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Growing challenges with phenotype-based practice, muscle biopsy in neuromuscular diseases? A few novel presentations

Sankar Bandyopadhyay
Penn State College of Medicine, USA

Introduction: Phenotype-based disease-classification and fixed histology, are not infallible.

Objective: Appreciation of novel phenotypes and histological findings.

Methods: Case studies. Histology.

Results: Case 1: 45 year-old woman with 10 year of proximal lower extremity, 2 year of bilateral finger flexion weakness with supportive objective evidence. CK 734. EMG and muscle biopsies: myopathic.

Presumptive Diagnosis: Early onset Inclusion Body Myositis.

Final diagnosis: Acid maltase deficiency (Adult onset Pompe disease), after Dried blood test and GAA sequencing, with novel IBM phenotype (MUSCLE & NERVE by same author)

Case 2: 53 year man with 2 years of progressive bilateral arms, forearms and thigh weakness. Objective weakness was corresponding. Prominent contractures of bilateral hamstrings and biceps. EMG and muscle biopsy: myopathic. Ck 162.

Phenotypic diagnosis: atypical late onset Emery-Dreifuss or Bethlem Muscular dystrophy.

Final diagnosis: 2B LGMD or Miyoshi myopathy.

Novel features: contractures, normal CK, hamstring type).

Case 3: 78 year man with 18 months of proximal lower extremity and distal upper extremity weakness. CK 379. EMG myopathic. Muscle biopsy: myopathic, rimmed vacuoles PLUS C5b-9 (seen in membrane attack complex disease e.g. dermatomyositis).

Final diagnosis: Sporadic IBM with Dermatomyositis histology profile.

Novel feature: C5b-9 positive staining in IBM (MUSCLE & NERVE by author). No response to immunosuppressives.

Case 4: 77 year old with 2 years of proximal LE and Grip weakness. CK 415. EMG and muscle biopsy: myopathic with rimmed vacuoles. TDP-43 positive (Sporadic IBM). Strongly positive C5b-9.

Final diagnosis: Sporadic IBM with Dermatomyositis histology profile.

Novel features: C5b-9 in IBM. TDP43 and C5b-9 co-existence.

Conclusion: Phenotype and histology based evidences are proving inadequate in neuromuscular diagnosis with growing evidences. Genotype is still a developing concept with caveats of unknown significance of variants.

Recent Publications

1. Bandyopadhyay S, Buchman SL. Prevalence of Vitamin D Deficiency in spring and Summer Months in Central Pennsylvania Neuromuscular Clinic: A Study Complementary to the Pilot Study Published in Journal of Clinical Neuromuscular Disease in June 2015 for Fall and Winter Months. J Clin Neuromuscul Dis. 2016 Jun; 17(4):223-5. PMID: 27224439.
2. Buchman S, Bandyopadhyay S, Baccon J, Wicklund M. A surprising case of inclusion body myositis with positive endomysial C5b-9 staining. Muscle Nerve. 2016 06; 53(6):991-2. PMID: 26659413.

3. Bandyopadhyay S, Reid D. Electrical myotonia in acid maltase deficiency disease. Muscle Nerve. 2015 Dec; 52(6):1141. PMID: 26235386.
4. Bandyopadhyay S, Wicklund M, Specht CS. Novel presentation of Pompe disease: Inclusion-body myositis-like clinical phenotype. Muscle Nerve. 2015 Sep; 52(3):466-7. PMID: 25846667.

Biography

Sankar Bandyopadhyay is an Associate Professor of Neurology in College of Medicine, Penn State University. He practices at Penn State Neuroscience Institute. He got his M.B.B.S degree from Calcutta National Medical College. He got a fellowship in Clinical Neurophysiology from Medical College of Georgia in 2001. His are of researches are bone health in neuromuscular diseases, vitamin-D deficiency, clinical neuromuscular disorders and others.

sbandyopadhyay1@pennstatehealth.psu.edu

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