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EFFECTIVE TREATMENT OF NEUROFIBROMATOSIS TYPE 1

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Relevance. Neurofibromatosis is a genetic autosomal dominant disorder mostly connected to begnin neurocutaneous neoplasm which is caused by the mutation of the suppression gene of neurofibromatosis type 1 (NF1) tumor gene which is located on chromosome 17 and it is the predominant type, it is found once in each 3500 newborns. If left untreated, the growth will lead to physiological deformities of the face and could transform in to malignant tumor. Despite the rarity of this disease, the issue of treating it, is relevant in modern medicine.

Aim: To study the clinical features and effectiveness of methods of diagnosis and treatment of type 1 neurofibromatosis in a patient, which will expand the understanding of the disease and improve approaches to its management.

Materials and methods. Medical histories in the 4th city children hospital with diagnosis neurofibromatosis type 1 where studied. In to special consideration was taken a patient who was recurrently admitted in 2024 to the department of maxillofacial surgery and with diagnosis neurofibromatosis type 1 who became the main object of observation for this research. In to consideration where taken all the MRI and CT and analyzed.

Results and their discussion. On the base of studying of the case histories it was brought to site that the patient was born with cafe-au-lait spots all over his body, but especially big in size was the spot along the zygomatic bone and partially on the frontal area, but after the age of 8 years old his mother started noticing deformities of this spot in the zygomatic area on the left side of the face in form of masses and growth of the soft tissue. Patient O, male, 19.01.2009. At 10 years old in 2019 they came to the 4TH children clinical hospital and were hospitalized to the maxillofacial department and a biopsy was performed 29.01.2020 the operation of incision biopsy with partially removal of the tumor was performed, he was diagnosed with neurofibromatosis type 1, histological picture: tissue fragment with light burgundy color of not specific form, with sizes 5x2x0,5см, 2x1x0,5см and 1x1x0,5см the histological conclusion showed neurofibromatosis 1st type. During surgery, the formation showed that it does not have clear borders, a capsule and bleeds. 10/27/2020 - the second surgical treatment was performed, with excision of an area of 4.7x4x1.2 cm, histological conclusion - diffuse neurofibroma. 03/11/2021 The child had 3 tumor fragments excised again - 1.5x0.8 cm, 4x2 cm and 4x2.5x1.0 cm, preserving the function of the facial nerve. On June, 09, 2022, surgical treatment was continued with further excision of part of the tumor and confirmation of the previously established diagnosis histologically. 02/15/2023 Surgical treatment was continued to remove part of the tumor, after which complete symmetry of the patient's face was obtained with complete preservation of the function of the facial nerve on the side of the tumor, but with a complication in the form of a deforming scar in the left buccal and temporal areas, which was surgically corrected on 03/26/2023. In 2024 the patient was discharged for outpatient treatment on a course of anti-scar therapy without a physiotherapeutic component.

Conclusions. The tumor does not have clear borders and capsule, which leads to significant difficulties during surgical treatment, especially when localized along the branches of the facial nerve. In addition, surgical treatment of a tumor often requires multiple staged interventions and long-term observation due to the possibility of recurrence and growth, as well as malignancy. Minimally invasive surgical treatment allows to achieve not only an acceptable, but often good aesthetic result.