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# Mutation load in carotid paragangliomas

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*Motivation and Aim*: Carotid paragangliomas are tumors of head and neck that have neuroendocrine origin and arise at carotid bifurcation. High vascularity and location of the tumors make them a surgical challenge. Carotid paragangliomas are belonged to tumors with uncertain potential for malignancy; in 5–15 % of cases, they become to be malignant. Treatment options for patients with carotid paragangliomas (particularly with metastasis) are limited. The development the effective management strategy of these tumors based on early diagnosis and new therapeutic approaches is important. In this work, we performed the analysis of the mutation load (ML) in carotid paragangliomas. It was shown that in many tumors ML is associated with sensitive to immunotherapy, which is very promising for tumor treatment [1].

*Methods and Algorithms*: In the study, we used the collection of blood and tissues (tumors and lymph nodes) derived from 12 patients with carotid paragangliomas at Vishnevsky Institute of Surgery, Ministry of Health of the Russian Federation. The isolation of DNA from the samples was performed using FFPET DNA Isolation Kit (Roche, Switzerland). DNA from blood was isolated with MagNA Pure Compact Nucleic Acid Isolation Kit I (Roche) on MagNA Pure Compact Instrument (Roche). Exome library preparation was performed using TruSeq Exome Kit (Illumina, USA) according to the manufacturer's protocol. Sequencing of exome libraries was carried out on NextSeq 500 System (Illumina) with 2x75 bp paired-end reads at EIMB RAS "Genome" center (http://www.eimb.ru/rus/ckp/ccu_genome_c.php).

*Results*: The average ML was estimated for examined samples as a number of mutations per megabase of coding regions and was compared with the data obtained [3]. The values were approximately equal (~7/Mb).

*Conclusion*: Additional analysis of exome data from tumors with paired blood and lymph nodes tissues allowed accurately estimating ML in carotid paragangliomas.

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