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Disturbance of the human gut microbiota in patients with Myotonic Dystrophy type 1.

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Abstract:

Myotonic dystrophy (MD) is an autosomal dominant genetic disorder found highly prevalent in the province of Québec, Canada. MD type 1 is characterized by an abnormal trinucleotide CTG repeat located in the 3' untranslated region of DMPK, the gene encoding the DM protein kinase located on chromosome 19q13.3. The core pathogenic feature of MD1 is the intra-nuclear blockage of RNA-binding proteins with the toxic RNA repeat, resulting in a wide array of nonfunctional proteins. Although MD1 is primarily characterized by progressive muscular weakness, there are many multisystemic symptoms: cognitive deficits, cardiac conduction abnormalities, diabetes, and cataracts, as well as endocrine and reproductive problems. Involvement of the gastrointestinal (GI) tract is also frequent and can affect the whole digestive tract from the pharynx to the anal sphincter. However, it is not clear if these GI symptoms are caused by biomechanical problems of the intestine or if the intestinal microbiota is involved. The objective of this study is to assess the role of the gut microbiota in the GI symptoms of MD1 patients. Stool samples from 50 MD1 patients were collected, and their close family members used as controls. These samples were sequenced by 16s MiSeq and analyzed with DADA2 to generate taxonomic signatures. Our analysis indicated that the status of MD1 significantly changes gut bacterial community structure as for the relative abundance of Firmicutes, Bacteroidota, and Actinobacteriota Phylum. These results might help understand how the gut microbiota, in addition to the biomechanics, can affect the gastrointestinal tract of MD1 patients.

BIOGRAPHY:

Manijeh has completed his first PhD in pharmaceutical biotechnology from Isfahan university of medical sciences and started her second PhD in Biochemistry at Universite de Sherbrooke. She works on the gut microbiome and its role in the gastrointestinal manifestations of Myotonic Dystrophy type 1 patients in both mice and human samples. Her H-index is 8 and has published about 24 papers in reputed journals.

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