Case 2004-9

Submitted by: Dr. Christopher Pierson, Children’s Hospital and Brigham and Women’s Hospital, and Dr. E. Tessa Hedley-Whyte, Massachusetts General Hospital, Boston, MA

Diagnosis: Merosin deficient congenital muscular dystrophy

Comment: There were increased variation of muscle fiber size and phagocytosis of fibers, on microscopical examination. Neither “ragged red” fibers nor nemaline rods were present. Merosin was absent from the sarcolemmal membranes, on immunocytochemistry. Merosin deficiency in a mouse model has been successfully treated with agrin, which makes a functional mimetic of merosin.

References:
