Evangelos Axiotis

Deciphering the Mechanism of Immune Dysfunction in Vici Syndrome

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SAPIENZA
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Faculty of Pharmacy and Medicine

Department of Molecular Medicine

PhD in Immunological and Hematological Sciences and Rheumatology

Curricula: Clinical Immunology

Deciphering the mechanism of immune dysfunction in Vici syndrome

Ph.D. candidate

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“What we observe is not nature itself, but nature exposed to our method of questioning”

Werner Heisenberg

I dedicate my Ph.D. thesis to the children with Vici syndrome and their families.

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Chapter 1

Introduction

1.1) Vici Syndrome

Vici syndrome (OMIM # 242840) is a rare genetic multisystem disorder characterized by five main clinical signs: 1) Agenesis of the corpus callosum (ACC) (total or partial lack of the truncus or "body" of the corpus callosum, connecting the right and left hemisphere of the brain); 2) Cataract (progressive clouding of the lens inside the eye which lead to a decrease in vision); 3) cardiomyopathy(cardiac changes pronounced in the left compared to the right ventricle); 4) Immunodeficiency (functional deficiency of the immune mechanisms); 5) hypopigmentation (variable from total albinism in a partial pigmentation of the retina). (Figure 1)

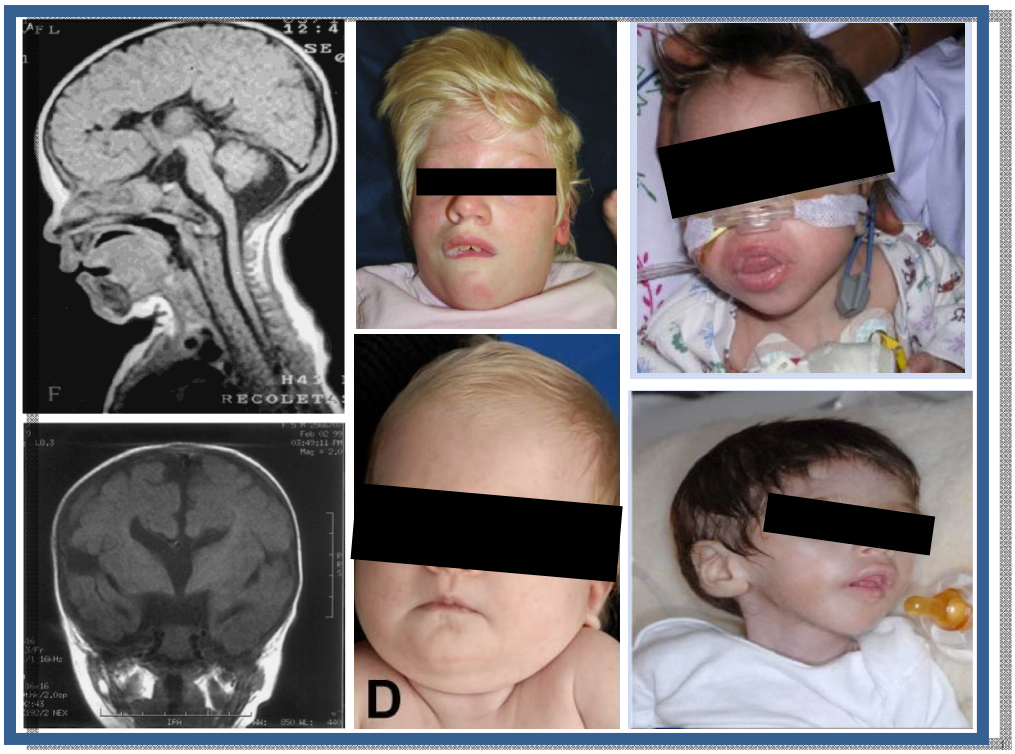

Figure 1. Clinical Features of patients with Vici Syndrome. The magnetic resonance brain imaging showed agenesis of the corpus callosum, while examination of the ocular fundus shows a strongly hypopigmental retina. Patients may present dysmorphic facial features, such as microcephaly, hypertelorism, micrognathia.