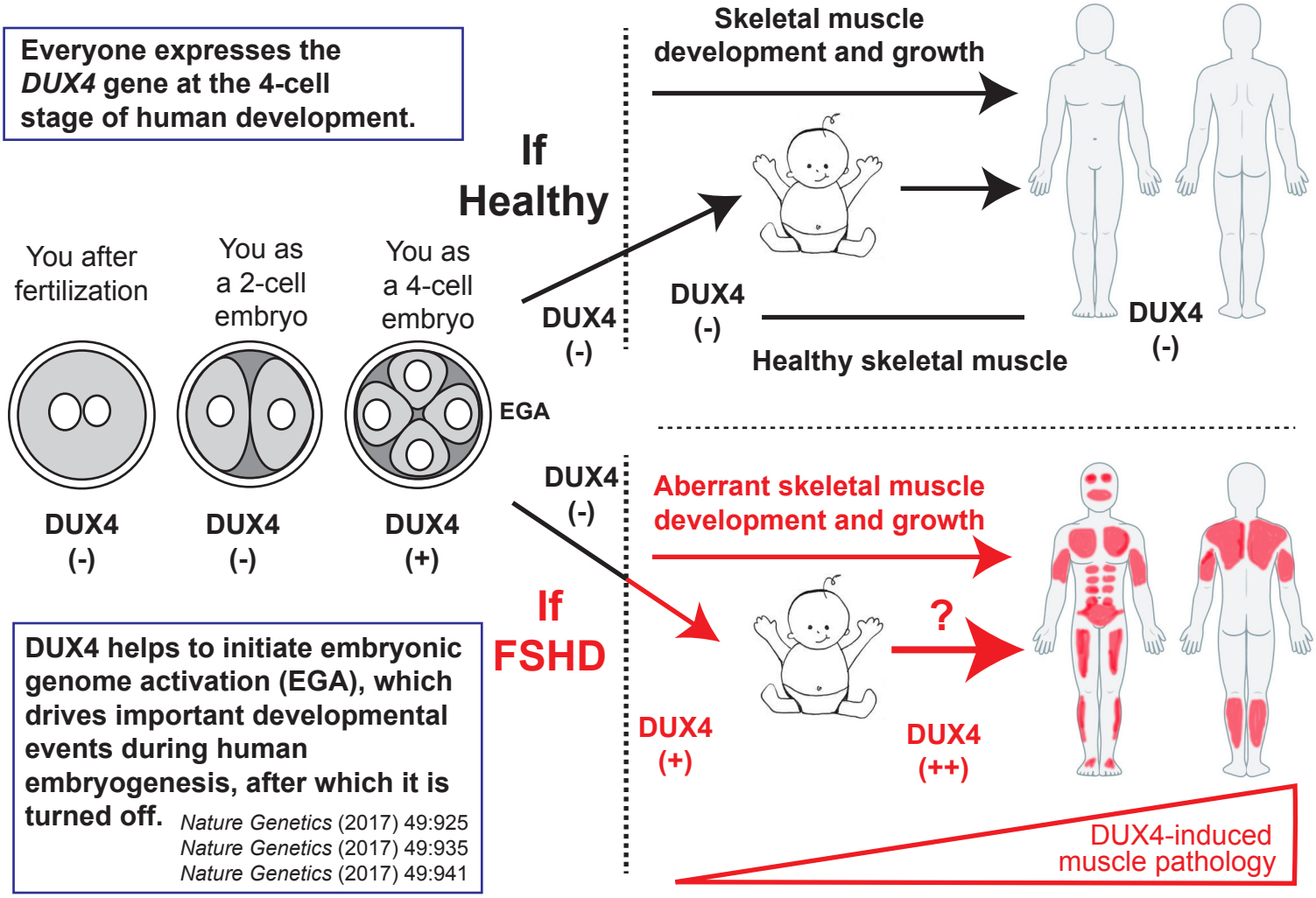
Why do we have the *DUX4* gene?

FSHD is a pathogenic gain-of-function disease caused by the aberrant expression of the *DUX4* gene in skeletal muscles.

However, *DUX4* gene expression is very important in driving normal early human development.

Everyone expresses the *DUX4* gene at the 4-cell stage of human development.



DUX4 helps to initiate embryonic genome activation (EGA), which drives important developmental events during human embryogenesis, after which it is turned off.

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In FSHD, the *DUX4* gene is aberrantly turned back on in skeletal muscle leading to expression of an early embryonic developmental program in the wrong cell type (muscle).

The onset of pathogenic *DUX4* expression, clinical presentation, and accumulated muscle pathology is variable between FSHD individuals.

The *DUX4* gene, its mRNA, and the DUX4 protein are the prime targets for FSHD therapeutic development.