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CLINICALY SIGNIFICANT DNA ALTERATIONS IN UKRAINIAN COLON ADENOCARCINOMA SAMPLES: ADVANTAGES AND LIMITATIONS OF USING AN NGS HOTSPOT PANEL

Aim. The study aimed to identify the changes of clinically relevant gene variants in colon adenocarcinoma samples of Ukrainian patients using the NGS Ampliseq Hotspot Cancer Panel (HSCP) (ThermoFisher Scientific). **Methods.** The study used 98 samples of Ukrainian patients aged 38–87 years, who had colorectal cancer (adenocarcinomas) of various localization and degree of differentiation. To identify clinically relevant gene variants, the HSCP data was analyzed using the Franklin by GennoX database. **Results.** A total of 133 clinically relevant (Tier 1–3, Pathogenic, Likely pathogenic) gene variant alterations (SNVs, INDELs) were found in 31 of 50 genes. Tier 1–2 gene variants rate was more than 50% of all genetic variants. The largest number of different mutation types of Tier 1–2 were found in the APC, KRAS, PIK3CA, TP53, and NRAS (25, 12, 9, 7 and 6 accordingly). According to the Oncogenic classification it was detected 11 oncogenic variants of Tier 1–2, and 15 oncogenic variants of Tier 3 gene variants. Among the clinically significant variants according to VAF, more than 50% were identified as Somatic SNVs (VAF < 10%) and Likely Somatic SNVs (VAF 10–30%). The rate of Likely Germline SNVs and INDELs was approximately 40%. The therapeutic significance for colorectal cancer (A-D) was found for 20 genetic variants Tier 1–2 of the KRAS, NRAS, BRAF, PIK3CA. Meanwhile, the therapeutic significance for colorectal cancer at the gene level was also found for variants of the ACT1, APC, FBXW7, GNAS, and SMAD4. The strengths of HSCP are that it enables the detection of clinically significant somatic mutations in the genes listed in the Ukrainian colon cancer treatment protocol. In addition, it detects other oncogenic mutations that are not yet included in the protocols but have proven clinical significance. The presence of germline mutations must be confirmed using DNA of normal cells. **Conclusions.** A spectrum of clinically significant gene variants was identified in colon cancer samples, which are important for the diagnosis and treatment of patients. Further confirmation using a larger number of samples and the inclusion of new gene regions are necessary to analyze the hereditary impact of mutations and to conduct a pharmacological assessment of treatment response in colon cancer. **Grants/Fundings.** This work was supported by a grant from the National Research foundation of Ukraine [grant number 2021.01/0024] and IMBG Simons Foundation Grant for Ukrainian institutions № SFI-PD-Ukraine-00017453.

Keywords: NGS, clinically significant gene variants, colon adenocarcinoma, hotspot panel.

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