

iPS cells and personalized medicine

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Personalized medicine

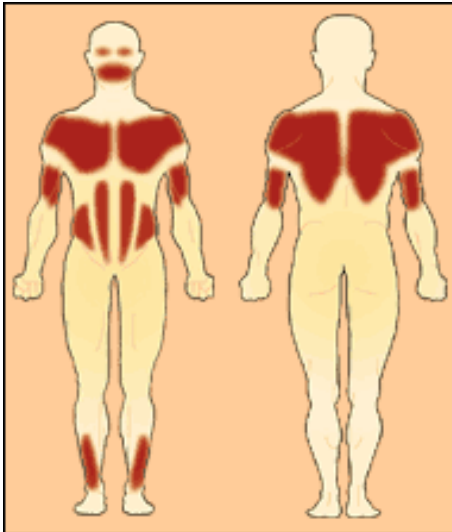
- I want to know the mechanism of the diseases ???
- Probably you will need a suitable model ???
- **I need a specific therapy (ASAP) ???**

- We will need a lot of money to achieve all of that !!!



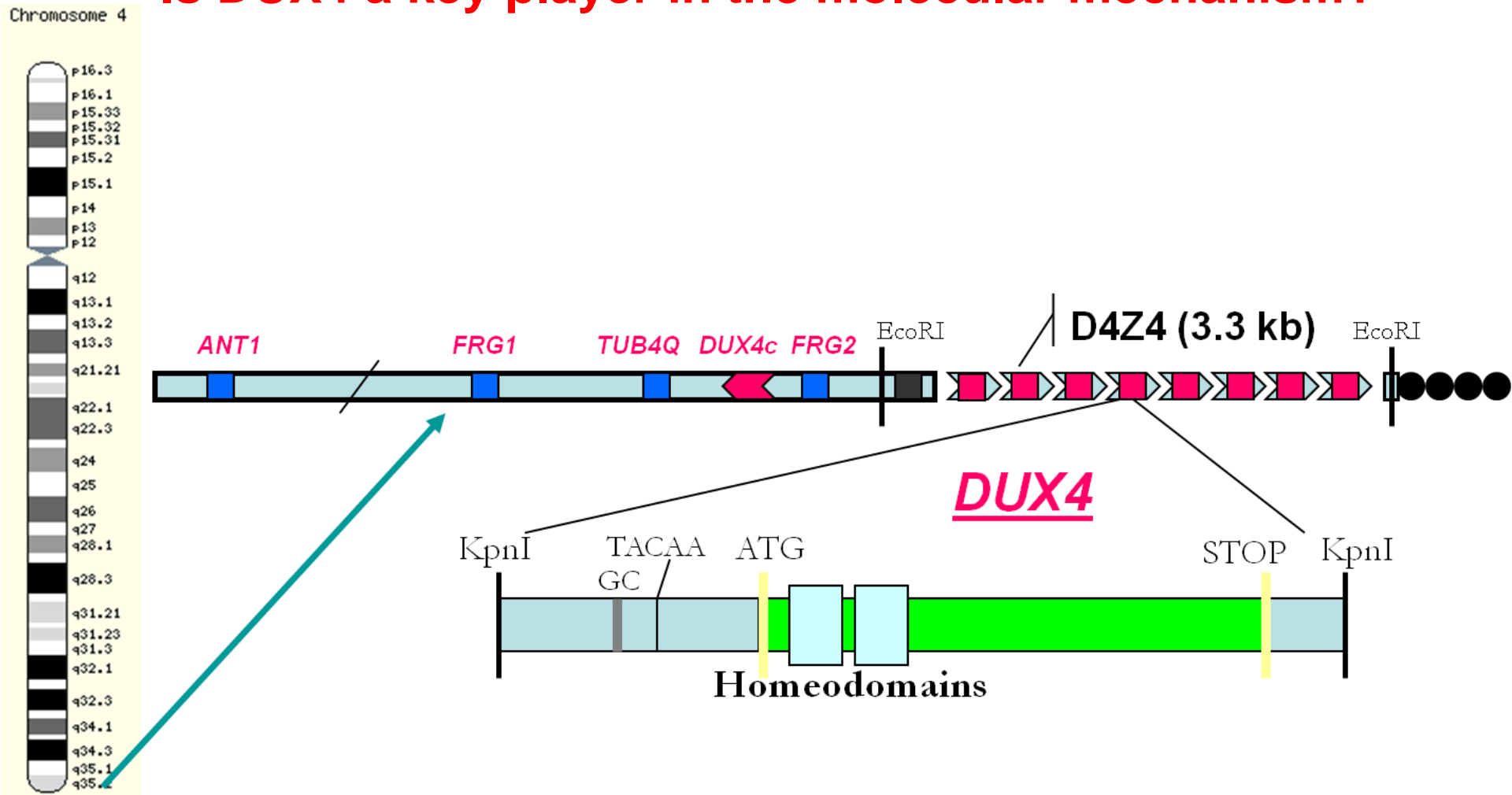
Facioscapulohumeral Muscular Dystrophy (FSHD)

- FSHD is an autosomal dominant inherited disorder
- Third most common inherited neuromuscular condition (1:8000)
- Disease onset is unusual before the age of 10
- Muscles on the face are first affected, then the symptoms are progressive
- **There is no specific treatment.**



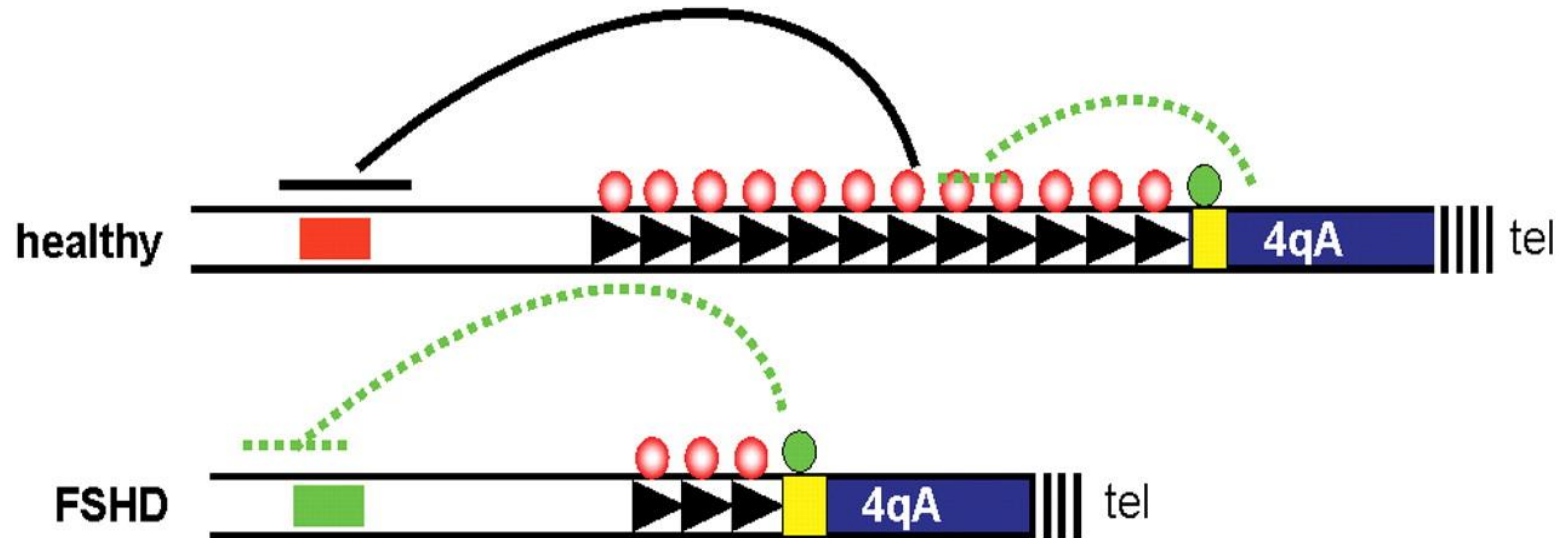
Deletion on chromosome 4 is an initial factor for FSHD

Is DUX4 a key player in the molecular mechanism?



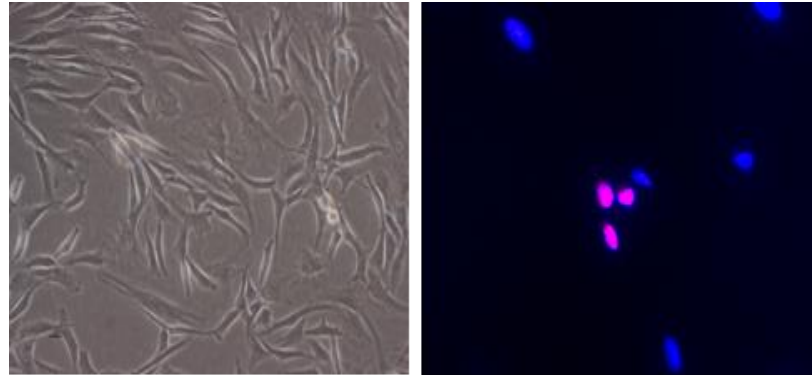
Predictions !!!

4q35A locus is open and transcriptionally active in FSHD



DUX4 and FSHD

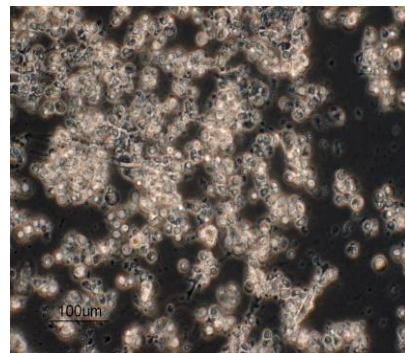
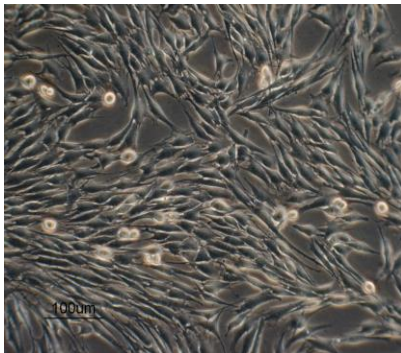
FSHD cells express DUX4



DUX4 induces cell death

Control

+ DUX4



DUX4 affects myogenesis

Control

DUX4

MyHC/DAPI

