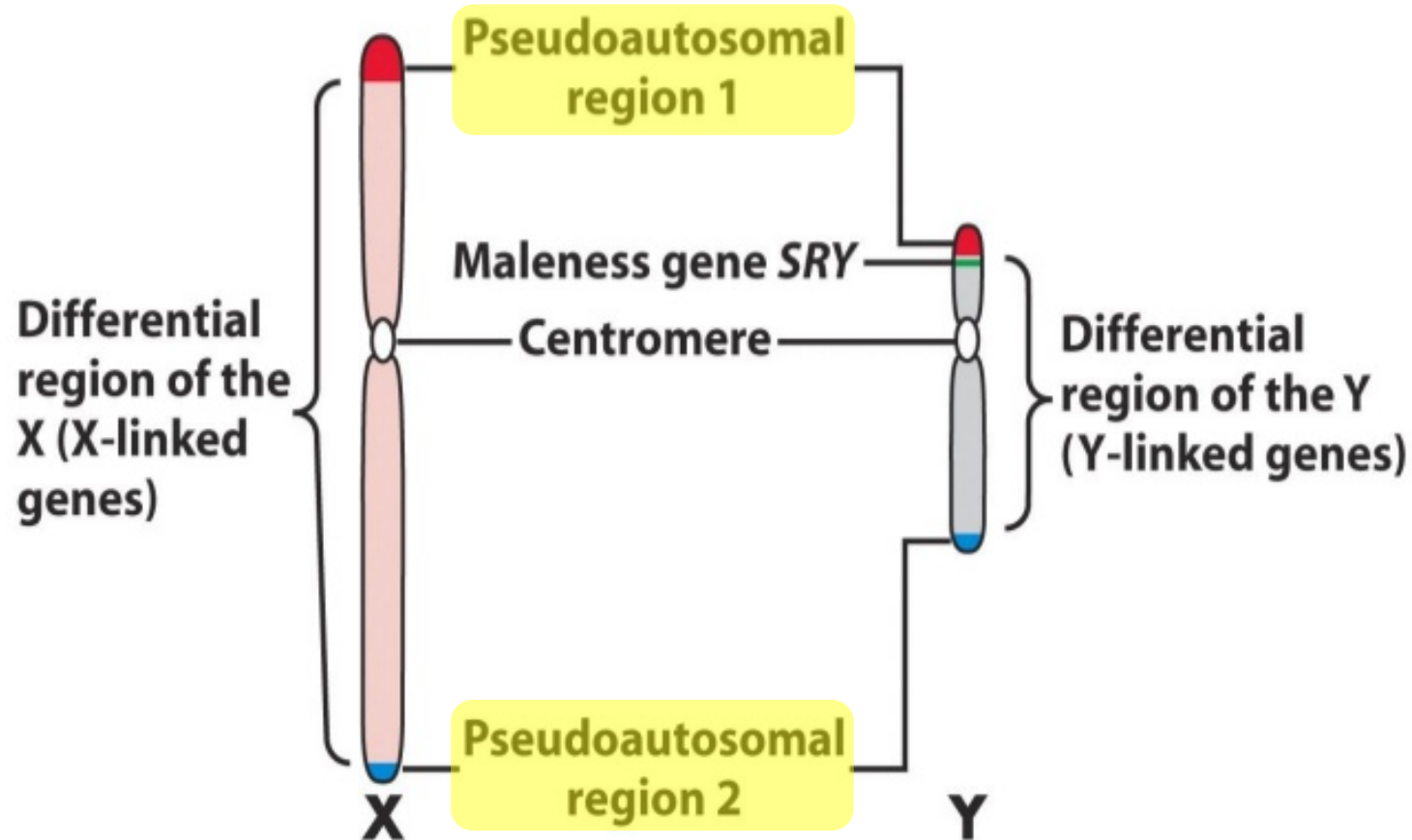
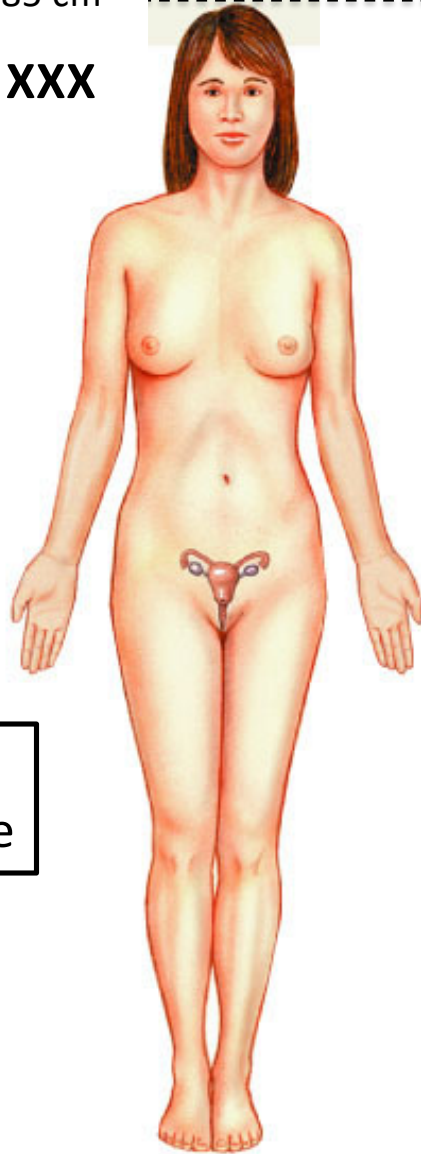


SRY = Sex determining Region on Y chromosome



Differential and pairing regions of human sex

185 cm
47, XXX



20 cm

46, XX



45, X

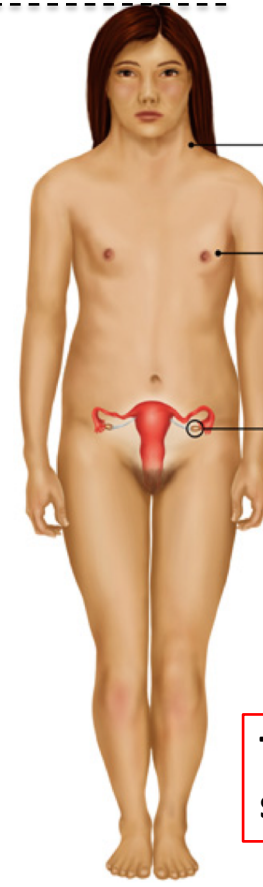
20 cm

145 cm

Large
neck

small
breast

empty
ovary



Dosage effect of
the X chromosome

**Turner
syndrome**

X chromosome dosage effect.



Linda HUNT (actrice)

Syndrome de Turner

45, X0



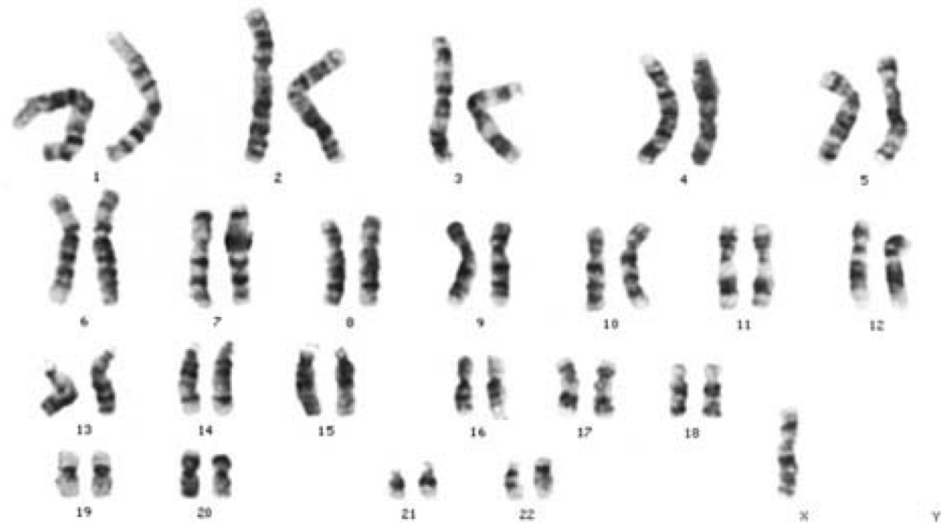
145 cm

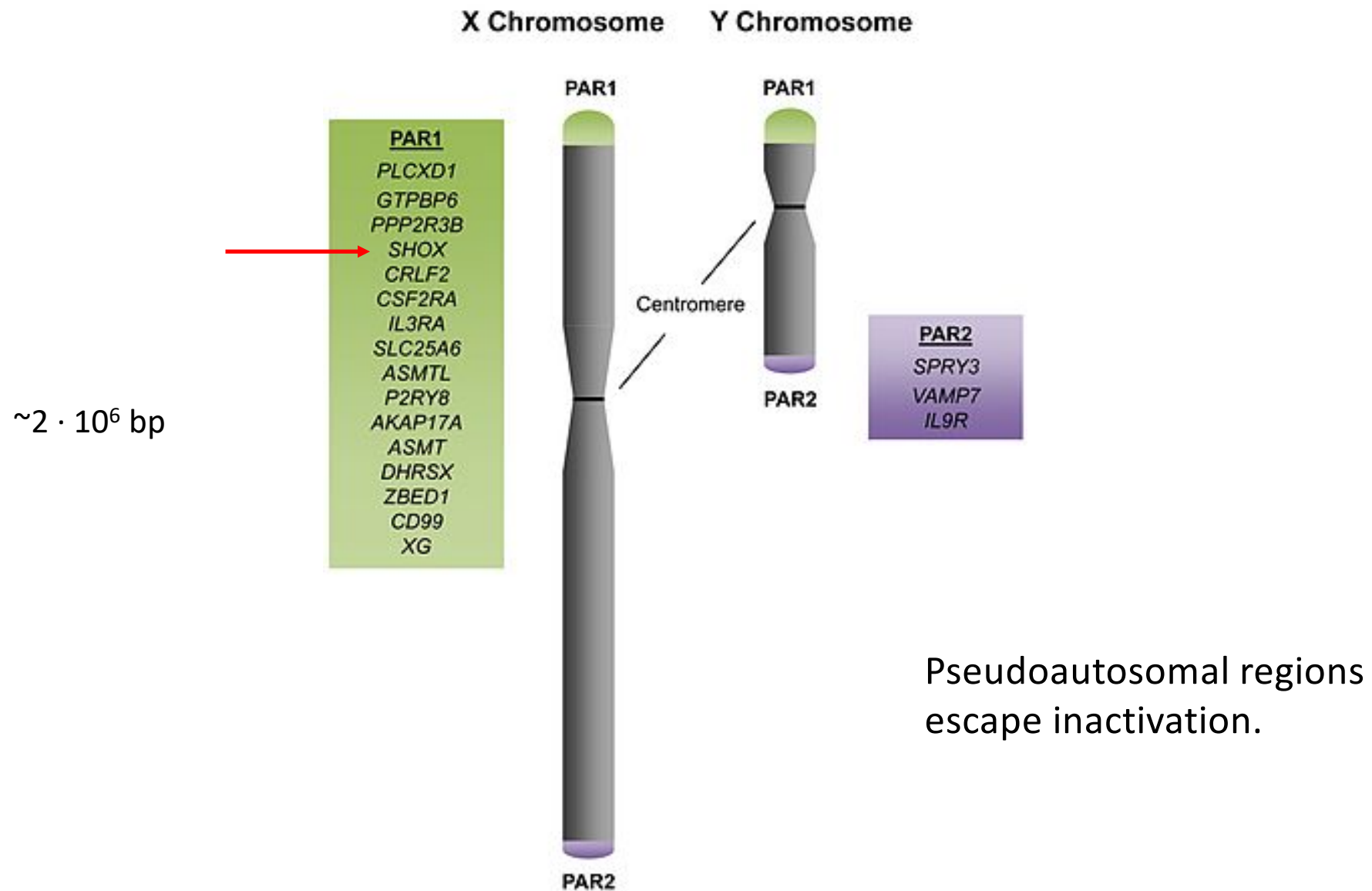
Gene SHOX :

1 copy → 145 cm

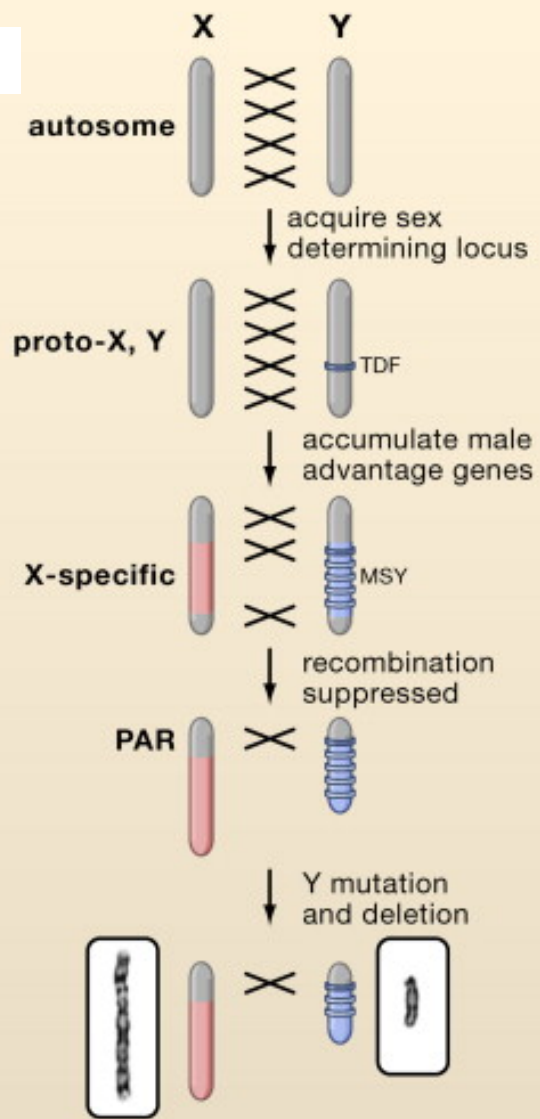
2 copies → 165 cm

3 copies → 185 cm

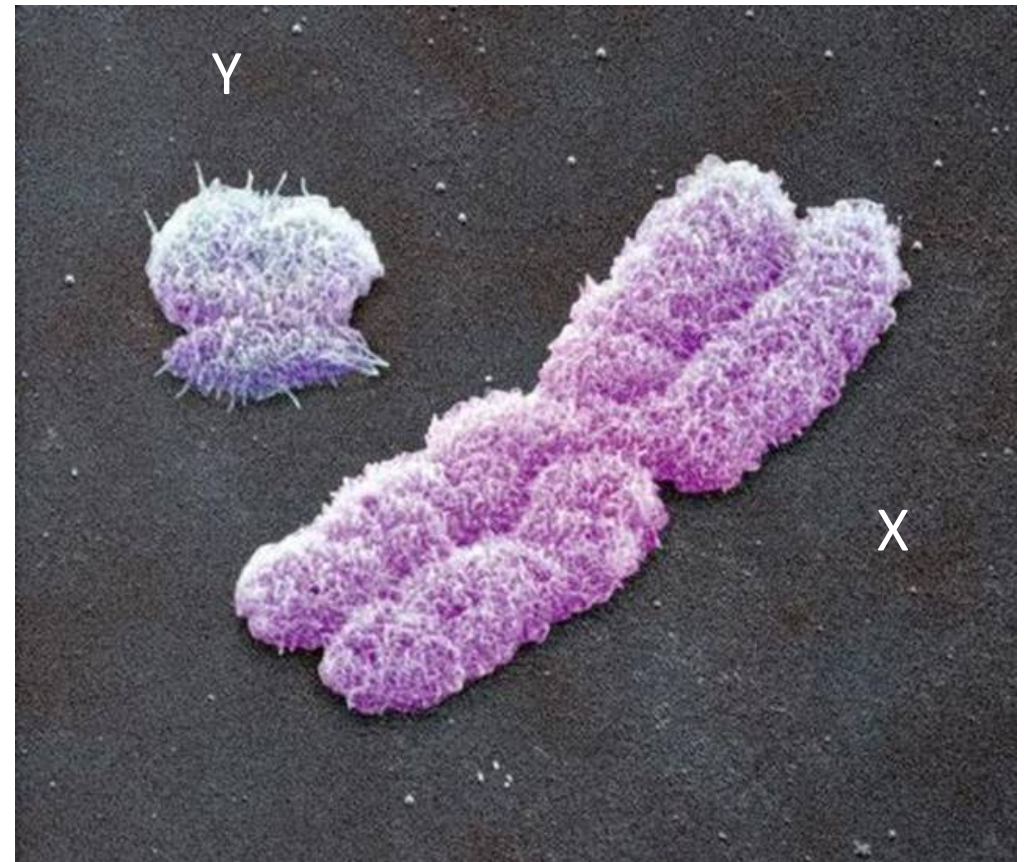




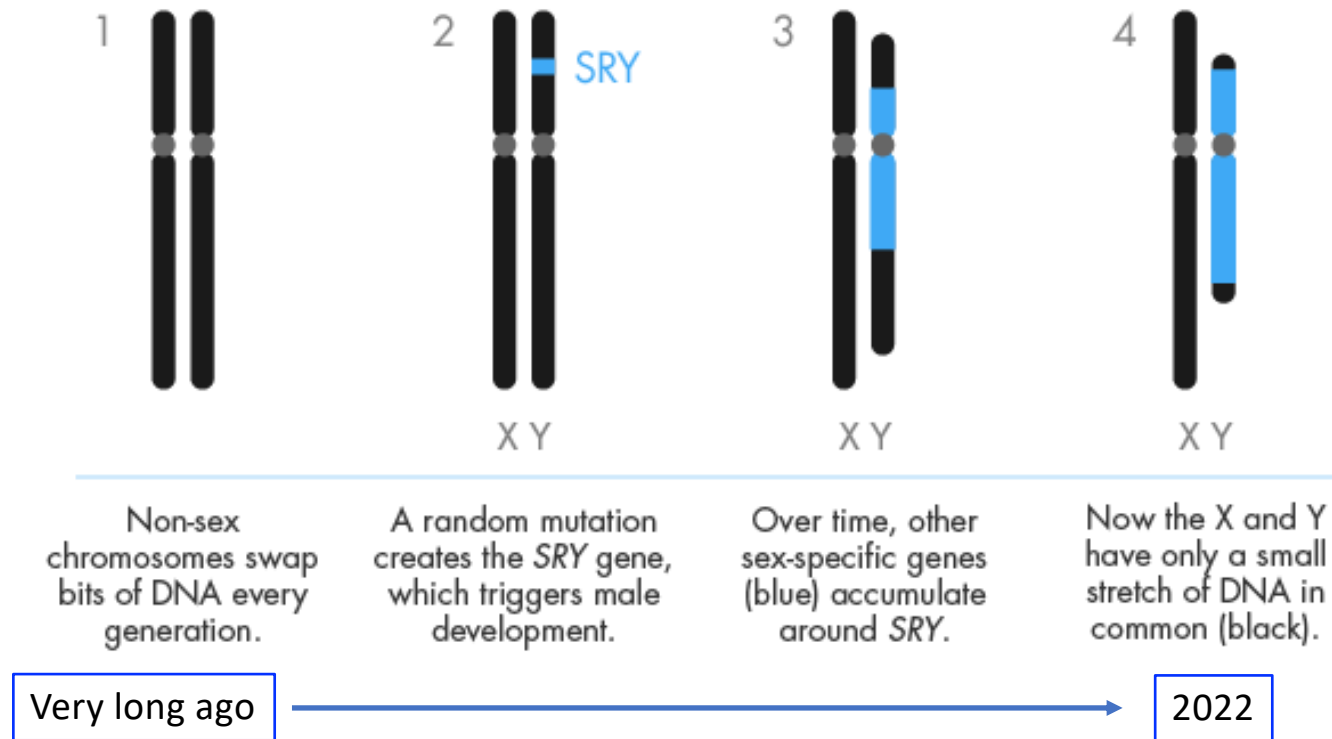
Very long ago



Scanning electron micrograph of X and Y chromosomes



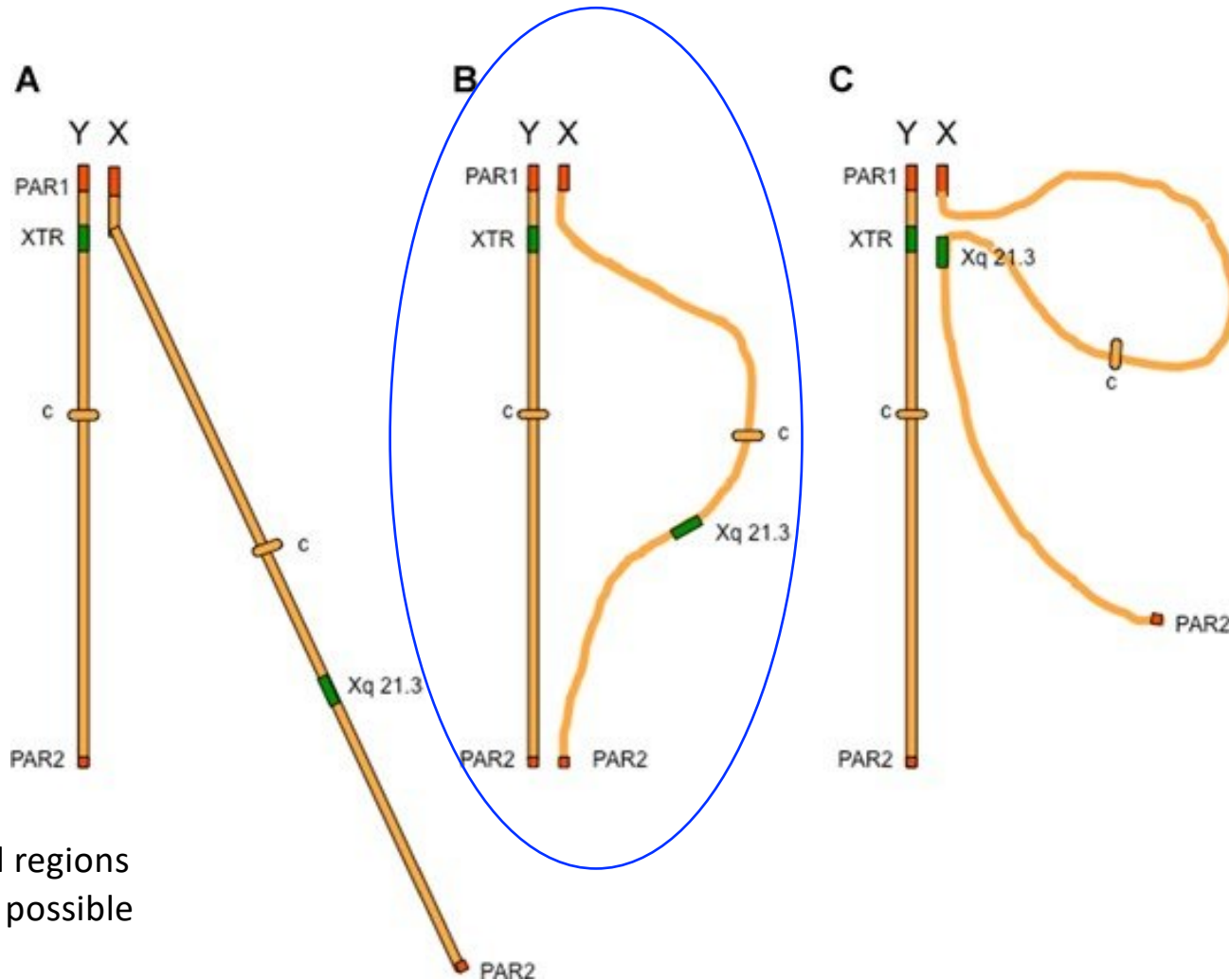
WHY THE Y SHRINKS



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.

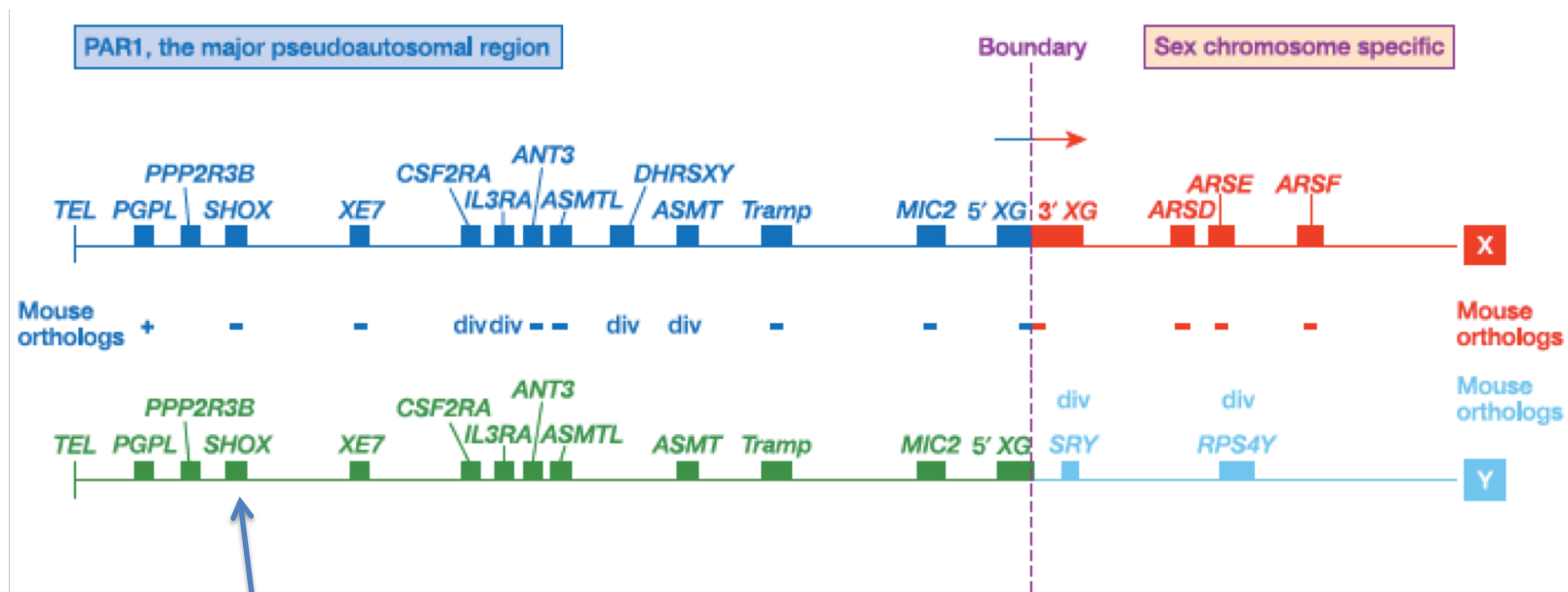
Male meiosis

Meiosis I :
homologous
chromosomes
must pair



The 2 pseudoautosomal regions
make pairing of X and Y possible
during meiosis I.

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



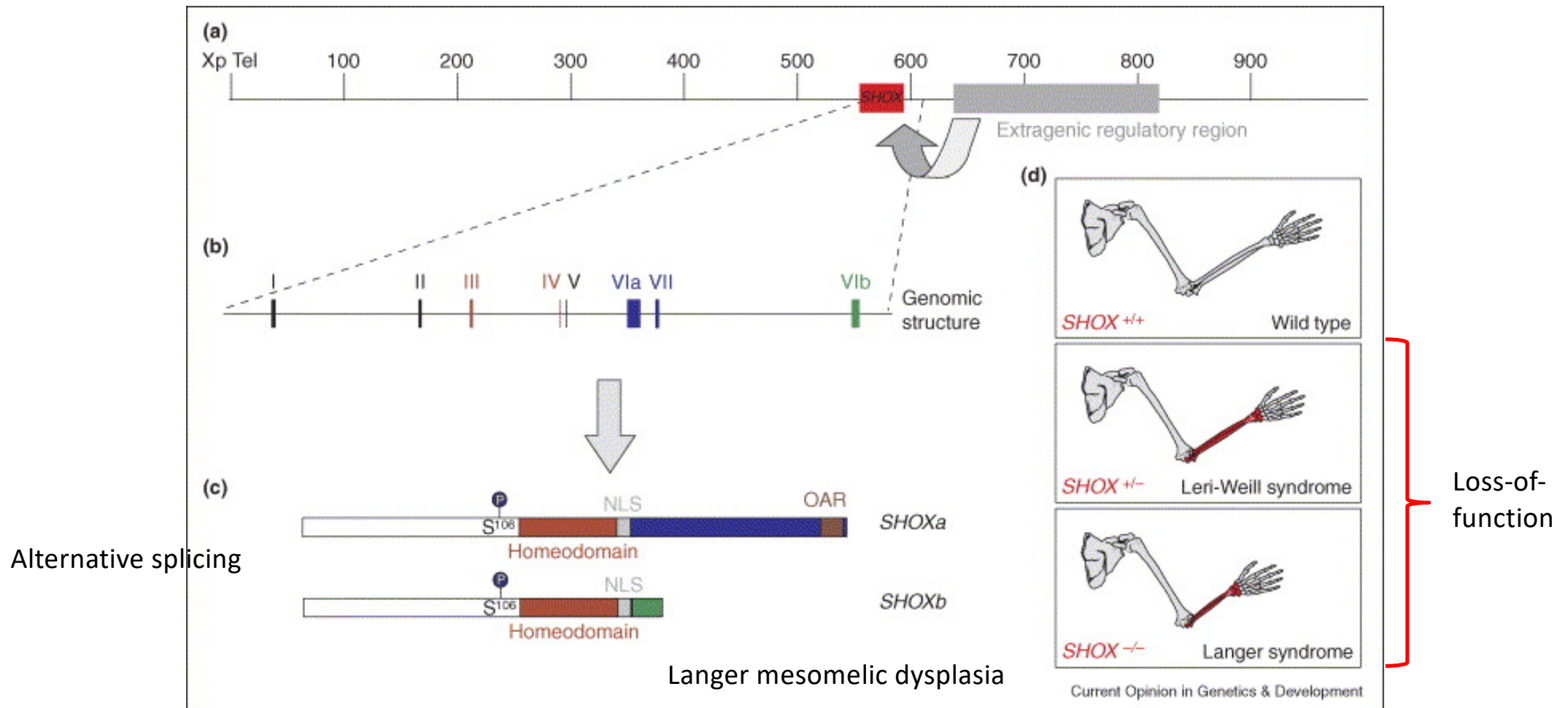
SHOX gene : gene determining body height

45, X0 → 145 cm

46, XX → 165 cm

47, XXX → 185 cm

SHOrt stature on X

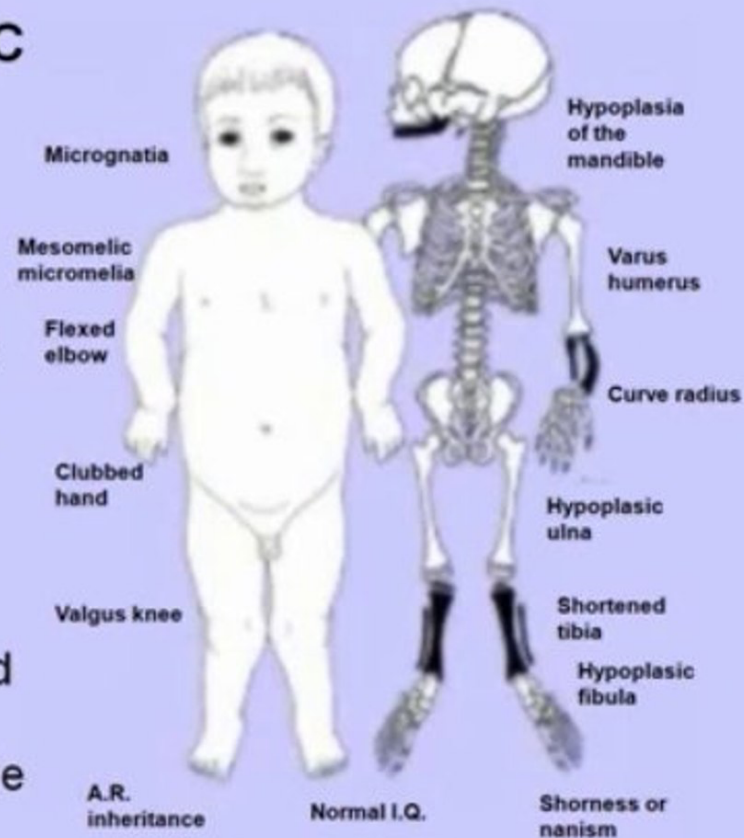


The shox protein is a **transcription factor**.

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

LANGER MESOMELIC DYSPLASIA (LMD)

Langer Mesomelic Dysplasia (LMD) is a rare subtype of mesomelic nanism characterized by the shortening of the middle segment of the limbs, aplasia or hypoplasia of the ulna and fibula, and variable hypoplasia of the mandible

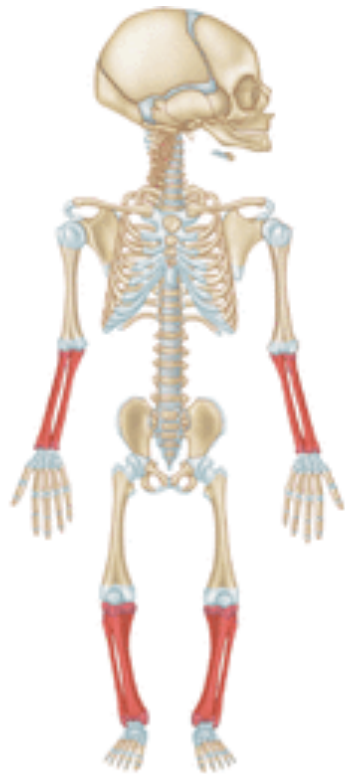


Modified by www.fondazioneacanepa.com

3 types of dysplasia



Rhizomelic

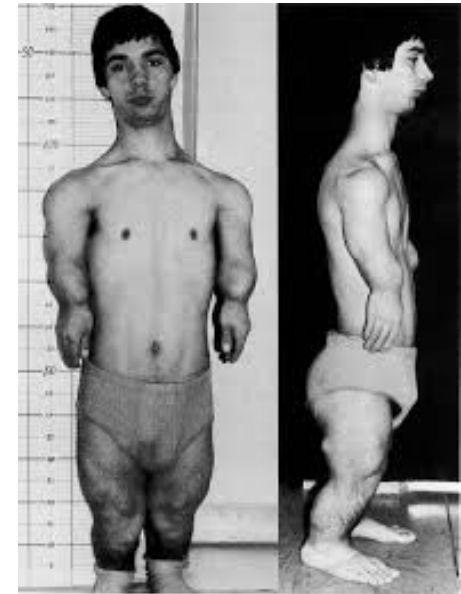


Mesomelic



Acromelic

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.



Clinical appearance of the proband, aged 12 years.

Rhizomelic and mesomelic limb shortening is present. Syndactyly and camptodactyly is present in the hands. Proximal placement of the big toes is evident.