Role of MyHC-embryonic protein in muscle development and disease

Dr. Sam J. Mathew, Assistant Professor at DBT's Regional Centre for Biotechnology (RCB), Faridabad, found out that loss of MyHC-embryonic protein leads to muscle abnormalities including alterations in muscle fiber type, fiber number and fiber size. Secondly, the MyHC-embryonic protein is required to regulate the rate of differentiation of the muscle stem cells during development, and this effect is mediated by fibroblast growth factor (FGF) signalling. Although MyHC-embryonic is expressed in all muscles, they find that loss of MyHC-embryonic has differential effects on distinct muscles, and the adult mice lacking MyHC-embryonic displayed abnormal curved spine (scoliosis), an abnormality seen in individuals with Freeman-Sheldon Syndrome (FSS).

Targeted mouse models, they have characterized the function of MyHC-embryonic and developmental myosins in general, during embryonic stages of development. Loss of function of MyHC-embryonic leads to scoliosis in adult mice, a defect seen in FSS patients, and this mouse model could thus be a valuable tool in understanding the defects underlying this congenital disorder.

Myosins are proteins which are present in all cell types, meant for fundamental cellular functions such as cell movement, cell division and transport of cargoes within cells. A specialized set of myosin proteins known as muscle myosins are expressed by the skeletal muscle, which are required for muscle contraction. While most muscle myosin proteins are expressed in the adult muscle, two are expressed only during embryonic development.

Very little is known about these developmentally expressed muscle myosins except that mutations in the gene encoding one of the myosins, *MYH3*, leads to Freeman-Sheldon Syndrome (FSS) in humans, a genetic disease causing severe musculoskeletal abnormalities including joint deformities, bent fingers, club feet, curved spine and facial anomalies. FSS patients have compromised movement, respiratory, speech and feeding problems and delayed growth and development.

Reference: https://dev.biologists.org/content/147/7/dev184507