

O N L U S



Associazione
Nazionale
di Volontariato
Cornelia
de Lange
www.corneliadelange.org

12° Congresso Nazionale sulla Sindrome Cornelia de Lange

Ilaria Parenti

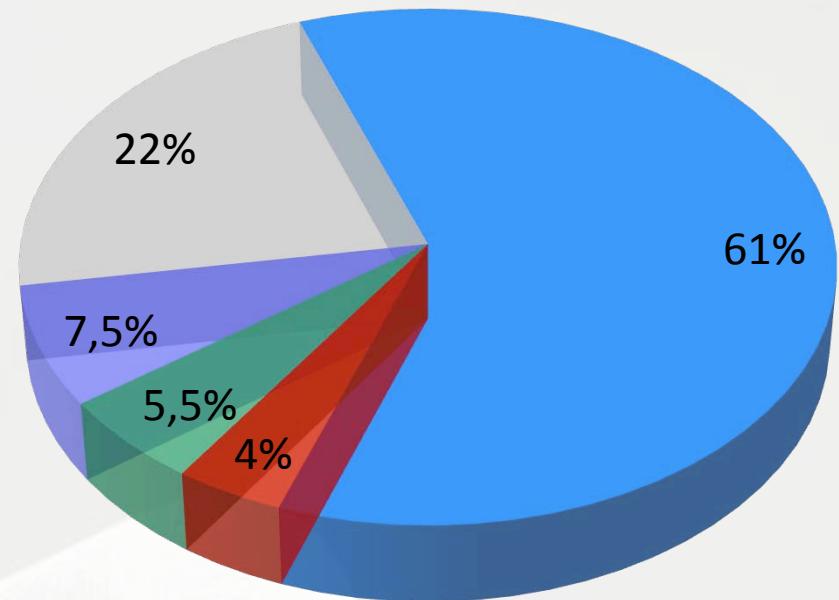
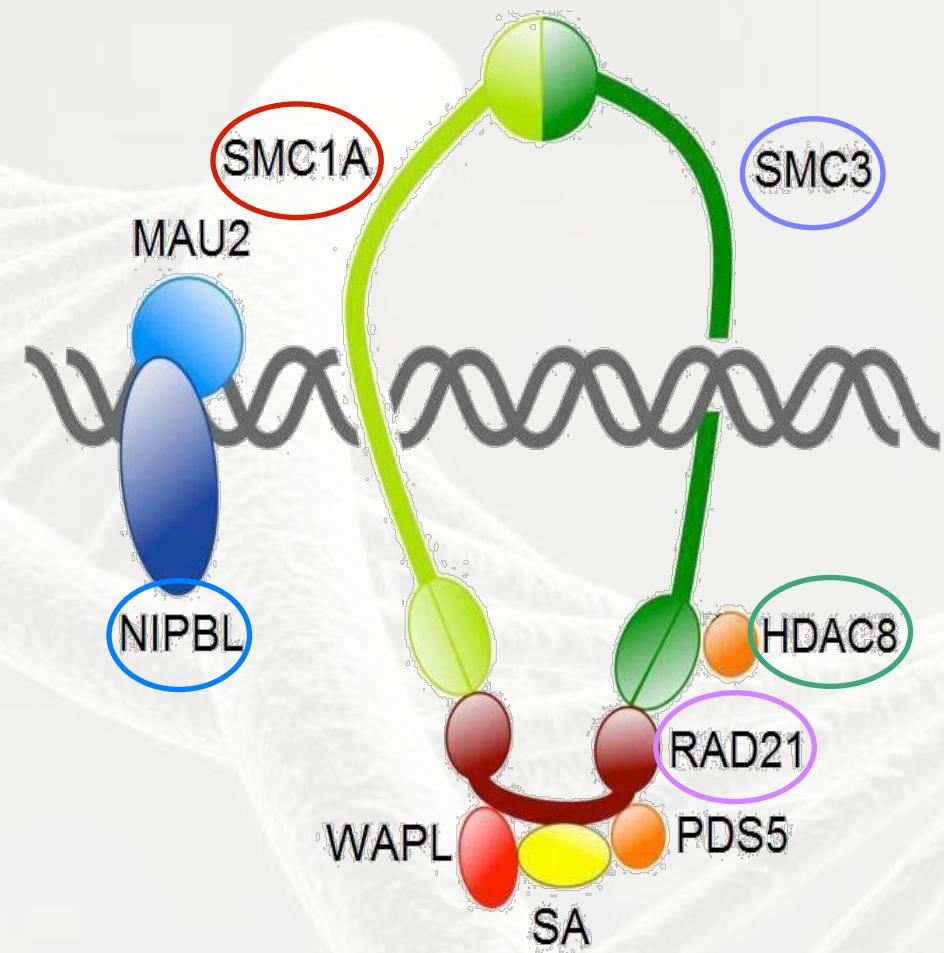
Pesaro, 30 aprile 2017



Institut für
Humangenetik
Lübeck

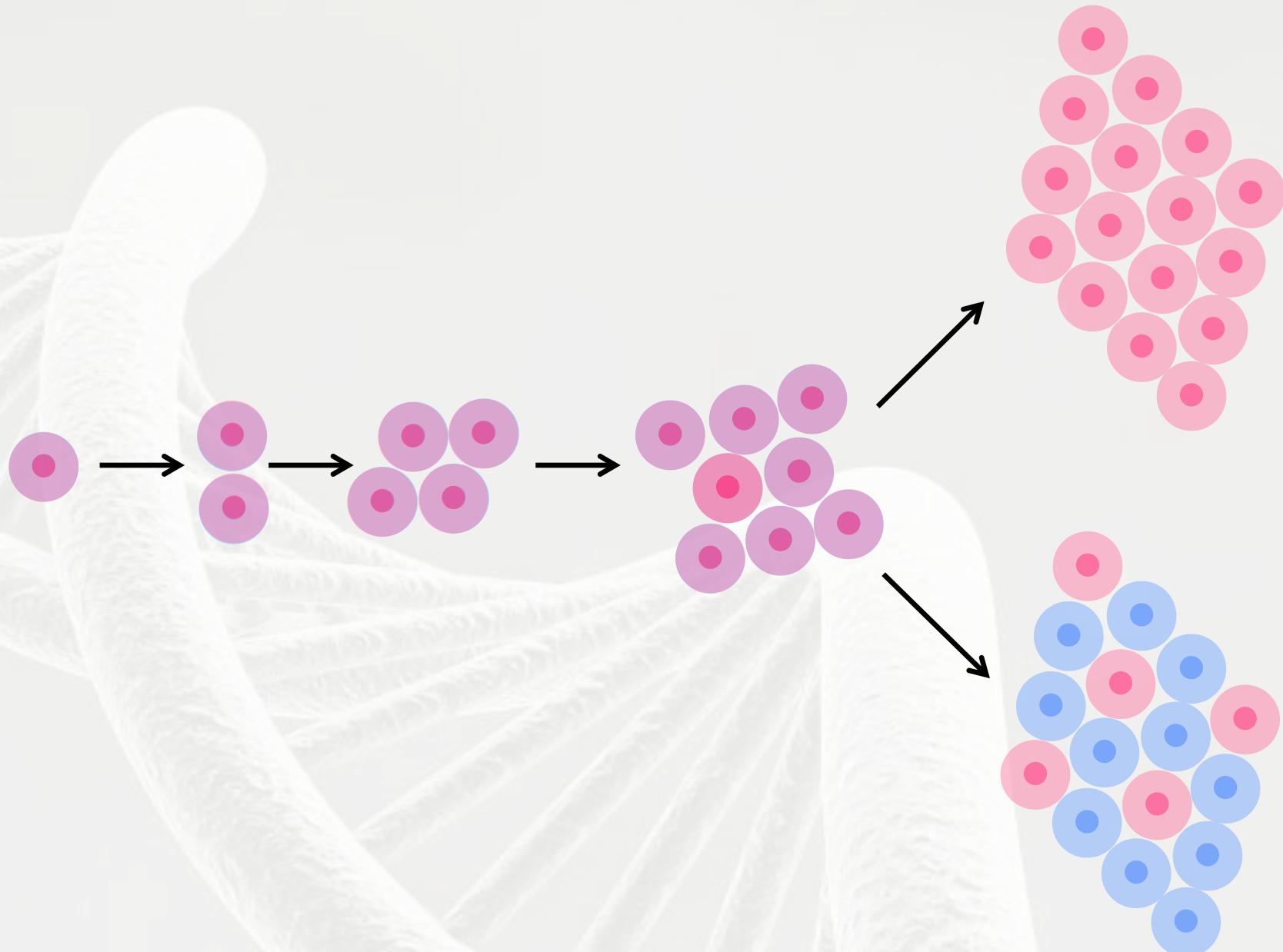


Coesine



- NIPBL*: 122 mutazioni
- SMC1A*: 8 mutazioni
- HDAC8*: 11 mutazioni
- SMC3*: 15 mutazioni
- Ignoto

Mosaicismo



Mosaicismo

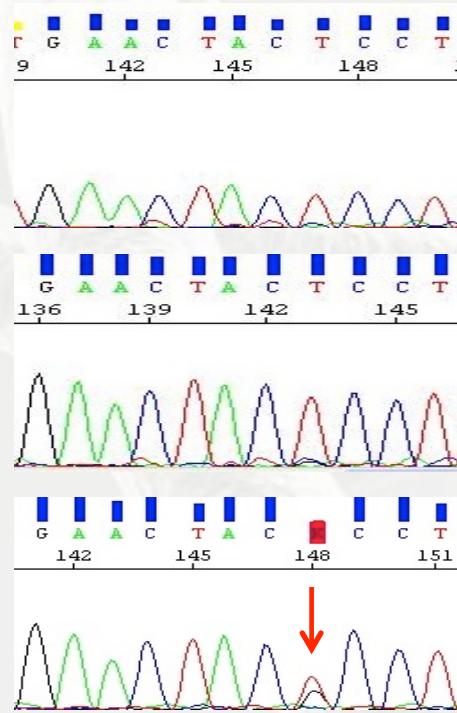
NIPBL: 9 mutazioni a mosaico

131 mutazioni totali (65%)



Susanne

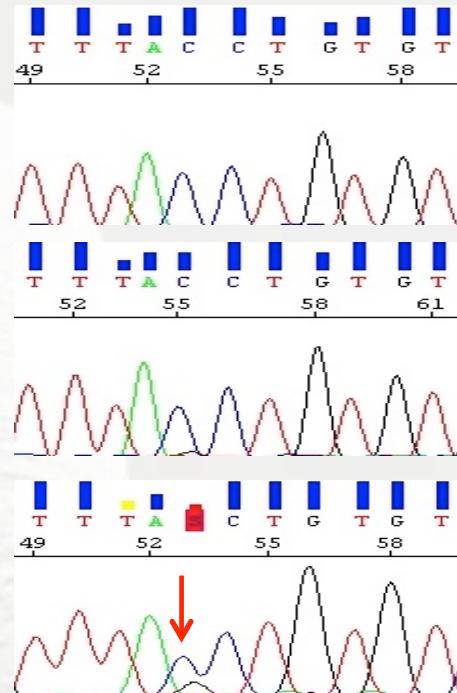
Sangue
Mucosa
Fibroblasti

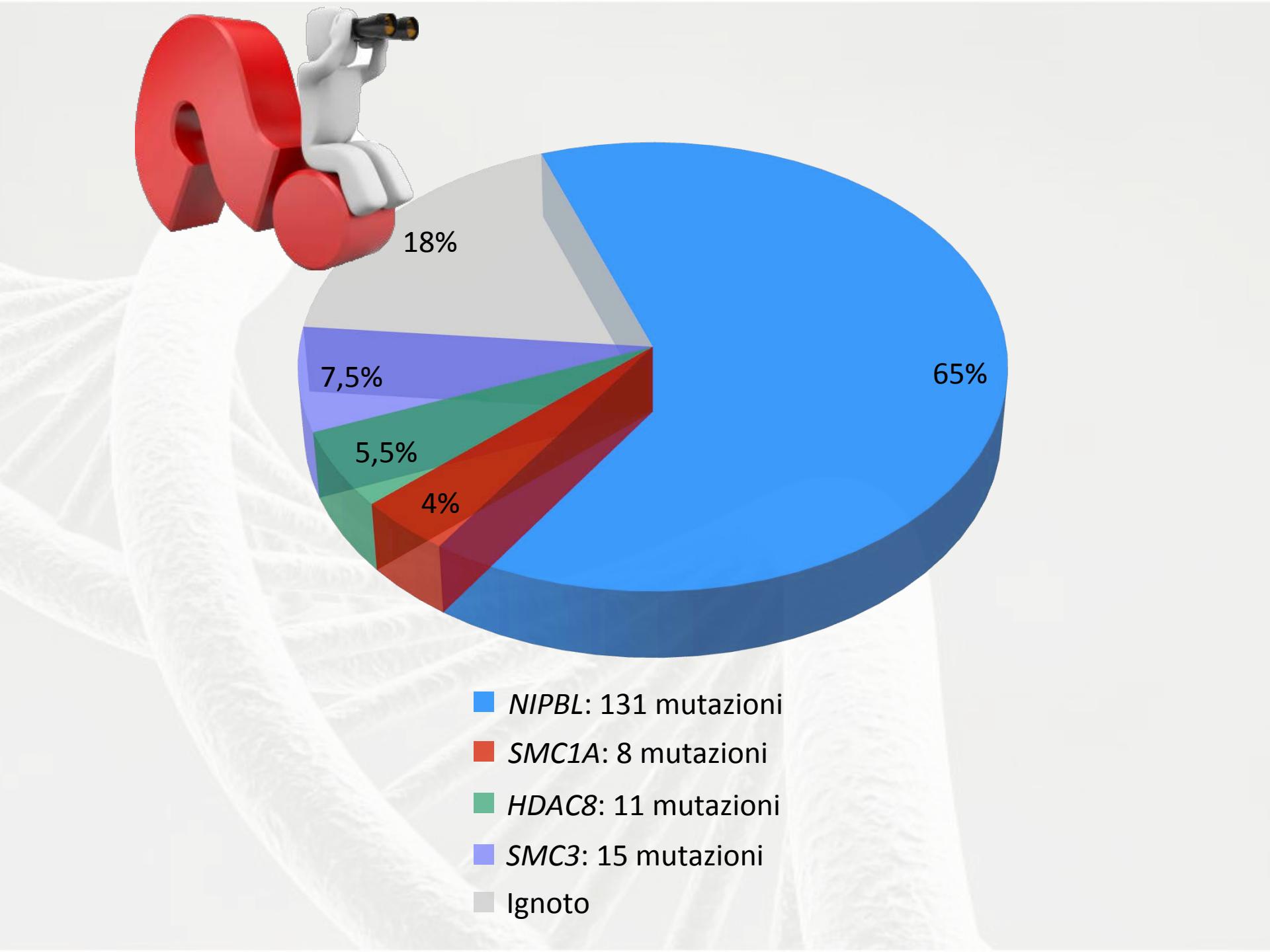


Lena-Marie



Xenia





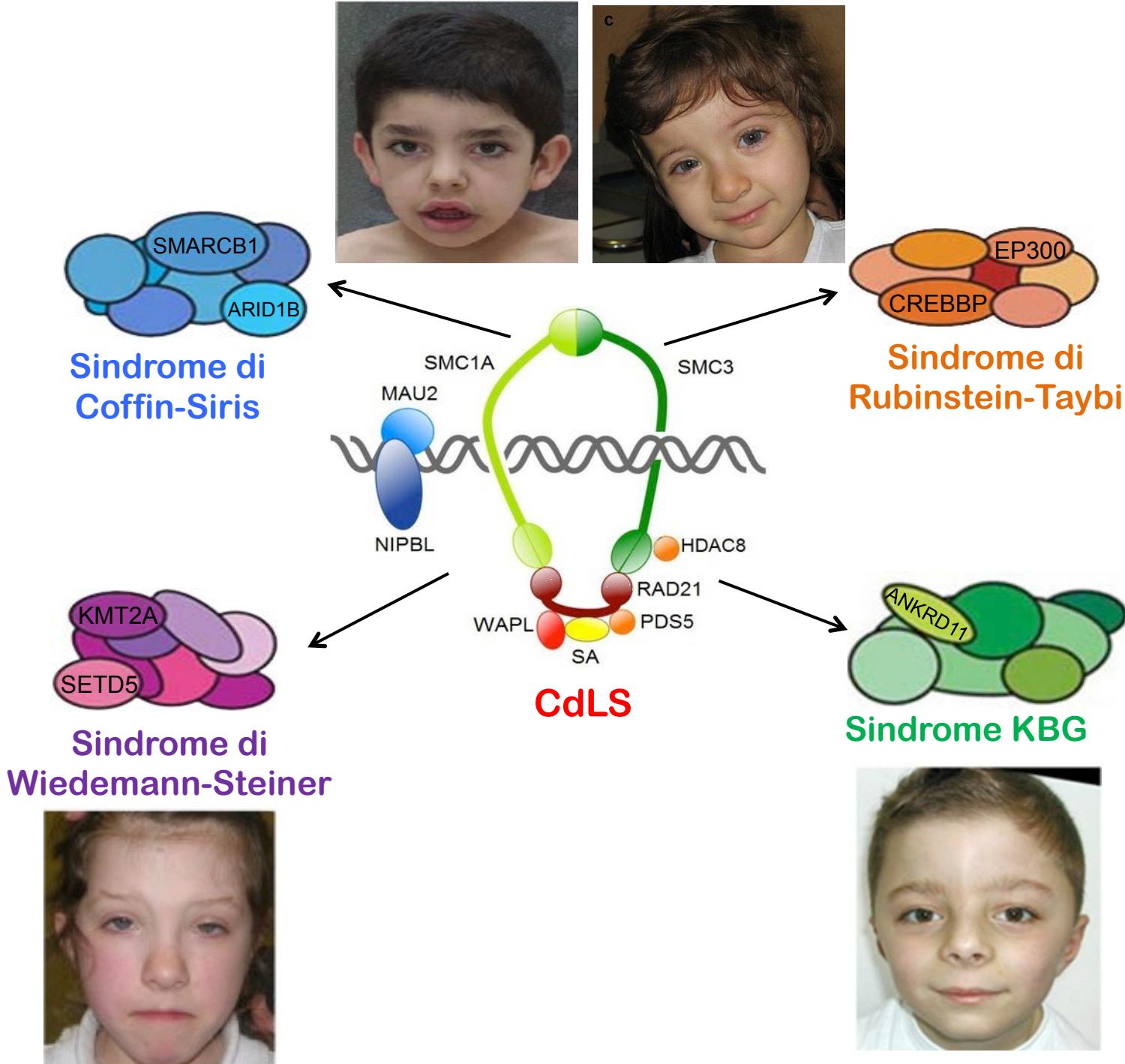
■ *NIPBL*: 131 mutazioni

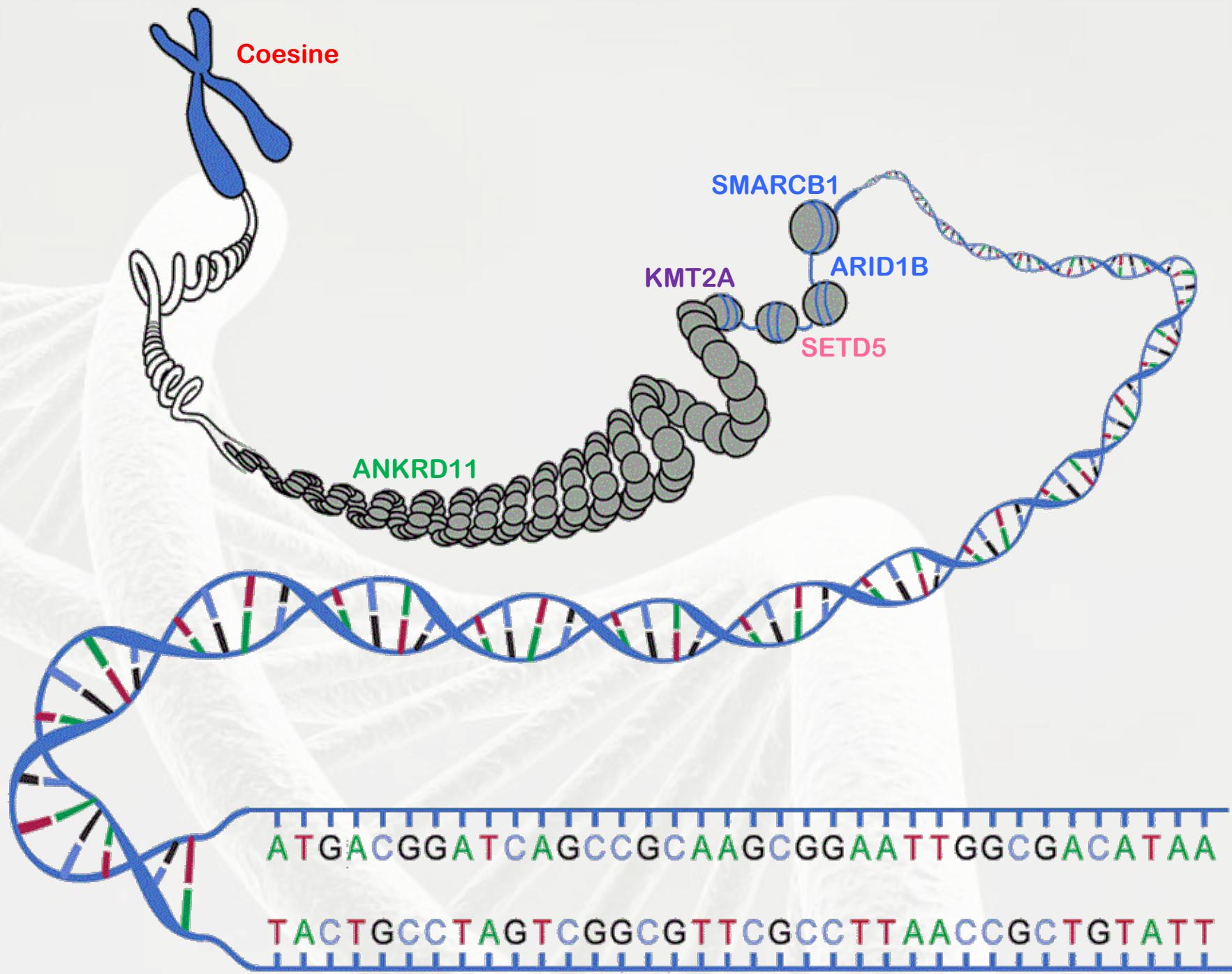
■ *SMC1A*: 8 mutazioni

■ *HDAC8*: 11 mutazioni

■ *SMC3*: 15 mutazioni

■ Ignoto





Disabilitá
intellettiva
SETD5



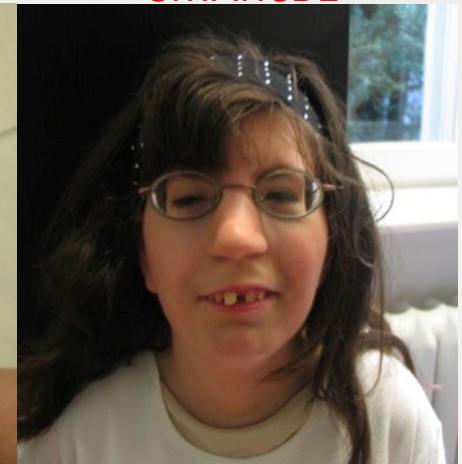
Sindrome di
Wiedemann-
Steiner
KMT2A



Sindrome KBG
ANKRD11



Sindrome di
Coffin-Siris
SMARCB1



ARID1B
Sindrome di
Coffin-Siris



ARID1B
Sindrome di
Coffin-Siris



ARID1B
Sindrome di
Coffin-Siris



NIPBL
CdLS

Grazie!!!



**Ai medici, ai pazienti e alle loro
famiglie!**