

Congenital myopathies: adventures in new gene discovery

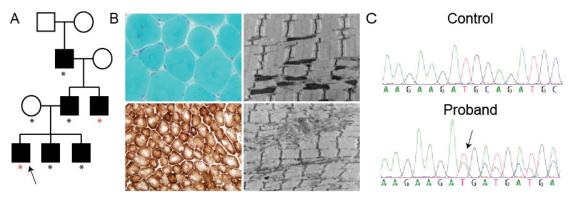


Figure: Novel TPM2 mutation in core-rod myopathy

Congenital myopathies are a heterogeneous group of disorders characterized by muscle weakness, lifelong disabilities and early mortality. To date, no therapies are available for these devastating diseases. One of the major barriers to therapy development is that nearly 50% of congenital myopathies are currently genetically unresolved. One of the overarching goals of my laboratory is new gene discovery for these disorders. We have developed a novel strategy for gene identification that includes application of next generation sequencing technology combined with utilization of the zebrafish model system. Using this approach, we have recently identified and characterized three novel genetic causes of congenital myopathy.

Dr. James Dowling

University of Michigan (Candidate for Faculty Appointment)

Host: Dr. Barbara Funnell

Date: Friday August 23, 2013

Time: 2:00 p.m.

Place: Donnelly CCBR Building

160 College St. Red Seminar Room