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GORLIN-GORLZ SYNDROME

Gorlin-Gorlz Syndrome, also known as nevoid basal cell carcinoma (NBCCS) is a rare autosomal dominant transmitted disease characterized by multiple skin impairment that have a high degree of malignancy. The estimated prevalence varies from 1/57.000 to 1/ 256.000, with a male-to-female ratio of 1:1. The most common phenotypic expressions of this syndrome are: multiple basal cell carcinoma (skin tumor derived from basal cells of epidermis and its annexes), multiple skin lesions on the head, neck and trunk simultaneously occurring especially in areas less exposed to solar radiation. It is frequently associated with bone cysts, skeletal abnormalities, ectopic intracranial calcifications and palmar and plantar hyperkeratosis, facial dimorphism (macrocephaly, cheilopalatoschizis), cleft eye abnormalities, cardiovascular and genitourinary anomalies. NBCCS is caused by mutation in the PTCH 1 gene and is transmitted as an autosomal dominant trait with complete penetrance and variable expressivity and therefore mutation gene analysis may confirm the diagnosis. Otherwise the diagnosis is established based on the major criteria or one major criteria and two minor criteria. Management of this disease is interdisciplinary (dermatologist, surgeon and neurologist) because life expectancy is not altered and morbidity not grow substantially. Treatment varies with gravity, extension and associated diseases from surgical excision (when basal cell carcinomas are limited), to laser ablation, PUV-A (UV-A photodynamic therapy) and topical treatment. In this paper we present a series of cases of Gorlin-Gorlz syndrome seen in dermatology department, that illustrate both the primary syndrome manifestations and their complications. So once again we highlight the importance of accurate and early diagnosis of Gorlin syndrome.

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ENHANCED SUBCUTANEOUS B16 MELANOMA CELL-DERIVED PRIMARY TUMOR GROWTH WITH REDUCED INTRATUMORAL ARGINASE ACTIVITY IN STAT6-DEFICIENT MICE

Introduction: Tumor tissue contains different immune cells which can generate an anti-tumor immune response. Paradoxically, developing tumors can often induce a pro-tumoral immune-microenvironment leading to tumor progression and metastasis. The STAT6 transcription factor is a central regulator of gene expression mediated by IL-4 and IL-13 during the differentiation of Th2-type T cells and M2 macrophages. Th2 type cytokines can be produced by tumor-infiltrating immune cells and, in some cases, also by tumor cells during tumor development. Th2 cells and M2 macrophages are known to inhibit anti-tumor immunity and promote tumor progression.

Aim: The main goal of our work was to determine the role of STAT6 transcription factor in primary tumor development using subcutaneous B16 mouse melanoma tumor model in STAT6 knock-out and wild type mice.

Methods: Subcutaneous tumors were removed 14 days after inoculation and individual tumor weights were measured. We also examined the basic structure and macrophage content of tumors using immunohistochemistry as well as the tumors' immunological status by the measurement of different immune cell markers at RNA level. We also measured the arginase activity of primary tumor lysates.

Results: Our results show a significantly increased subcutaneous tumor growth in STAT6-deficient mice compared to the wild type animals. The histological structure of the tumor tissues were similar in both groups, but peripheral tumor cell infiltration into the neighboring tissues was enhanced in STAT6 KO mice. Gene expression analysis revealed a significant down-regulation of M2 macrophage marker arginase 1. Furthermore, in accordance with the gene expression levels, arginase activity of whole tumor lysates was also reduced in case of STAT6-deficient mice-derived tumors.

Conclusion: These results raise the question about the possible anti-tumor role of STAT6-induced arginase 1 expression/activity in the regulation of primary tumor growth.