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REFERENCES AND DIAGNOSES

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Case 2004-9

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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:

Moll J, Barzaghi P, Lin S, et al: An agrin minigene rescues dystrophic symptoms in a mouse model for congenital muscular dystrophy. Nature 2001; 413:302-307.

Pegoraro E, Mancias P, Swerdlow SH, et al: Congenital muscular dystrophy with primary laminin alpha2 (merosin) deficiency presenting as inflammatory myopathy. Ann Neurol 1996; 40:782-791.

Tubridy N, Fontaine B, Eymard B: Congenital myopathies and congenital muscular dystrophies. [Review] Current Opinion in Neurology 2001; 14:575-582.