The Role of Muscle Biopsy Pathology in Diagnosing Genetic Myopathies
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I. Muscle biopsy histopathology as a biomarker of genetic disease
   A. Structural and ultrastructural changes
      1. congenital myopathies
      2. congenital myasthenic syndromes
      3. autophagic vacuolar myopathies
      4. myofibrillar myopathies
      5. other myopathies
   B. Immunostaining abnormalities
      1. muscular dystrophies
      2. autophagic vacuolar myopathies
   C. Western blotting in muscular dystrophies

II. Approaches to diagnosis
   A. Traditional approach
      1. clinical evaluation
      2. muscle biopsy
      3. genetic testing
   B. Emerging approach to diagnosis
      1. clinical evaluation
      2. genetic testing
      3. muscle biopsy for confirmation (verification)
      4. muscle biopsy to obtain diagnostic clues
   C. Genetic testing results that may lead to muscle biopsy
      1. no mutation(s) found
      2. single mutant allele in a recessive disorder
      3. variants of unknown significance
      4. sequence variant(s) in a novel gene
   D. Circumstances that drive the approach to diagnosis
      1. relative costs of testing modalities
      2. resources to pay for testing
      3. clinical acumen
      4. diagnostic acumen
      5. availability of technology (access to technology)

III. Clinical trial endpoints