Zebrafish help identify mutant gene in rare muscle disease

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Zebrafish with very weak muscles helped scientists decode the elusive genetic mutation responsible for Native American myopathy, a rare, hereditary muscle disease that afflicts Native Americans in North Carolina.

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Provided by University of Michigan

Scientists led by John Kuwada, professor of molecular, cellular and developmental biology at the University of Michigan, and Hiromi Hirata of the National Institute of Genetics in Japan originally identified the gene in mutant zebrafish that exhibited severe muscle weakness. Native American myopathy causes muscle weakness from birth and other severe problems that can lead to death before adulthood.

The findings appear in the journal *Nature Communications*.

The responsible gene encodes for a muscle protein called Stac3, which in turn regulates a physiological process required for muscle contraction. The muscles of zebrafish and people with the genetic mutation don't make normal Stac3 protein and the muscles don't contract effectively.

Scientists established the importance of Stac3 for muscle function in zebrafish by studying the small fish physiologically and genetically. Scientists then looked at the human version of the gene, and found that the gene was mutated in people suffering from Native American myopathy.

For many degenerative muscle diseases few drugs help, largely because scientists don't know the genes responsible for many of these muscle diseases, making it difficult to develop drugs and other therapies that target the condition. The discovery of the gene for Native American myopathy, however, may help develop drugs to treat the myopathy, as well as other related muscle diseases.

More information: