CASE STUDY
NATIONAL INSTITUTES OF HEALTH

Empire Genomics Contributes to First Report of FGFR2 Amplification in Primary CRC Tumor

Background

Colorectal cancer (CRC) is the third most common malignancy diagnosed in both genders and a leading cause of morbidity and mortality. Amplification of FGFR2 is frequent in gastric and breast cancers, but has never before been reported in primary colorectal cancer. Some studies have indicated that certain FGFR tyrosine-kinase inhibitors (TKIs) have activity against the FGFR2-amplified colorectal cell line, NCI-H716. For this reason, it is clinically important to characterize colorectal tumors for FGFR2 amplification, as this could help identify patients who could benefit from FGFR TKI therapies.

Objectives

In this study, several analyses were performed in an attempt to describe and verify FGFR2 amplification identified by clinical next-generation sequencing (NGS) in a patient with primary colorectal cancer. The usefulness of somatic copy-number alterations (SCNA) prediction from NGS data was reported, as copy-number variations can have predictive value in and of themselves.

Approach

Genomic, histologic, and immunohistochemistry analyses of this clinical case of primary CRC with FGFR2 amplification were used to reveal clinically relevant information regarding these tumors, and how CRC patients may benefit from particular therapies. Next-generation sequencing (NGS), copy-number analysis, microarray, immunohistochemistry, and fluorescence in situ hybridization (using the BAC clone and CEP 10 from Empire Genomics) were all utilized as part of the experimental procedure.

Results

Results of extensive analyses, made possible in part by devices manufactured by Empire Genomics, make this the first report to identify FGFR2 amplification in a CRC tumor sample obtained directly from a patient's primary tumor and the first non-cell-line derived, clinical case to harbor this particular alteration. Phenotypic and genotypic similarities with the CRC cell line NCI-H716 were unveiled in this clinical case of primary CRC. Clinically relevant conclusions can be inferred, and the usefulness of clinical NGS panels in uncovering novel, actionable somatic mutations in cancer was demonstrated.

FGFR2 amplification in colorectal adenocarcinoma
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Lead Organization

National Institutes of Health

Diseases

- Colorectal Cancer

Biomarkers Mentioned

- FGFR2
- TP53

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