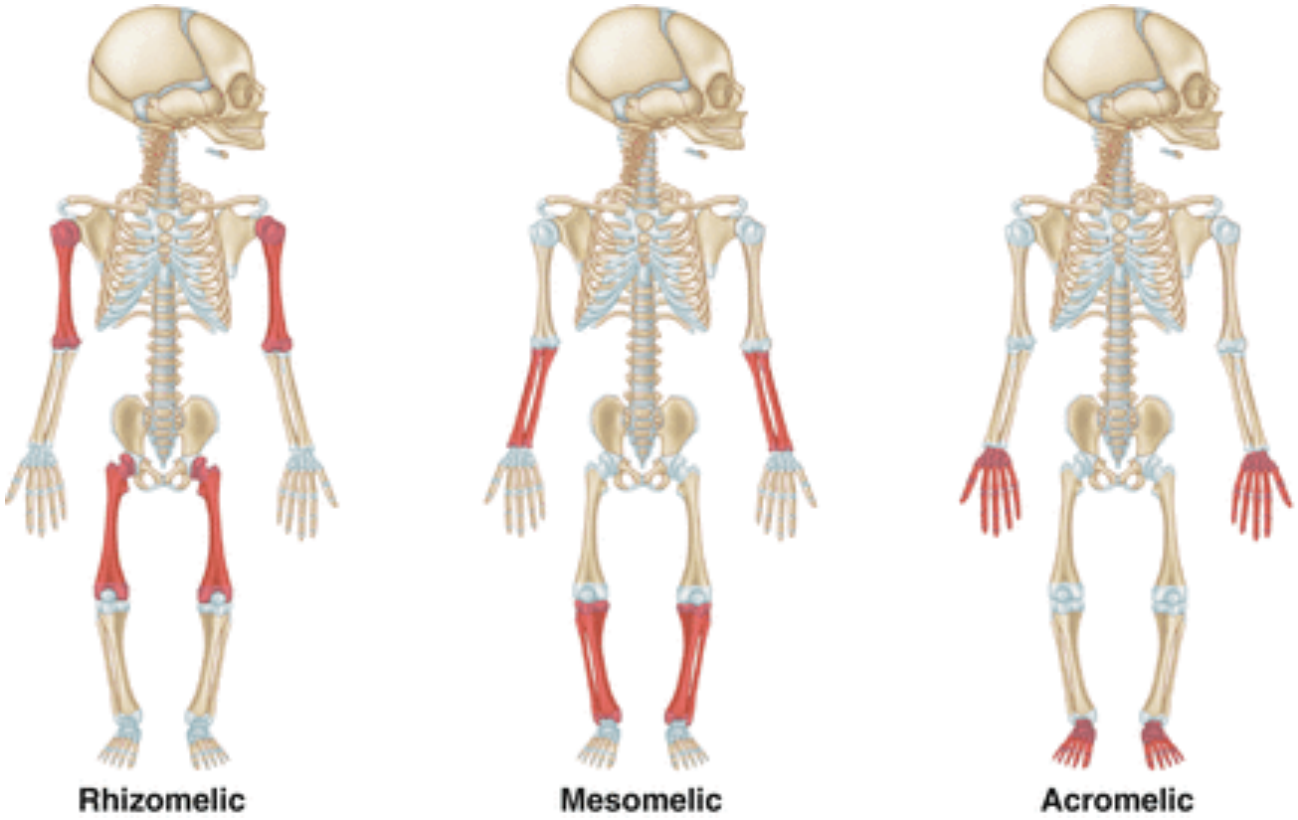


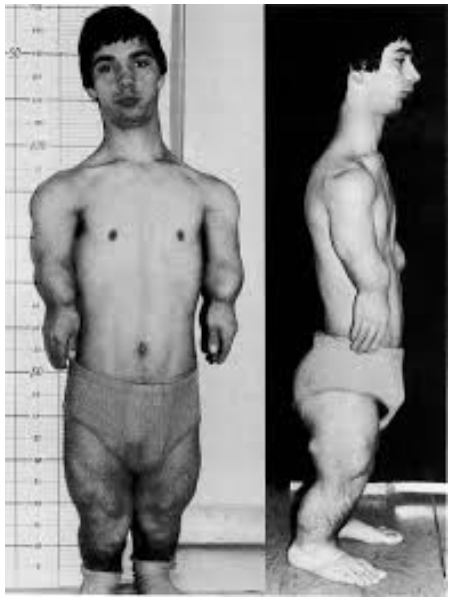
Langer mesomelic dysplasia

https://www.dailymotion.com/video/x17hzxl



Mesomelic dysplasia: Langer type





Two patients with Langer type mesomelic dysplasia are reported. This is one of the rare but well differentiated and easily recognizable mesomelic dysplasias.