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Case Report

A Case Report on Immunosuppressive Drugs and Neurofibromatosis Type 1

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Abstract

In this report, we aim to highlight the favoring effect of immunosupressants on the progression of NF1 lesions in a patient with café au lait maculae treated with immunosupressants for corticosteroid dependent nephrotic syndrome. The patient's disease progressed after two years of treatment and regressed upon stopping immunosuppressive treatment. Neurofibromatosis type 1 is one of the most common genetic diseases, which can be complicated during its course by the appearance of serious complications, in particular tumors of the central nervous system.

***Keywords**: neurofibromatosis type 1(NF1), immune system, immunosupressants, central nervous system tumors , optic nerve glioma*

Abbreviations:

NF1: neurofibromatosis type 1; MRI: Magnetic Resonance Imaging; MMF: Mycophenolate Mofetil

Introduction

Vonreckling or NF1 One of the most prevalent genetic disorders with autosomal dominant transmission is Hausen's disease.

Although it can affect multiple organs, the hallmark symptoms include early-life onset of café au lait maculae and tumors of the peripheral and central nervous systems. The phenotypic of this condition is highly variable, even within families, with both severe and rough versions coexisting.

Case Report

The patient, a 15-year-old boy with no family history of NF1, had been treated with corticosteroids for corticosteroid-dependent nephrotic syndrome for six years. Because of the patient's corticodependence, an immunosuppressant (MMF) was started at age ten.

Clinically, the examination was distinguished by the presence of cafe au lait maculae at the trunk and limbs with varying diameters.

The patient first appeared at age 12 with pyramidal syndrome, which is characterized by lower limb predominance ataxia, weakness, hyperflexia, spasticity, and Babinsky sign.

After a cerebromedullar MRI, two lesions were detected: a right optic nerve glioma with a diameter of 4.7 mm and a bulkiest 8 mm by 7 mm left thalamus lesion. Rathke's pouch cyst and several intra parenchymal hamartomas were mostly found on the left.

In the literature, the Journal of Hurt and Lung Transplantation, volume 23, issue 6, June 2004, documented two observations of disease progression in patients following lung transplantation.

Since the MMF was discontinued, the patient is only receiving corticosteroids for treatment.

An MRI was performed three months after the MMF was stopped, showing the removal of parenchymal lesions, a distinct 9 mm left thalamic lesion, and a persistent 4,5 mm diameter right optic nerve glioma.

An MRI three months later showed diffuse enlargement of the right optic nerve at a thickness of 5.3 mm, but no abnormalities at the brain level.

Multiple cutaneous neurofibromas occur in the trunk during the course of the corticosteroid treatment.

Discussion:

Dominant mutations in the neurofibromine-encoding gene are associated with NF1.

The gene NF1 suppresses tumors.

Leukemias, pheochromocytomas, malignant nerve sheath tumors, gliomas, and neurofibromas of NF1 patients have all been shown to have the mutation; yet, NF1 has other functions that account for cognitive impairments, vasculopathy, and other symptoms of NF1.

It is becoming increasingly evident that immune system function and immune cell promotion of tumor genesis in NF1 are significantly impacted by haploinsufficiency for NF1 mutation.

RAS signaling is downregulated by neurofibromin, and NF1 patients have overactive RAS phenotypes in a variety of cell types.

The role of the RAS pathway is to transfer signals from extracellular milieus to the nucleus of cells, where particular genes are triggered for the growth, division, and differentiation of cells. NF1 mutation results in alterations in the amounts of cytokines, T cells, and B cells. indicating alterations in immune system activity.

Clarification of the causal connection between immune cell changes and cancer has been aided by mice models of NF1 related malignancies.

Studies have indicated that immunosuppressive medications (trial recipients) raise the risk of several malignancies.

Disease progression when using immunosupressants may be explained by the correlation between immunosupression and an increased risk of tumorigenesis in NF1.

Additional findings imply that steroids may accelerate the formation of neurofibromas.

Conclusion

NF1 is associated with an increased risk of cancer, particularly tumors of the peripheral and central nervous systems. While more research is necessary to fully understand the role of NF1 in this association, patients with NF1 must be closely monitored when receiving immunosuppressant medication.

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