

Identifying Novel Druggable Pathways for FSHD Therapeutics

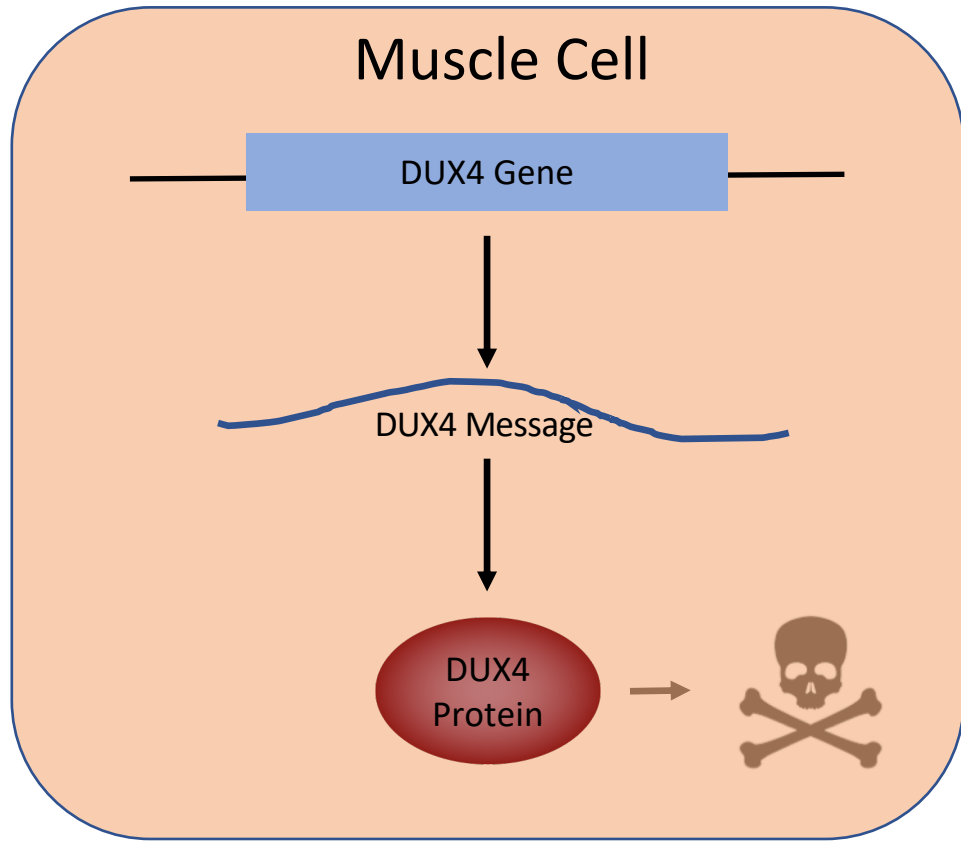
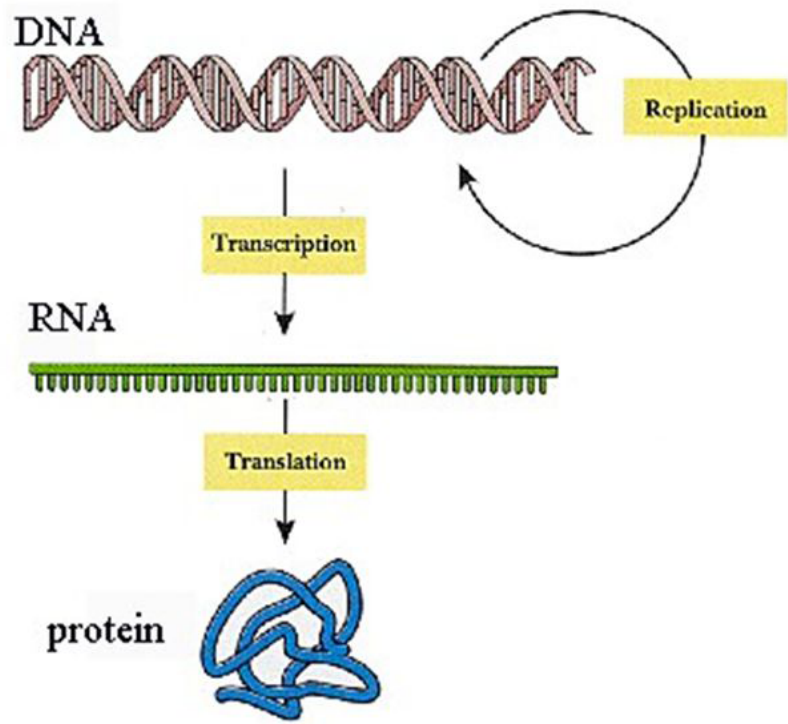
Alec DeSimone, PhD

University of Massachusetts Medical School

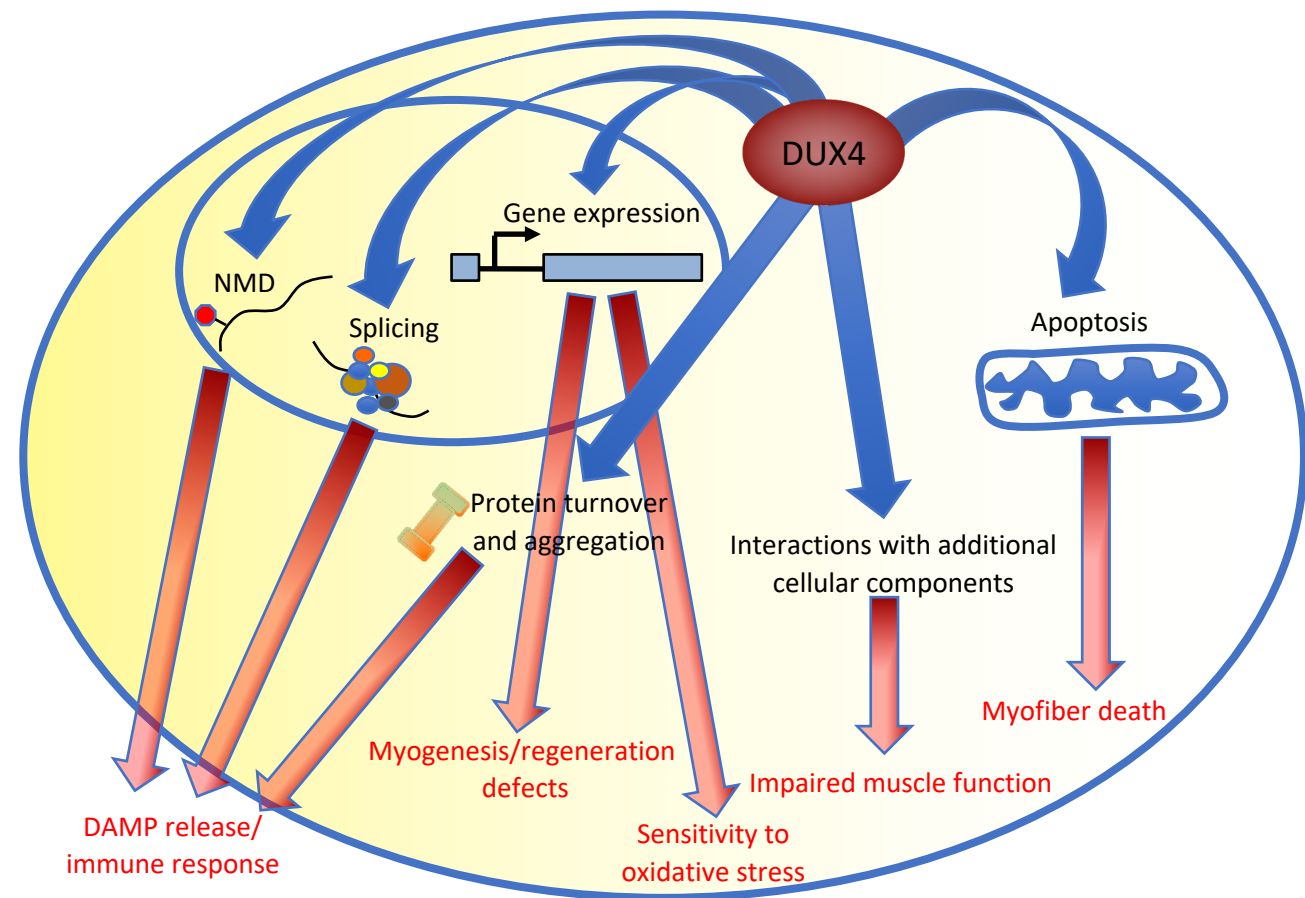
Emerson Lab

11/4/18 FSH Soc. New England Chapter/Wellstone Meeting

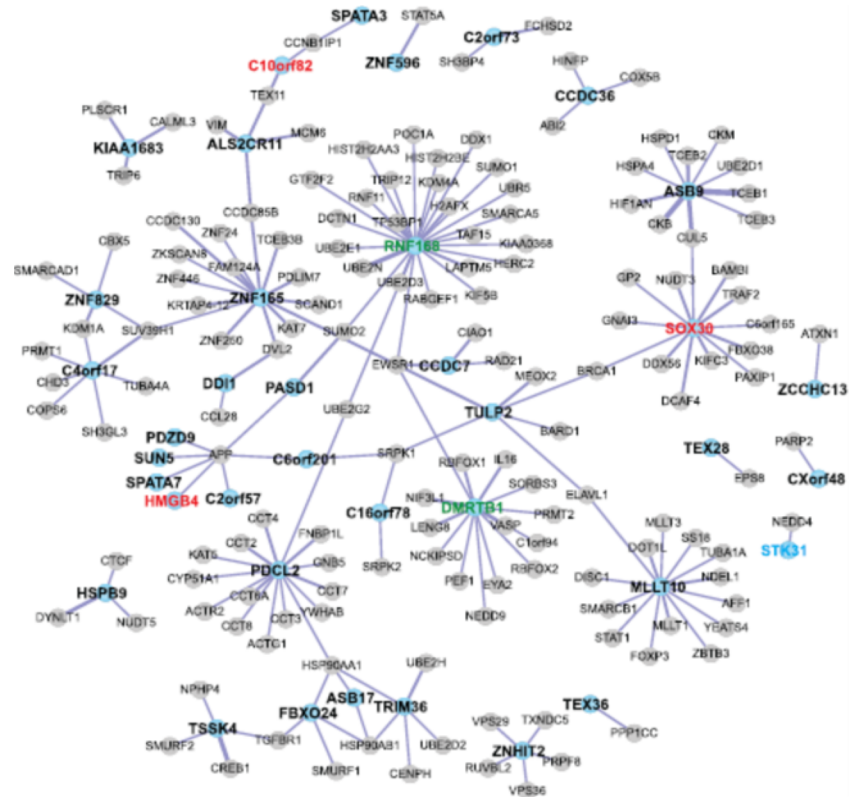
Expression of the DUX4 Gene Muscle Cells Causes Pathology



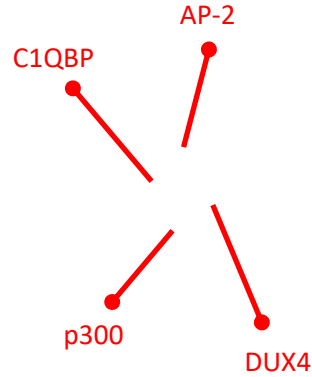
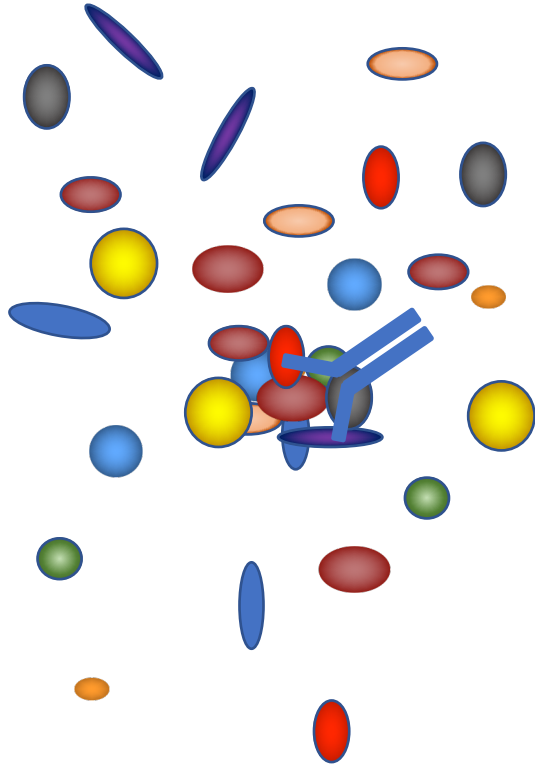
Expression of the DUX4 Gene Muscle Cells Causes Pathology



A cluster of several colorful, egg-shaped objects. One red egg in the center is labeled 'DUX4'. Other colors include yellow, blue, green, orange, and grey. The objects are arranged in a loose group, with some overlapping.



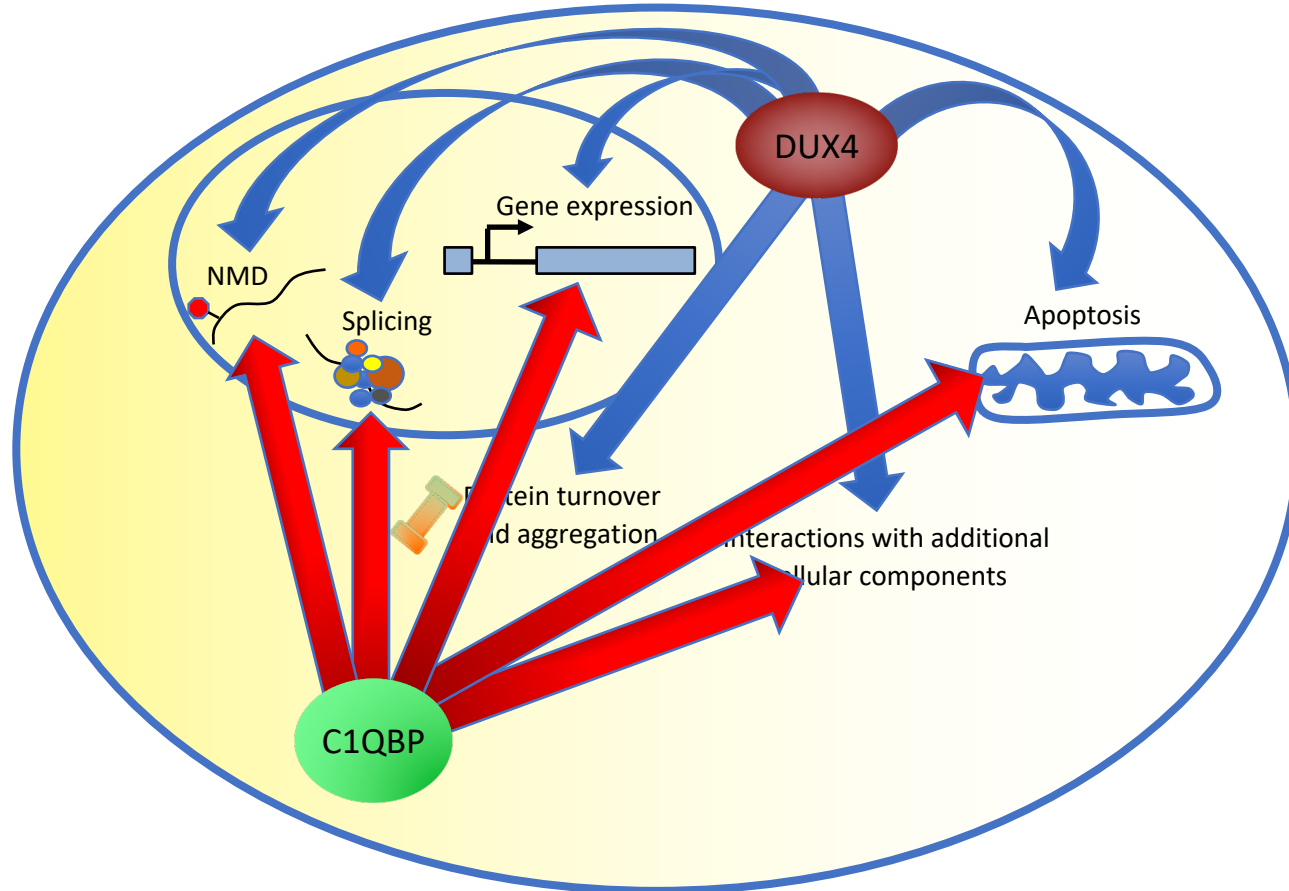
Identifying DUX4 Protein Networks



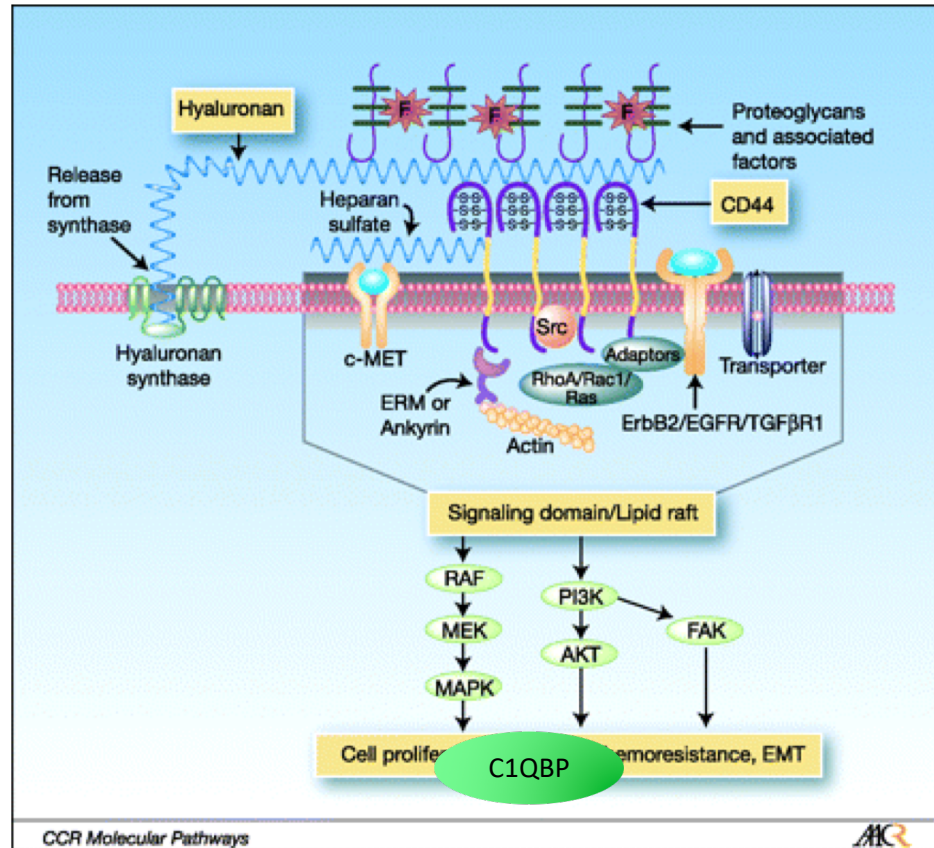
Mass Spectrometer



DUX4 Interacts with a Protein in the Hyaluronan (HA) Network



DUX4 Interacts with a Protein in the Hyaluronan (HA) Network



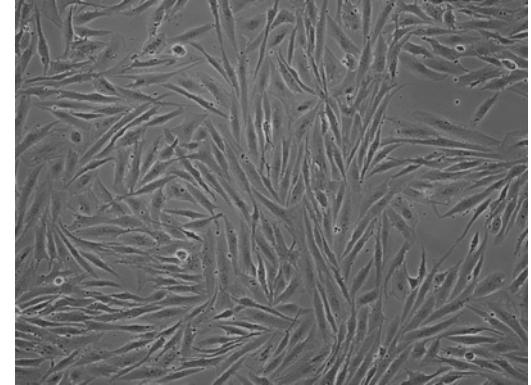
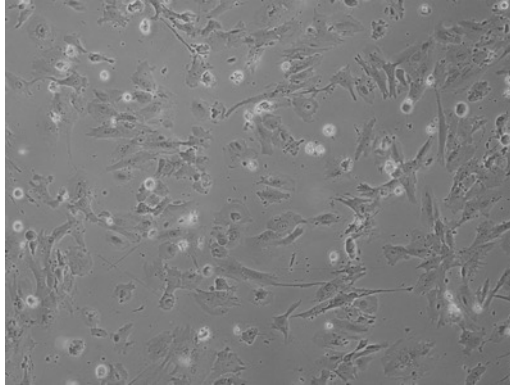
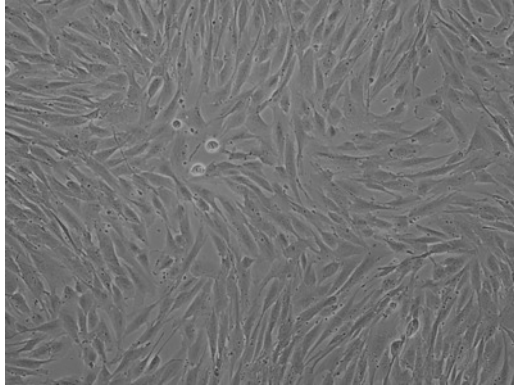
Eliminating HA Signaling Protects Cells from DUX4

Healthy Cells

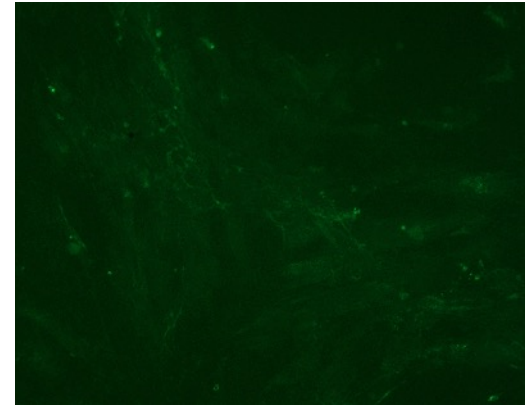
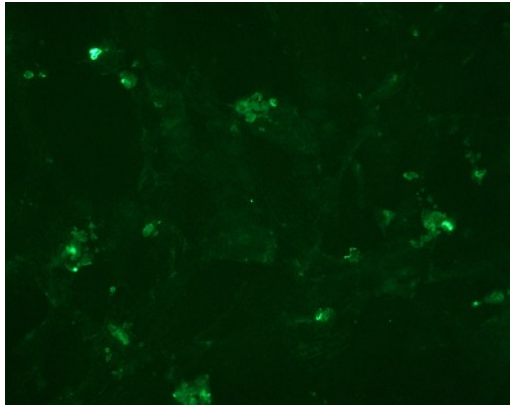
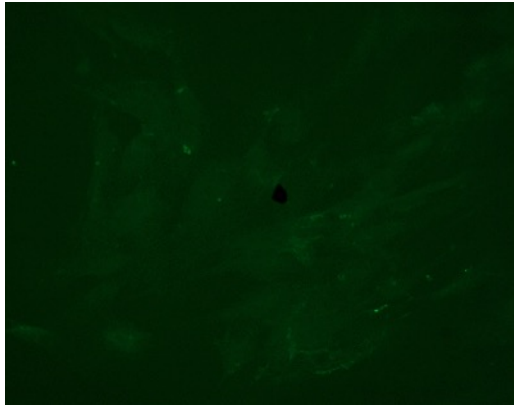
Cells Expressing DUX4

Cells Expressing DUX4
+ HA Inhibitor

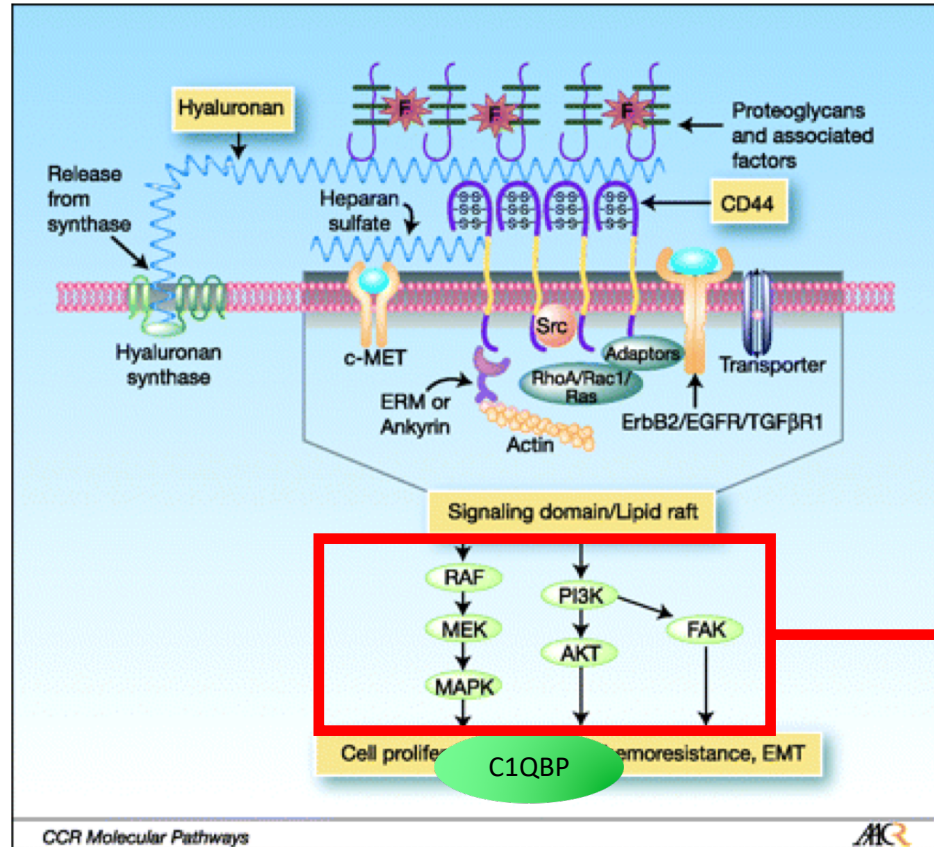
Light
Microscope



HA Stain
(Fluorescence
Microscope)

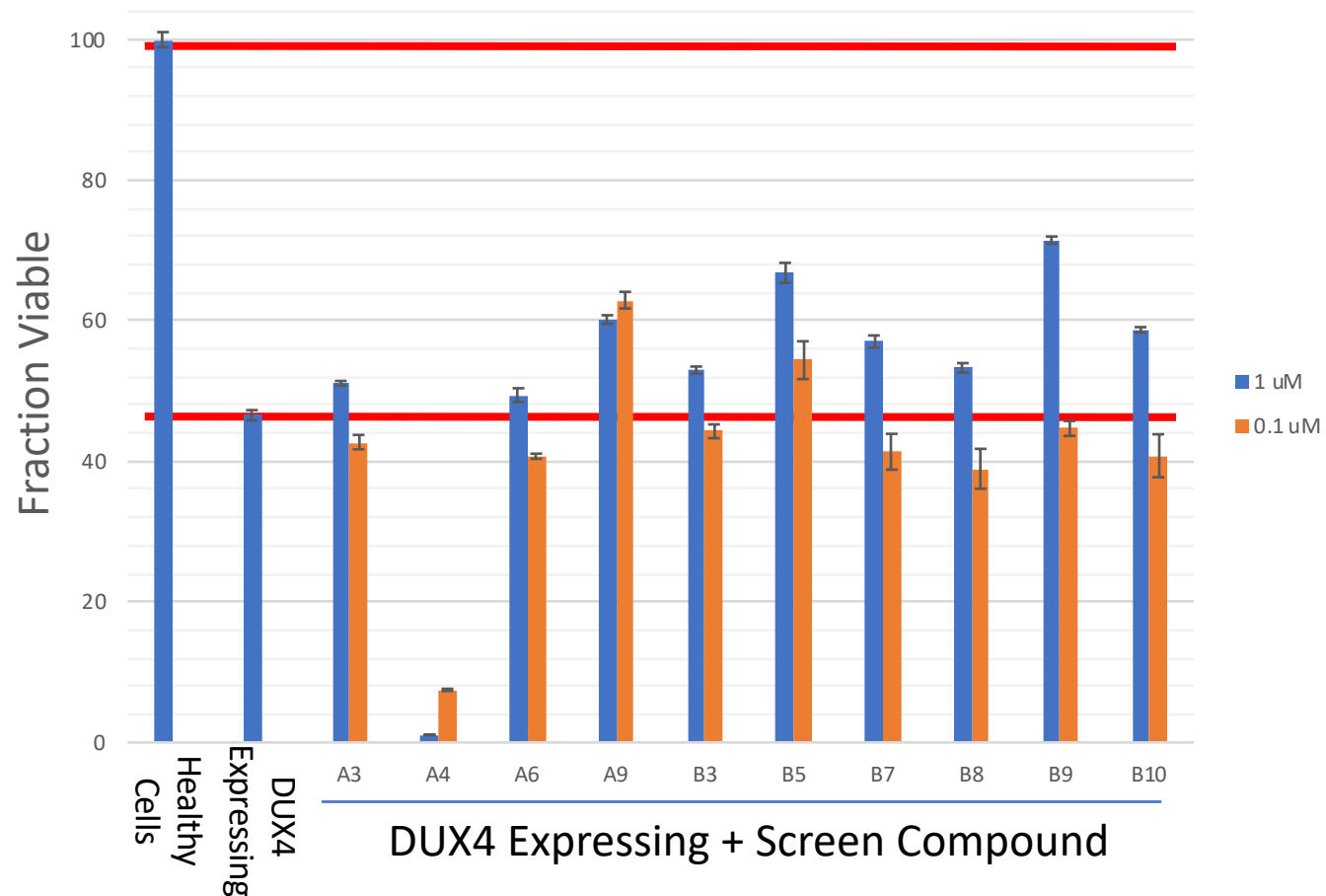


Known Drugs can Affect HA Signaling Networks



Signaling Pathways with well-known inhibitors already in use as therapeutics

Components of the HA Network Can be Targeted with Drugs



Acknowledgements

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