

Molecular genetic characteristics of salivary gland tumors in differential diagnosis and prognosis prediction

Abstract:

In the presented manuscript the author summarizes the current knowledge on molecular biomarkers of salivary gland cancer, focusing on tumor-type specific fusion oncogenes and their use as diagnostic, prognostic and therapeutic biomarkers. In detail, the author deals with adenoid cystic carcinoma (AdCC), the second most common salivary gland cancer. New facts of its biological behavior as well as new fusion oncogenes probably responsible for its carcinogenesis were described in the last few years.

A retrospective case series evaluating 27 patients suffering from AdCC, who were treated at the University Hospital in Pilsen in the last 30 years (1986-2016), is presented in this study. The following characteristics were observed: age, gender, tumor location, clinical stage at diagnosis, presence of regional and distant metastases, tumor grade, duration of follow-up, treatment and outcomes. Detection of the 1p36 deletion and the t(6;9)(q22–23;p23–24) chromosomal translocation resulting in the *MYB–NFIB* gene fusion were performed.

The incidence of AdCC in minor salivary glands, submandibular gland, parotid gland and sublingual gland was 41 %, 26 %, 22 % and 11% respectively. The following staging was observed: the 1st stage in 26 %, the 2nd stage in 18 %, the 3rd stage in 26 % and the 4th stage in 30 % of cases. Metastases to regional lymph nodes were diagnosed in 26 % and distant metastases in 30 % of patients (55 % to lung, 27 % to liver, 9 % to bones and 9 % of peritoneal metastases). The average follow-up was 76.4 ± 67.0 months (range 7-287 months). An outcome of the treatment during follow-up was as follows: 59 % of patients were with no evidence of the disease, 22 % of patients died because of the disease and 19 % of patients were alive with a recurrence or metastases of AdCC. The *MYB-NFIB* fusion transcript was detected in 79% of cases (19/24) and the 1p36 deletion in 13% of cases (3/23).

In line with a recent literature the high incidence of regional lymph node metastases as well as the *MYB-NFIB* fusion oncogene was confirmed in our retrospective case series of 27 patients with AdCC. The *MYB-NFIB* gene fusion could currently only be used as a potential diagnostic tool in difficult histopathological cases of AdCC, especially in late distant metastases.

Keywords:

Salivary gland cancer, fusion oncogene, biomarker, targeted therapy, adenoid cystic carcinoma, *MYB-NFIB*