

Problem solving 5.

- Ullrich Congenital Muscular Dystrophy
- Bethlem Myopathy,

UCMD is a relatively severe disorder with onset in young infants and children. Symptoms include neonatal hypotonia, progressive weakness, proximal joint contractures, and laxity of distal joints.

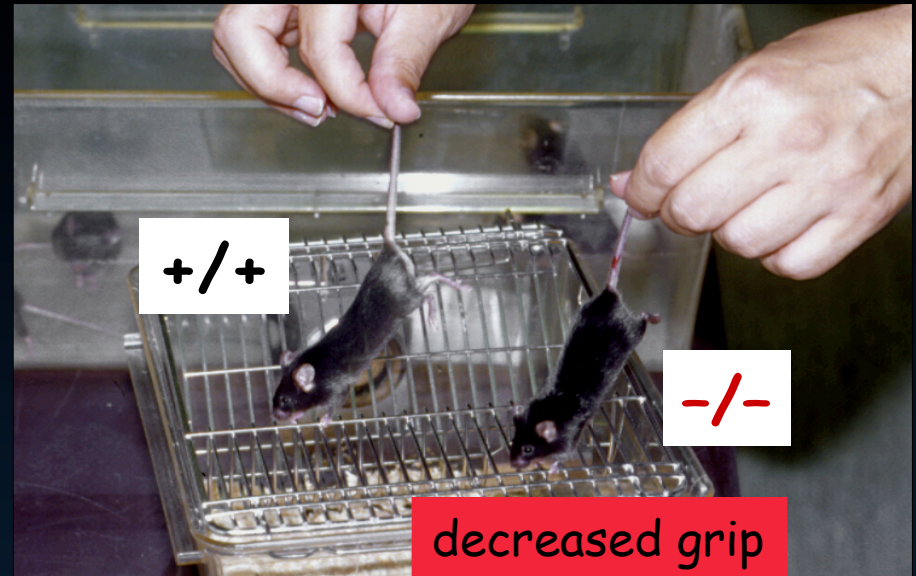
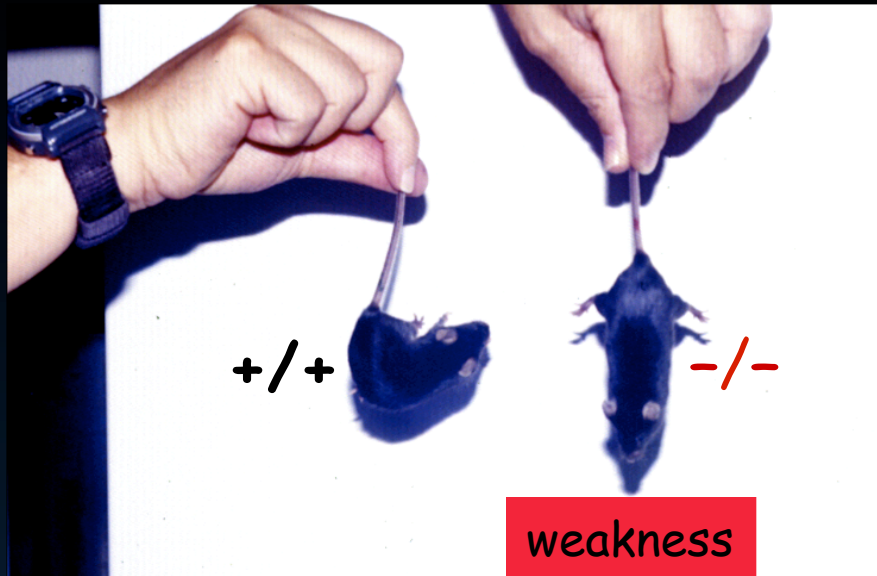
BM is a milder syndrome characterized by slowly progressive proximal weakness and contractures in distal joints with onset in the first or second decade.

a. In a given disease, muscles are affected

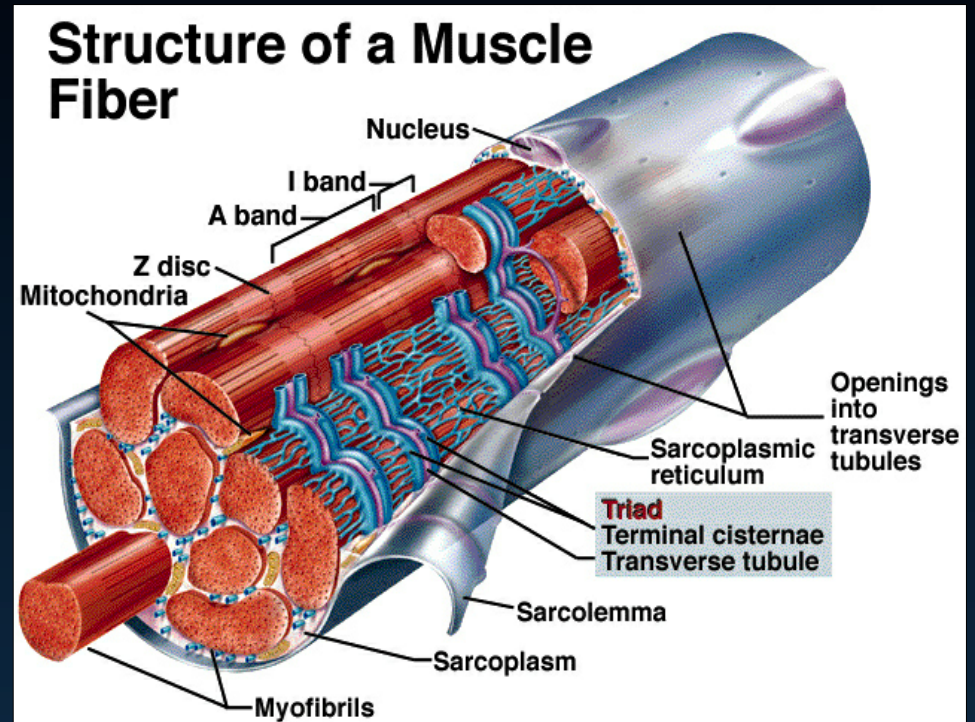
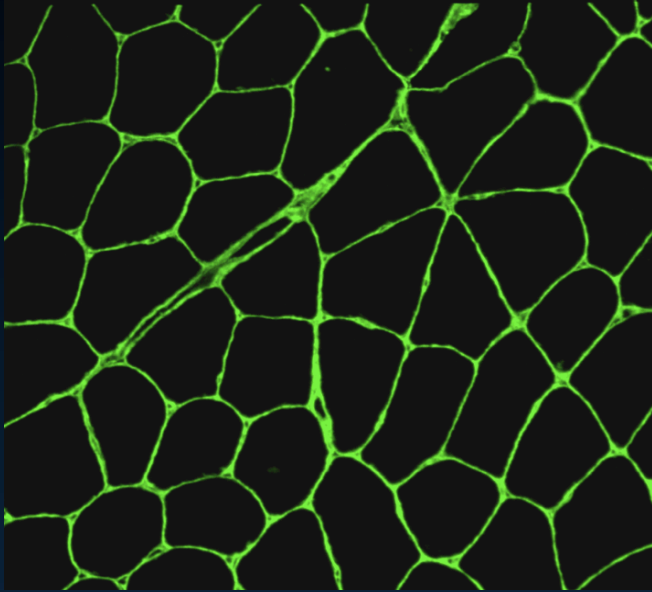
b. The muscle fibers have a defect in autophagy

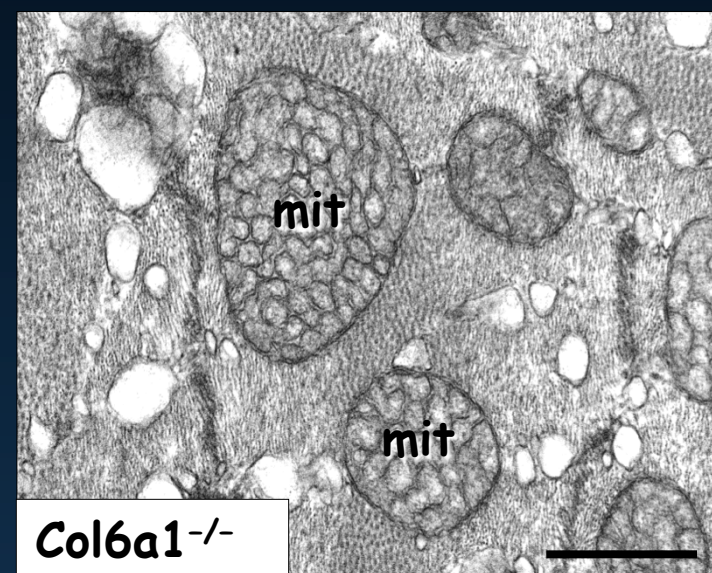
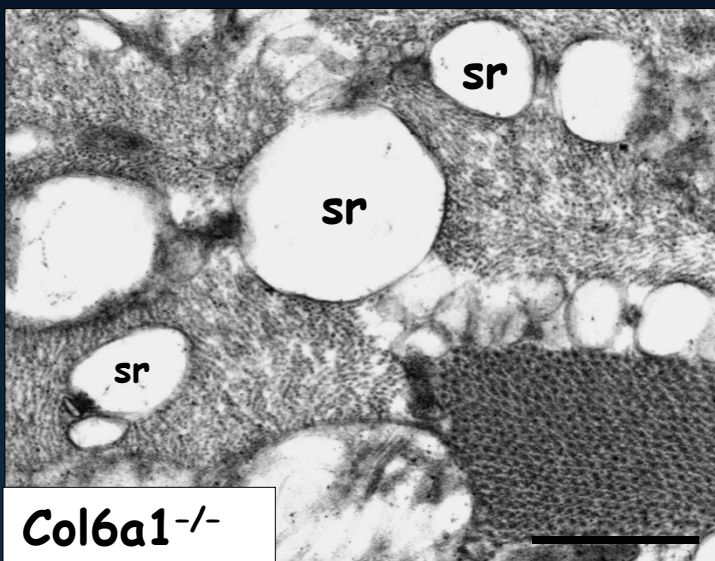
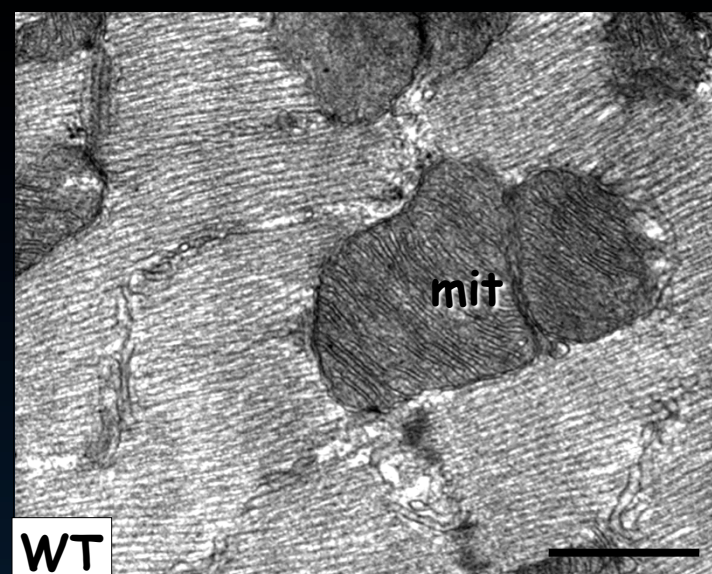
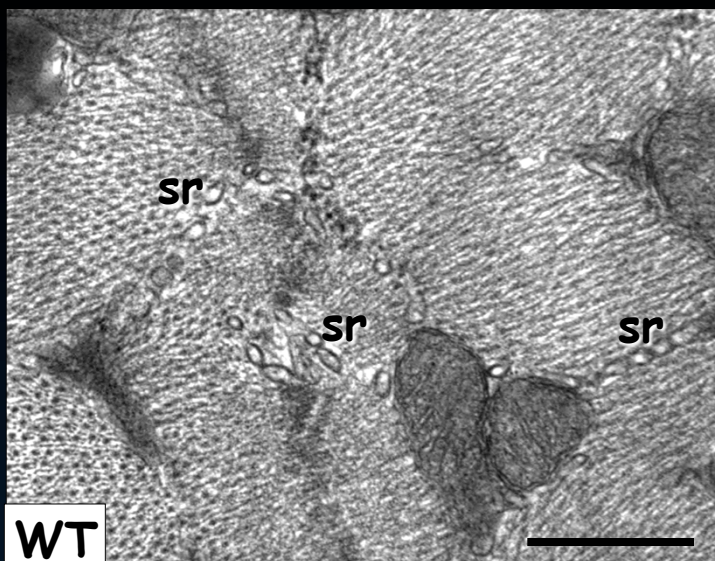
Is it possible to manipulate autophagy to cure this disease?

COLLAGEN VI NULL (*Col6a1*^{-/-}) MICE

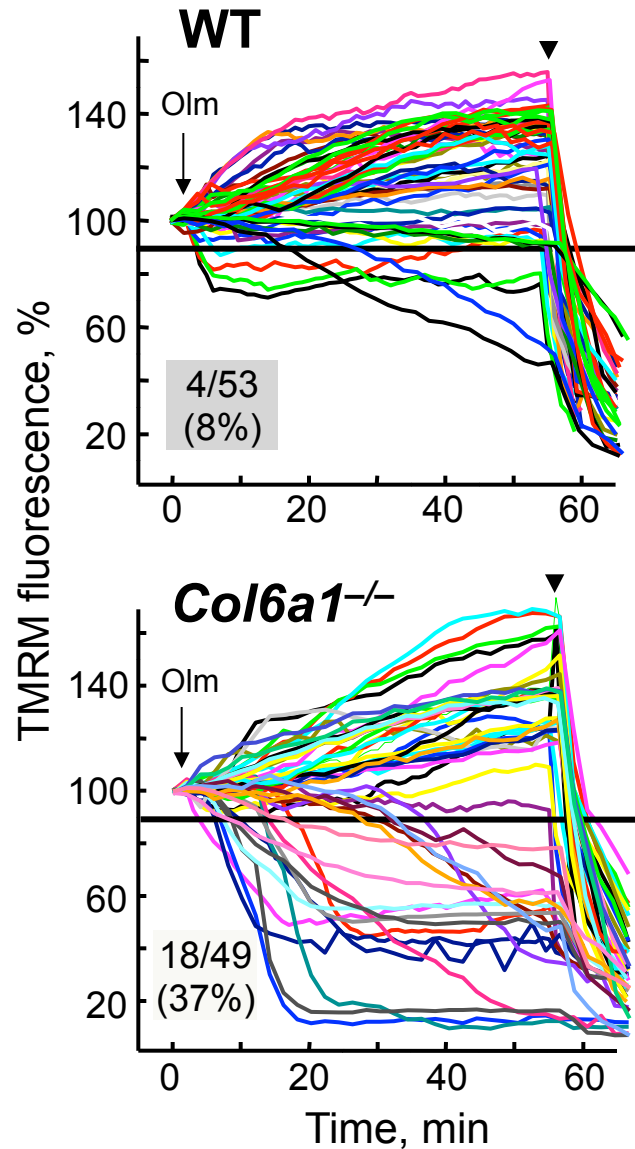


● Ultrastructural analysis of skeletal muscle





● latent mitochondrial dysfunction



● spontaneous apoptosis

