# High throughput gene sequencing to identify new genes that cause myotubular and Centronuclear Myopathy

The project, which will be run by Dr Jocelyn Laporte and colleagues in the department of translational medicine at IGBMC in France, will use next generation sequencing to identify novel genes implicated in centronuclear myopathies. The life-threatening congenital myopathies are present in all populations, affecting children as well as adults. Considerable progress in human genetics within the past 25 years led to the identification of the molecular basis for 50% of these pathologies. However, the causative mutations in half of patients are still unknown. This is mainly due to genetic heterogeneity (mutation in several genes causing the same or very similar disease) and to the lack of large families and large panels of patients. To date, molecular approaches used for identifying implicated genes correspond to gene by gene explorations, starting from the most pertinent one. Considering that the human genome contains 3 billion nucleotides - enough letters to fill 6000 novels - and that a change of a single nucleotide can trigger the myopathy, the quest for the "wrong letter" is often time-consuming and laborious. The next generation sequencing technology allows the massive parallel analysis of all genes of the human genome at once and is a promising approach for the efficient identification of the unknown genes. This is the important first step to provide an accurate genetic counselling, to improve health care and disease management and to identify novel drug targets that may be more accessible for therapeutic development.

In 1996 Jocelyn Laporte led the discovery of the x-linked gene, MTM1, which is the cause of the majority of cases of myotubular/centronuclear myopathy.

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