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[Unraveling the pathophysiology of Bethlem Myopathy using a unique zebrafish model for the disease].

[Article in French; Abstract available in French from the publisher]

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Abstract in [English](#), [French](#)

Bethlem myopathy (BM) is a neuromuscular disease characterized by joint contractures and muscle weakness. BM is caused by mutations in one of the genes encoding one of the three α -chains of collagen VI (COLVI), a component of the skeletal muscle extracellular matrix. Nowadays, an unresolved question is to understand how alteration of COLVI located outside the muscle cells leads to functional modifications in muscle fibers. The zebrafish model $col6a1^{\Delta ex14}$ is currently the unique animal model of the disease since it is the only model to reproduce a mutation that is the most frequently found in BM patients. In patient and $col6a1^{\Delta ex14}$ zebrafish muscles, the structure of the sarcoplasmic reticulum has been found to be altered, thus suggesting dysfunction in intracellular Ca^{2+} handling and/or in ion channels that are known to control Ca^{2+} homeostasis and to play pivotal roles in muscle function and pathogenesis. Therefore, our project aims at exploring the properties of ion channels and intracellular Ca^{2+} regulation using electrophysiological approaches and intracellular Ca^{2+} measurement at rest and during activity in isolated muscle fibers from $col6a1^{\Delta ex14}$ zebrafish. On one hand, this project should contribute to decipher how alteration in an extracellular matrix component transduces pathogenic signals within muscle fiber and should possibly lead to identify therapeutic targets for this currently incurable disease. On the other hand, because functional studies on zebrafish muscle cells are scarce, this project will provide a sound database on the electrophysiological properties of this cell model.

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