

X-linked myopathy with excessive autophagy (XMEA) is an extremely rare genetic disorder characterized by muscle disease (myopathy). The disorder is fully expressed in males only and is characterized by slowly progressive muscle weakness, especially in the legs. Onset is usually during childhood often between 5-10 years of age. XMEA occurs due to mutations of an unidentified gene on the X chromosome. The disorder is inherited as an X-linked recessive trait. A diagnosis of XMEA is made based upon a thorough clinical evaluation, a detailed patient history and surgical removal and microscopic evaluation (biopsy) of affected muscle tissue.