NOVA SCOTIA Health Research FOUNDATION

**PROJECT FACT SHEET**

# Muscles Matter—Even in the Womb

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Dr. Boris Kablar, with help from NSHRF, is helping us understand the roles that muscles play in fetal development. His research may eventually help us understand diseases associated with muscular atrophy.

Muscular atrophy, wasting, and loss rob thousands of Canadians of their mobility every year. Diseases related to muscular atrophy are genetic, and SMA (Spinal Muscular Atrophy) Support Inc. estimates that one in 40 people carry a recessive gene for muscular atrophy.

Central to our evolving understanding of these diseases are the roles that muscles play in the development of embryos. Even in the womb, fetuses use muscles, and this affects the development of their skeletal system and other organs. In fetuses affected by genetic diseases relating to muscular atrophy, the development of the skeleton and organs is likely affected by abnormalities in the developing muscles. However, researchers do not fully understand the roles played by developing muscles.

Boris Kablar, Associate Professor, Department of Anatomy & Neurobiology, Faculty of Medicine, Dalhousie University, in collaboration with Brian Hall of Dalhousie's Biology Department, has contributed to our understanding of the interplay of genes, bones, and muscles in a new study Supported by the Nova Scotia Health Research Foundation.

"We used mouse embryos genetically engineered without skeletal muscle to explore the development of the skeletal system in the absence of muscle movement. We focused on three areas: the role of skeletal muscle in the development of bones and joints; the role of muscle in the maintenance and fusion of cartilage; and the development of the inner ear," says Dr. Kablar.

The study concluded that fetal development is strongly influenced by mechanical cues from the skeletal musculature. Research indicated that without skeletal musculature, multiple bone defects occur including enlarged and fused cervical vertebrae, posture abnormalities, shortening and fusion of long bones, and underdevelopment of the shoulder blade and collar bone.

The researchers found that an absence of skeletal muscle also leads to cleft palate and cleft sternum. In contrast, the development of the skull remained essentially normal.

Dr. Kablar also discovered that muscular cues are important for the development of the inner ear. In the absence of skeletal muscle, sensory cells of the inner ear related to angular acceleration fail to fully differentiate.

“We would like next to explore the role of skeletal muscle in the development of cleft plate, cleft sternum, and the molecular basis of secondary cartilage development,” says Dr. Kablar.

This research is a novel approach to the study of the dependence of skeletal and neural development on the muscles of the developing fetus, and Dr. Kablar’s findings help us understand how the muscles of the fetus affect its development.

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