

Characterization of fecal and oral microbiome associated with Lynch syndrome

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Introduction. Lynch syndrome (LS) is a type of inherited cancer syndrome associated with a genetic predisposition to different cancer types that affect different body districts, such as for example colorectal cancer (CRC). LS is an autosomal dominant condition associated with germline alterations of genes related to the DNA mismatch repair process. In recent years, the development of microbiome analysis has led to a new era of microbial discovery, thus unveiling important associations between microbiota and several pathological conditions including CRC. At the moment, there are still no relevant studies that have investigated the possible involvement of the microbiota in affecting LS onset therefore we decided to perform microbiome analysis of fecal and salivary samples of patients affected by LS and their relative age and sex matched controls.

Materials and methods. Total DNA from fecal and salivary samples of patients with LS and matched controls was extracted using specific DNA extraction kit. From the total DNA, we specifically amplified the V3-V4 region of the bacterial 16s rRNA gene. The obtained amplicons were purified, indexed and, after a second purification step, quantified and pooled, then loaded on the MiSeq Illumina system. Sequences with high quality score and length >250bp were used for the taxonomic analysis with QIIME software (v1.9.1).

Results. In fecal samples, we observed differences in the relative abundance of Bacteroidaceae and Prevotellaceae, increased in the LS group, and the family of Ruminococcaceae, decreased in the LS group. We did not find any statistical significant difference in alpha and beta diversity. In salivary samples, we observed differences in the relative abundance of Veillonellaceae, Prevotellaceae and Streptococcaceae, increased in the LS group, and Neisseriaceae and Pasteurellaceae, decreased in the LS group. We also found a statistically significant difference in the alpha diversity observed otus and Shannon index, lower in LS patients, and the beta diversity index allows us to barely distinguish the two populations of LS patients and controls.

Conclusions. In this study, we analyzed for the first time the microbiome composition of fecal and salivary samples obtained from patients with LS and their relative age- and sex-matched controls. The results obtained so far show microbiome variations in fecal and salivary samples already associated with inflammatory and pathological conditions in other settings. This data suggest the possible role of pro-inflammatory bacteria in the development of tumors in a condition with a clear proneoplastic genetic background, such as LS.